Medical eponyms

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Abernethy malformation congenital anomaly of the splanchnic vasculature, arises from defects in vitelline vein formation; in type 1, portal vein completely diverted into IVC and complete absence of formation of intrahepatic portal vein; in type 2, portal venous system is formed, but there is an abnormal communication with systemic veins, usually IVC; associated with hepatic encephalopathy and hepatopulmonary syndrome; more common in dogs, particularly Yorkshire terriers

Adamkiewicz, artery of artery responsible for anterior spinal syndrome; can be severed in AAA repair

Adamson's fringe in tinea capitis, the location of the terminal tuft of hyphae; weakest point of hair located just about Adamson's fringe

Addison's disease primary adrenal insufficiency, bilateral adrenal destruction by TB used to be most common cause, now only accounts for 7-20% of cases; now autoimmune disease 70-90%, remainder caused by infectious disease, metastasis or lymphoma, adrenal hemorrhage, infarction, or drugs

Adie's pupil tonic pupil, larger than contralateral unaffected pupil, reacts sluggishly to changes in illumination, see also Holmes-Adie syndrome; seen in young women; no neurologic significance

Adler sign for distinguishing appendicitis from adnexal or uterine pain, if the point of maximal tenderness shifts medially with repositioning on the left lateral side, etiology generally gynecologic

Adson's sign in thoracic outlet syndrome, decrease in ipsilateral radial pulse and/or presence of subclavian bruit while patient extending neck maximally, rotating head towards side being tested, and holding breath

Ahumada-del Castillo syndrome galactorrhea-amenorrhea not associated with pregnancy

Aicardi syndrome partial or complete agenesis of corpus callosum, infantile spasms, mental retardation, lacunae of the retina; occurs only in females; likely X-linked dominant with lethality in males; described in 1965

ainhum disease autoamputation of a digit, usually of the fifth toe from a constricting scar in the form of a band or groove; usually from trauma acquired from walking barefoot; more common in tropics in patients of African descent; "ainhum" means "fissure" in one of the indigenous language in Brazil

Alagille syndrome inherited cholestatic syndrome, associated with biliary hypoplasia (ductopenia), vertebral anomalies, prominent forehead, deep-set eyes, peripheral pulmonic stenosis

Albers-Schönberg disease osteopetrosis or marble bone disease

Albert's test for diagnosing hemineglect; patient presented with multiple lines on a page and asked to cross them out; if >70% of crossed out lines are ipsilateral to motor deficit, hemineglect is diagnosed; described in *Neurology* 1973; 23:658-64

Albright's hereditary osteodystrophy polyostotic fibrous dysplasia, short stature, round face, skeletal anomalies (brachydactyly, also see Archibald's sign), and heterotopic calcification, precocious puberty, café-au-lait spots on skin, low calcium, high phosphate, resistance to elevated PTH levels from mutation in $G_s$alpha coupling PTH receptor to adenylyl cyclase

Alder-Reilly anomaly large, dark, pink-purple granules in cytoplasm of neutrophils; autosomal recessive trait resulting in abnormal granule development in neutrophils resembling severe toxic granulation

Alexander's disease leukodystrophy-like neurodegenerative disease presenting in infancy or childhood; characterized by Rosenthal fibers

Alexander's law peripheral vestibular nystagmus increases in intensity when the gaze is in the direction of the fast phase, and decreases in intensity when the gaze is away from the fast phase

Allen's sign in pulmonary embolism, fever, tachycardia, and tachypnea, present in only 23% of cases

Allen's test for demonstrating patent ulnar artery and an intact superficial palmar arch, patient's hand is initially held high while fist is clenched and both radial and ulnar arteries are compressed, hand then lowered and fist is opened; after ulnar pressure released, color should return to the hand within 6 seconds

Allis sign in congenital hip dislocation, difference in knee height when child is supine with knees flexed and feet are flat on examination table

Alport's syndrome hereditary nephritis accompanied by deafness and various eye disorders, including lens dislocation, posterior cataracts, and corneal dystrophy; a few families have large platelets (20-25 fL), thrombocytopenia, and leukocyte inclusions which resemble May-Hegglin anomaly

Alström's syndrome obesity, autosomal recessive, childhood blindness due to retinal degeneration, nerve deafness, vasopressin-resistant diabetes insipidus, and in males, hypogonadism with high plasma gonadotropin levels (end organ resistance to hormones), also baldness, hyperuricemia, hypertriglyceridemia, aminoaciduria

Alzheimer's disease most frequent cause of dementia, pathologically characterized by neurofibrillary tangles, neuritic plaques, and granulovacuolar degeneration, degeneration of nucleus basalis of Meynert (principal origin of cholinergic innervation); described in 1906

Amadori product products of early non enzymatic glycosylation of proteins

Andermann syndrome familial progressive sensorimotor neuropathy with agenesis of the corpus callosum, mental retardation; found in high frequency in

	Quebec; associated with mutation in SLC12A6, a K-Cl transporter
Andersen disease	type IV glycogen storage disease (or amylopectinosis), from branching enzyme deficiency, presents with cirrhosis with hepatosplenomegaly and failure to thrive in the first 18 months of life
Andersen's syndrome	periodic muscle paralysis, arrhythmias, short stature, macrocephaly, hypertelorism, brachydactyly, tapering fingers; from mutation in *KCNJ2* which encodes a potassium channel Kir2.1; also with dental abnormalities, incomplete secondary dentition, cleft palate, first described in 1971; also long QT syndrome 7 (LQT7)
Angel's sign	performing an otoscopic exam while patient is blowing against a pinched nose; increases sensitivity of otoscopy for the detection of a perforated tympanic membrane by demonstrating fluid or pus in the external canal; described in 1994
Angelman syndrome	"happy puppet" syndrome characterized by severe developmental delay, frequent laughing, easily excitable personality; from maternal deletion of 15q11-13; associated with mutation in maternally-imprinted ATP10C, a putative aminophospholipid translocase
Anitschkow myocytes	in rheumatic fever, large mesenchymal cells in myocardial lesion
Antley-Bixler syndrome	trapezoidocephaly-synostosis syndrome; characterized by midface hypoplasia, humeroradial synostosis, bowing of femora, fractures; associated with inactivating mutations in FGFR2; occasionally associated with ambiguous genitalia
Anton's syndrome	denial of cortical blindness; a form of anosognosia
Apert syndrome	autosomal dominant disorder from mutation in FGFR2 characterized by craniosynostosis, mid-facial malformations, symmetric bony syndactyly of hands and feet, and varying degrees of mental retardation, from missense mutations in the fibroblast growth-factor-receptor 2 (FGFR2) genes
Apgar score	developed in 1952, five parameters assessed: heart rate, respiratory effort, muscle tone, reflex irritability, and color; score 7 or greater said to be indicator of good health; 5 minute score most reliable; 5 min score of 0-3, mortality 244/1000 v. score of 7-10, mortality 0.2/1000 (*N Engl J Med* 2001;344:467)
Apley grind test	for meniscal tears, flex patient's leg to 90 degrees, then grind the tibial condyles against the femoral condyles with rotation motion in the varus and then valgus positions
Apley scratch test	asking patient to scratch the back from above or bottom, looking for furthest point reached (T4-T5 former, T7-T8 latter normal); if can't reach, infraspinatus/teres minor tendonitis tear or subscapularis tendonitis/tear respectively
Apley's law	in pediatrics, the farther a chronically recurrent abdominal pain is from the umbilicus, the greater the likelihood of an organic cause for the pain
Apt test	test which differentiates fetal from maternal hemoglobin in infant's gastric contents, vomitus, or stool
Archibald's sign	in pseudohypoparathyroidism of Albright's hereditary osteodystrophy, characteristic shortening of the fourth and fifth digits as dimpling over the knuckles of a clenched fist
Argyll Robertson pupils	small irregular pupils, usually but not always caused by CNS syphilis, they accommodate, but don't react; absence of miotic reaction to light, both direct and consensual, with preservation of a miotic reaction to near stimulus; lesion in tectum of midbrain; named after Douglas M. C. L. Argyll Robertson.
Arnold's nerve	auricular branch of vagus nerve supplying posterior and inferior meatal skin of ear; stimulation can elicit cough reflex
Arnold's reflex	ear cough reflex mediated by Arnold's nerve
Arnold-Chiari malformation	downward displacement of the cerebellar tonsils and medulla through the foramen magnum, results in pressure atrophy of displaced brain tissue, hydrocephalus from obstruction of the CSF outflow tract, is almost always characterized by the presence of a thoracolumbar meningomyelocele
Arthus reaction	localized area of tissue necrosis resulting from acute immune complex vasculitis; type III hypersensitivity
Aschoff body	pathognomonic finding in rheumatic carditis, an area of local fibrinoid necrosis surrounded by inflammatory cells including lymphocytes, plasma cells, and macrophages that later resolve to fibrous scar tissue
Asherman's syndrome	intrauterine synechiae, typically occurs after curettage of the uterus, presenting as amenorrhea
Asherson's syndrome	catastrophic antiphospholipid antibody syndrome
Ashman's phenomenon	relationship of aberrancy to changes in the preceding cycle length; may persist for several cycles, usually exhibits RBBB morphology
Askin's tumor	malignant small-cell tumor of the thoracopulmonary region; member of Ewing sarcoma family or primitive neuroectodermal tumors
ASPEN syndrome	Association of Sickle cell disease, Priapism (usually major), Exchange transfusion, Neurological events including headache, lethargy, and seizures; may be due to abrupt cerebral hypoxemia from too-rapid exchange or release of vasoactive cytokines released from penile blood vessels; described in *J Urol.* 1993; 150:1480-1482.
Asperger's disorder	severe and sustained impairment in social interactions and the development of restricted, repetitive patterns of behavior, interests, and activities, but intellectually normal and no language delays, but with abnormalities of spoken language
Auenbrugger's sign	epigastric bulge due to a massive pericardial effusion
Auer rods	present in acute myelogenous leukemia or refractory anemia with excess blasts; granules form elongated needles, granules are all azurophilic, contain peroxidase; fused lysosomes

Auerbach's plexus	myenteric plexus, between the longitudinal and circular layers of muscle; provides motor innervation to the two muscle layers and secretomotor innervation to the mucosa
Auspitz's sign	for psoriasis; sign is positive when slight scratching or curetting of a scaly lesion reveals punctate bleeding points within the lesion; suggests psoriasis, but is not specific
Austin Flint murmur	diastolic rumble in aortic regurgitation heard at cardiac apex, thought to be due to aortic jet impinging on the mitral valve, causing vibrations and also from simultaneous diastolic filling of the left ventricle from the left atrium and aorta closing the mitral valve in diastole, producing physiologic stenosis; named after Austin Flint (1812-1886)
Australia antigen	HBsAg, found in the serum of an Australian aborigine
Austrian triad	clinical triad of pneumococcal pneumonia, meningitis, and endocarditis (classically aortic valve endocarditis associated with aortic regurgitation); described by Robert Austrian
Babinski sign	upper motor sign that indicates dysfunction of fibers within the pyramidal system; described in 1896 by Babinski, student of Charcot
Bachmann's bundle	anterior internodal tract in atrial conduction system
Bainbridge reflex	compensatory increase in heart rate caused by a rise in right atrial pressure
Baker's cyst	popliteal cyst, a synovial cyst within the popliteal fossa
Balint's syndrome	optic ataxia (inability to visually guide limb movements), ocular ataxia (inability to direct eyes to a precise point in the visual field), inability to enumerate objects in a picture or extract meaning from a picture, and inability to avoid objects in one's path, simultanagnosia, from infarction in unilateral or bilateral visual association due to watershed stroke between distal PCA and MCA
Balkan nephropathy	degenerative interstitial nephropathy seen in Balkan areas (tributaries of Danube River), see tubular proteinuria, glycosuria, RTA, azotemia, associated with increased risk of upper tract transitional cell carcinoma
Ball's disease	intracerebral leukocytostasis, potentially fatal complication of acute leukemia (especially AML) when peripheral blast cell count >100,000/uL; leukemic cells capable of invading through endothelium and causing hemorrhage into brain; not generally seen with CLL or CML
Ballance's sign	tender mass in the left upper quadrant due to a spleen hematoma
Baló's disease	variant of multiple sclerosis, see concentric rings of demyelination separated by bands of preserved myelin
Baltic myoclonus	see Unverricht-Lundborg disease
bamboo spine	seen in ankylosing spondylitis
Bancroft's sign	in deep vein thrombosis, compression of calf forward against tibia causing more pain than horizontal compression when gastrocnemius muscle is lifted; also referred to as Moses's sign
Bang's disease	brucellosis
Bannayan-Ruvalcaba-Riley syndrome	see Bannayan-Zonana syndrome
Bannayan-Zonana syndrome	hamartoma syndrome characterized by macrocephaly, multiple lipomas, and hemangiomas, speckled penis; associated with with germ line *PTEN* mutations; also known as Bannayan-Ruvalcaba-Riley syndrome
Bannwarth's syndrome	in early disseminated Lyme disease, triad of lymphocytic meningitis, cranial nerve palsies (especially VII nerve which may be bilateral), and radiculoneuritis
Banti's syndrome	splenomegaly, hypersplenism, and portal hypertension, noncirrhotic, arises after subclinical occlusion of the portal vein, usually years after occlusive event
Bantu siderosis	unusual form of iron overloading resembling hereditary hemochromatosis in South African blacks ingesting large quantities of alcoholic beverages fermented in iron utensils
Bárány test	see Dix-Hallpike test
Bardet-Biedl syndrome	mental retardation, pigmentary retinopathy, polydactyly, obesity, and hypogenitalism; genetically heterogeneous disorder with linkage to 7 loci; has been incorrectly called Laurence-Moon-Bardet-Biedl syndrome in the past
Barlow's disease	mitral valve prolapse
Barlow's maneuver	for congenital hip dislocation, patient placed in supine position and attempt made to push femurs posteriorly with knees at 90 degrees/hip flexed and hip will dislocate
Barr body	condensed, inactive X-chromosome in females; dense, stainable structure
Barraquer-Simons syndrome	acquired partial lipodystrophy; presents usually around 8-10, preceded generally by an acute viral infection; spares legs and hips; 1/3 of patients develop membranoproliferative glomerulonephritis; associated with accelerated complement activation and a serum immunoglobulin G, called C3 nephritic factor that are felt to cause lysis of adipose tissue
Barré-Liéou syndrome	cervicosympathetic syndrome, characterized by neck pain and dizziness due to arthritic or traumatic damage to the cervical spine
Barrett's esophagus	esophageal strictures and epithelial metaplasia from squamous epithelium to a specialized columnar epithelium with intestinal metaplasia in 10% of severe GERD
Barth's syndrome	infantile X-linked dilated cardiomyopathy, short stature, myopathy, cyclic neutropenia; from mutation in G4.5 which encodes for

	tafazzin (a putative acyl transferase that has been associated with altered metabolism of the mitochondrial phospholipid cardiolipin)
Bartholin gland	paired glands located near vaginal opening; maintain moisture of vaginal vestibular surfaces; can form abscesses or cysts; homologous to Cowper's gland; described by Danish anatomist Caspar Bartholin in 1677
Barton's fracture	intra-articular fracture of dorsal margin of distal radius; extends into radio-carpal joint
Bartter's syndrome	hypokalemic, hypochloremic metabolic alkalosis with normal or low blood pressure despite increased renin and aldosterone levels and hyperplasia of juxtaglomerular apparatus due to mutations affecting diuretic-sensitive sodium-transport proteins
Bassen-Kornzweig syndrome	congenital abetalipoproteinemia
bat wing edema	pulmonary edema in perihilar distribution in approximately 5% of cases
Bateman's senile purpura	purpura following trauma to severely sun-damaged skin of the dorsal forearm of elderly persons; months may be required for resolution of pigmentation from hemorrhage
Batista procedure	for treatment of heart failure, removal of portion of left ventricular free wall, i.e. partial left ventriculectomy
Batson's plexus	portal vertebral venous communications, may be responsible for isolated bone metastases in sacrum or vertebral bodies from colorectal cancer
Batten's disease	later-onset ceroid lipofuscinosis, CLN3, also used to describe juvenile form specifically; in general a group of conditions characterized by mental impairment, worsening seizures, and progressive loss of sight and motor skills related to buildup of lipopigments
Battle's sign	ecchymoses over the mastoid process in basilar skull fractures, generally occurring approximately 48 hours after event
Bazex's syndrome	acrokeratosis paraneoplastica, consists of erythematous to violaceous psoriaform plaques occurring predominantly in acral areas; associated with Hodgkin's disease and squamous cell carcinomas of digestive tract
Bazin's disease	nodular vasculitis or erythema induratum, a form of panniculitis classically associated with tuberculosis characterized histologically by caseation necrosis; described by Bazin in 1861
BCG	Bacille bilié de Calmette-Guérin; Leon A. Calmette, French bacteriologist, 1863-1933; Camille Guérin, French bacteriologist, 1872-1961; attenuated strain of *Mycobacterium bovis* bacille Calmette-Guérin
Beau's lines	horizontal depressions across nail plate seen as nail grows out, caused by a transient arrest in nail growth, can occur during acute stress (e.g., high fever, circulatory shock, myocardial infarction, pulmonary embolism)
Bechterew's disease	ankylosing spondylitis
Beck's triad	in pericardial tamponade, distended neck veins, distant heart sounds, hypotension, i.e. rising venous pressure, falling arterial pressure, and decreased heart sounds, Claude S. Beck thoracic surgeon 1935
Becker's muscular dystrophy	X-linked, normal levels of dystrophin but function altered, average onset 11 y.o., age at death 42 y.o., CK elevated
Becker's sign	in aortic regurgitation, visible pulsations of the retinal arterioles
Beckwith-Wiedemann syndrome	exomphalos, macroglossia, gigantism; associated with neonatal hypoglycemia
Beevor's sign	lesions of T9-T10 paralyze lower but spare upper abdominal muscles, resulting in upward movement of umbilicus when abdominal wall contracts
Behçet's disease	triad of aphthous ulcers, genital ulcerations, and ocular inflammation (posterior uveitis). Associated with erythema nodosum, cutaneous pustular vasculitis, also synovitis, CNS vasculitis involving brain stem, thrombophlebitis, and positive pathergy response. Described by Turkish dermatologist Behçet in 1937; also known as Silk Road disease due to clustering of cases along the Silk Road.
Bell's palsy	peripheral seventh nerve palsy; seen as a complication in diabetes, tumors, sarcoidosis, HIV, and Lyme disease
Bell's phenomenon	physiological upward rotation of the eyeball triggered by contraction of the ipsilateral orbicularis muscle with resulting closure of the eyelid
Bence Jones proteins	free Ig light chains seen in plasma cell dyscrasias (e.g. multiple myeloma, AL-amyloidosis, light-chain deposition disease, Waldenström's macroglobulinemia, MGUS, heavy-chain disease (mu) (rare), lymphoproliferative disease (rare), rifampin therapy (rare), filtered by glomerulus and then reabsorbed tubular cells; proteins are toxic to tubule cells; described by Henry Bence Jones
Benedikt syndrome	clinical picture from paramedian midbrain infarction from occlusion of the paramedian penetrating branches of the basilar artery affecting the third nerve root fiber, red nucleus, cerebral peduncle resulting in ipsilateral medial rectus palsy with a fixed dilated pupil and contralateral tremor, chorea, and athetosis
Bennett's fracture	fracture of the base of the first metacarpal with involvement of carpometacarpal joint
Bentall procedure	for treating ascending aortic aneurysms, composite prosthetic graft consisting of prosthetic aortic valve sewn onto end of graft
Berardinelli-Seip syndrome	congenital generalized lipodystrophy, apparent at birth, infants look very muscular due to absence of fat, associated with diabetes, hepatomegaly, acanthosis nigricans, enlarged external genitalia, and increased rate of skeletal growth.
Berger's disease	IgA nephropathy, ESRD develops in 15% of cases at 10 years and 20% at 20 years, treated with steroids in certain instances
Bergman minimal model	determinants of glucose disposal: phi-1 (acute insulin secretion), phi-2 (sustained insulin secretion), Si (insulin sensitivity), Sg (glucose sensitivity)

Bergman's triad	seen with fat emboli syndrome: 1. mental status changes; 2. petechiae (often in the axilla/thorax); 3. dyspnea
Bergmann gliosis	in ethanol abuse, proliferation of astrocytes adjacent to lost Purkinje cells between depleted granular cell and molecular layer of cerebellum
Bernard-Soulier disease	inherited disorder of platelet function where there is absence of Gp Ib/IX, the von Willebrand receptor, characterized by giant platelets and greater than expected bleeding for the degree of thrombocytopenia
Bernheim effect	in aortic stenosis, right ventricular failure preceding left ventricular failure from hypertrophied ventricular septum bulging into and encroaching on right ventricular filling
Bernheim effect, reverse	in pulmonary embolism, right ventricular failure causing septum to bulge into and compromise left ventricular filling
Bernstein test	to test for GERD, acid perfusion test of esophagus with 0.1 N HCl and see if reproduces chest pain; limited sensitivity and specificity though
Berry's ligament	thickened fasica next to the trachea; binds thyroid gland to cricoid cartilage; recurrent laryngeal nerve tends to run underneath it
Berry's sign	in malignant thyromegaly, absence of carotid pulsation from tumor encasing carotid and muffling pulsations
Bertin, renal columns of	the spaces between adjacent pyramids where cortical tissue extends into
Best disease	vitelliform macular dystrophy type 2 (VMD2), characterized by gradual loss of visual acuity starting in their teenage years; accumulation of lipofuscin in retinal pigment epithelium; markedly abnormal electro-oculogram (EOG) in all stages of progression and in phenotypically normal carriers; autosomal dominant, from mutation in VMD2 gene; first described in 1905
Betz cells	large pyramidal cells in layer 5 of primary motor cortex largest neurons in mammalian CNS; 30-40,000 Betz cells in precentral gyrus in one side of the brain
Bezold's abscess	abscess of mastoid tip
Bezold-Jarisch reflex	activation of receptors in the atria, great veins, and left ventricle causing increased parasympathetic tone and decreased sympathetic activity leading to a combination of hypotension and bradycardia with a sudden increase in coronary flow
Bickers-Adams syndrome	Sex-linked hydrocephalus, aqueductal stenosis, mental deficiency, and flexion deformities and spasticity of the extremities. X-linked (Xq28)
Bickerstaff's encephalitis	brain stem encephalitis
Bielschowsky's tilt test	in trochlear nerve palsy (which paralyzes the superior oblique muscle), elevation in the affected eye is greatest when the head is tilted toward the side of the involved eye and abolished by tilt in the opposite direction
Bier block	regional anesthesia of an extremity by placing a tourniquet and then infusing local anesthetic into a vein
Biette's collarette	in syphilis, a thin white ring of scales on the surface of a lesion
Billroth I	antrectomy with gastroduodenostomy
Billroth II	antrectomy with gastrojejunostomy
Billroth's cords	splenic cords found in the red pulp between the sinusoids, consisting mainly of fibrils and connective tissue cells
Bing's sign	extensor plantar response by pricking the dorsal surface of the big toe with a pin suggesting upper motor neuron defect
Bing-Horton syndrome	erythroprosopalgia, attacks of facial pain associated with marked reddening of the ipsilateral half of the face associated with tearing and watery discharge from the nose, occurs during sleep and is of brief duration, believed to be due to irritaiton in greater petrosal nerve
Binswanger's disease	subcortical arteriosclerotic encephalopathy, associated with hypertension; characterized by multiple lacunar infarcts and progressive demyelination limited to the subcortical area with characteristic sparing of cortex
Biot's breathing	succession of hyperpnea/hyperventilations and apneas (seen in increased ICP, drug-induced respiratory depression, brain damage, usually medullary level), but lacks typical crescendo-decrescendo pattern, abrupt beginning, and regularity of Cheyne-Stokes breathing (see also Cheyne-Stokes, cerebral level)
Birbeck's granules	aka Langerhans's granules; a small tennis racket-shaped membrane-bound granule with characteristic cross-striated internal ultrastructure seen in Langerhans cell histiocytosis
Birt-Hogg-Dube syndrome	genodermatosis characterized by hair follicle hamartomas, renal cell cancer, and spontaneous pneumothorax; caused by mutation in gene encoding folliculin; also associated with trichodiscomas and acrochordons; associated with mutation in *folliculin*
Bishop's score	scoring system for determining whether or not induction of labor will be successful, based on 5 criteria (scored from 0-3): position, cervical consistency, degree of effacement, dilatation, and station; score >9 suggests that induction will be successful
Bitot's spots	in vitamin A deficiency, small, circumscribed, lusterless, grayish white, foamy, greasy, triangular deposits on the bulbar conjunctiva adjacent to the cornea in the area of the palpebral fissure of both eyes
Bjork-Shiley valve	single tilting disk prosthetic valve, production stopped in 1986; large valves removed from market in October 1985 because of strut fracture

Blalock-Taussig shunt	for treating tetralogy of Fallot, direct end-to-side subclavian artery to pulmonary artery anastomosis on side opposite to arch of aorta (classic); alternatively graft placed between subclavian artery and pulmonary artery (modified)
Bland-White-Garland syndrome	anomalous origin of the left coronary artery from the pulmonary artery; presents in infancy with myocardial ischemia and CHF; though delayed presentations also occur including sudden death in adulthood
Blaschkow, lines of	patterning of a variety of linear nevi as well as linear patterning of commonly acquired diseases such as psoriasis, scleroderma, and lichen planus, including V shape over the upper spine, S shape of the abdomen, inverted U shape from the breast area, and perpendicular lines down lower extremities; cannot be explained by distribution of cutaneous nerve, lines of cleavage, nor blood vessels or lymphatics; first described in 1901
Blau's syndrome	granulomatous synovitis, non granulomatous uveitis, cranial neuropathies; resembles childhood sarcoidosis; no lung involvement; associated with mutations in *NOD2/CARD15* gene; first described in 1985
Blomstrand dysplasia	rare lethal disorder characterized by an increase in bone density and advanced skeletal maturationfrom inactivating mutation in PTHR-1 gene (see also Jansen metaphyseal chondrodysplasia where there is an activating mutation)
Bloom's syndrome	severe immunodeficiency, growth retardation, progeria, and predisposition to several types of cancers associated with hypersensitivity to a variety of DNA-damaging agents
Blount's disease	idiopathic varus bowing of tibia
Blumberg sign	rebound tenderness
Blumer shelf	carcinomatous metastasis from a primary site high up in the peritoneal cavity, may accumulate in the stomach, felt through the anterior rectal wall as a hard shelf in the rectovesical or rectouterine pouch (pouch of Douglas)
Boas's sign	right subscapular pain due to cholelithiasis, <7% sensitive
bobble-head syndrome	in children with progressive hydrocephalus, rapid, rhythmic bobbing of the head
Bochdalek's hernia	hernia through the posterior diaphragm, usually on the left, presents in infancy
Bockhart's impetigo	follicular impetigo
Boerhaave's syndrome	pressure rupture of the esophagus; can give rise to Hamman's sign
Bogota bag	temporary abdominal closure with filleted intravenous bag
Bogros space	space bounded by the peritoneum above and the fascia transversalis below where the lower part of the external ilica artery can be seen without cutting the peritoneum; called also retroinguinal space
Bohr effect	fall in pH leading to decrease in oxygen affinity of hemoglobin
Bohr equation	Vd/Vt, for determining ratio of physiologic dead space
Bombay phenotype	blood type O when A or B blood type expected; these individuals are recessive for H allele and do not make H antigen; H antigen required as precursor for A or B antigen to be expressed; first described in Bombay; explains a child with O blood type when a parent is e.g. AB blood type; rare phenotype, more common in consanguineous matings
Bonnet's sign	banking of veins distal to AV crossings (grade 3) in hypertensive retinopathy; compare with Salus's sign and Gunn's sign
Bonnevie-Ullrich syndrome	skeletal and soft tissue abnormalities (e.g., lymphedema of hands and feet, nail dystrophy, skin laxity), short stature, webbed neck.
Bordet-Gengou medium	for identifying *Bordetella pertussis*, medium contains high percentage of blood (20-30%) to inactivate inhibitors in blood; also has potato and glycerol
Bornholm disease	coxsackie virus producing pleurodynia, fever, cough, sore throat, myalgias in shoulder, chest, and abdomen; Bornholm is a Danish island in Baltic sea
Boston sign	in thyrotoxicosis, jerking of the lagging lid
Bouchard's nodes	bony spurs at PIP in osteoarthritis
Bourneville's disease	tuberous scelrosis
Bouveret syndrome	gastric outlet obstruction from gallstone impaction of the duodenum
Bowditch staircase	increased heart rate increases the strength of contraction in a stepwise fashion as the intracellular calcium increases over several beats
Bowen's disease	squamous cacrinoma in situ, seen generally on sun-exposed areas
Bowman's capsule	double-walled structure that surrounds the glomerulus
Boxer's fracture	fracture of the metacarpal neck, classically of small finger
Bradbury-Eggleston syndrome	pure autonomic dysfunction characterized by low circulating catecholamines

Brainerd diarrhea	acute onset of watery diarrhea lasting four weeks or longer, can occur in outbreaks or sporadically; named after first outbreak in 1983 in Brainerd, Minnesota
Brandt-Daroff maneuver	home treatment maneuvers for benign positional vertigo, used when office treatment maneuvers; series of provocative maneuvers done three sets per day for two weeks (Arch Otolaryngol 1980; 106:484-5)
Branham's sign	bradycardia after compression of AV fistula
Braxton-Hicks contractions	painless contractions of the uterine muscles during the second and third trimesters of pregnancy
Brechenmacher fibers	tracts which connect the atrium to the His bundle
Brenner tumor	benign ovarian tumor composed of epithelial cells in clusters within a deep fibrous stroma
Bricker procedure	creation of a urinary diversion by removing a piece of ileum, connecting the ureters to it, and then creating a stoma through the abdominal wall
Bright's disease	acute glomerulonephritis
Brill-Symmer's disease	nodular lymphoma
Brill-Zinser disease	recrudescent form of epidemic typhus (*Rickettsiae prowazekii*); occurs 10-50 years after primary infection; presents abrupty with chills, devers, headache, malaise; rash after 4-6 days after onset of symptoms
Briquet's syndrome	somatization disorder
Broca's area	left frontal speech area, important for articulating speech; in Broca's aphasia, because Broca's area near motor cortex and underlying internal capsule, a right hemiparesis and homonymous hemianopsia is almost always present in this type of aphasia
Brock's syndrome	right middle lobe atelectasis; more common in children with history of asthma or atopy
Brockenbrough sign	in hypertrophic cardiomyopathy, increase in murmur after a premature ventricular contraction, related to increase in LV systolic pressure, increase in systolic gradient between LV and aorta and consequent decrease in systolic pressure and pulse pressure
Brodie's abscess	small, intraosseus abscess that frequently involves the cortex and is walled off by reactive bone
Brodie-Trendelenburg test	for testing great saphenous vein and branches communicating with deep femoral vein; limb is elevated until drained of venous blood and then tourniquet is applied; normally arterial flow from below fills veins in about 35 s; however faster filling indicates incompetence of communicating vein
Brodman area	neuro anatomical classification of the cortex based on its cyto architecture into 52 areas; e.g. precentral gyrus Brodman 4, striate cortex Brodman 17
Brooke ileostomy	proctocolectomy with permanent ileostomy; a procedure used to treat ulcerative colitis, etc.
Broselow tape	tool for estimating weight and endotracheal tube size in pediatric population; developed by Broselow, an emergency physician in Hickory, North Carolina in the mid 1980s and by Luten
Brown's syndrome	in rheumatoid arthritis, vertical diplopia, clicking sensation when looking up and medially, and an apparent inferior oblique palsy, apparently from stenosing tenosynovitis of the superior oblique tendon and sheath; also congenital form
Brown-Séquard syndrome	loss of tactile sense, vibration sense, and limb position sense on the ipsilateral side and loss of pain and temperature sense on the contralateral side
Bruce protocol	multistage exercise treadmill test (seven 3 minute stages), described in 1963; developed by Robert A. Bruce (1916-2004), the "father of exercise cardiology"
Bruch's membrane	basal layer of the chorid between the choriocapillaris and the retinal pigment cell layer; deposits in this membrane in macular degeneration are known as drusen
Bruck syndrome	skeletal disorder resembling osteogenesis imperfecta with severe bone fragility and deformity; associated with congenital joint contractures; due to deficiency in telopeptide lysyl hydroxylase
Brudzinski sign	after flexing the neck, flexion of hips and knees in reaction suggests meningeal inflammation
Brueghel syndrome	dystonia of the motor trigeminal nerve producing a widely opened mouth, named after painting by Flemish painter Brueghel (*Neurol* 1996;46:1768)
Brugada syndrome	defect in an ion channel gene resulting in abnormal electrophysiologic activity in the right ventricle and characterized by (1) ST segment elevation in V1-V3, (2) right bundle branch block, (3) sudden cardiac death, (4) grossly normal heart; accounts for 40-60% of idiopathic ventricular fibrillation
Brunner's glands	in duodenum, submucosal mucous glands that secrete bicarbonate, glycoproteins, and pepsinogen II, virtually indistinguishable from pyloric mucous glands
Brushfield's spots	in Down's syndrome, small white spots on the periphery of the iris
Bruton's tyrosine kinase	mutation causes X-linked agammaglobulinemia (XLA also associated with defect in intact membrane-bound μ chain (it's essential for B-cell development) Btk found only in B cells
Budd-Chiari	occlusion of the hepatic vein, associated with polycythemia vera, pregnancy, postpartum state, oral contraceptives, paroxysmal

syndrome	nocturnal hemoglobinuria, and intra-abdominal cancers, particularly hepatocellular carcinoma
Buerger's disease	thromboangiitis obliterans, a nonatherosclerotic segmental inflammatory disease that most commonly affects the small and medium-sized arteries, veins, and nerves of the arms and legs. Acute phase reactants normal; strong association with tobacco use
Buerger's sign	in peripheral vascular disease, red foot becomes pale with elevation
Bunina bodies	intraneuronal inclusions felt to be pathognomonic for motor neuron disease
Burgdorf's reaction	acral erythema involving palms and soles after chemotherapy, originally reported in patients with AML receiving cytarabine
Burkitt's lymphoma	highly aggressive B cell lymphoma with tumor manifesting at extranodal sites (with endemic African variety, characteristically facial tumors); one of the fastest growing tumors in humans with doubling time 24-48 hours; associated with translocation of c-myc gene on chr 8 with IgH locus (chr 14), kappa (chr 2), or lambda light-chain (chr 22) locus; associated with EBV infection in African variety; named after Dennis Burkitt, British surgeon (1911-1993)
Burnett's syndrome	far-advanced milk-alkali syndrome, due to long-standing calcium and alkali ingestion; severe hypercalcemia, irreversible renal failure, and phosphate retention, may be accompanied by ectopic calcification; see also Cope's syndrome
Burow's solution	aluminum acetate in water, invented in mid-1800s by ophthalmologist Karl Burow (a.k.a. by its tradename Domeboro)
Burton's line	line at the interface of teeth and gums seen in chronic lead toxicity; from reaction of circulating lead with sulfur ions released by oral microbial activity
Buruli ulcer	caused by *Mycobacterium ulcerans*; painless nodule to large, undermined ulcerative lesions that heal spontaneously but slowly; named after Buruli region of Nile River
Buschke, scleredema of	uncommon dermatosis characterized by thickened, indurated skin associated with diabetes, sometimes with erythema; also known as scleredema diabeticorum
Buschke-Löwenstein tumor	verrucous carcinoma involving penile glans and prepuce, associated with HPV
Byler's disease	progressive familial intrahepatic cholestasis from impaired biliary secretion of both bile acids and phosphatidylcholine, leads to death from liver failure before adolescence
Cabot ring	in asplenia or malfunctional spleen, nuclear remnants on red blood cells as a thin, darkly-stained ring that follows the margin of the red cell
Cabrera's sign	in left bundle branch block complicated by MI, notching at 0.05 s in ascending limb of S wave in V3, V4; 27% sens for MI
Cacchi-Ricci disease	medullary sponge kidney disease
CADASIL	cerebral autosomal dominant arteropathy with subcortical infarcts and leukoencephalopathy, rare hereditary cause of stroke that may involve *Notch3* gene characterized by recurrent strokes (usually infarcts) and dementia
Cagot ear	absence of ear lobe; assocated with region in Pyrennes
caisson disease	decompression sickness
Cajal, interstitial cells of	cells present as networks of cells associated with neural plexues within gut musculature; required for normal intestinal motility
Calabar swellings	in loiasis, swellings 5-10 cm in diameter from localized areas of angioedema associated with migration of adult *Loa loa* worms through subcutaneous tissues
Call-Exner bodies	in granulosa cell tumors, small follicles filled with eosinophilic secretion; an important diagnostic feature
Call-Fleming syndrome	sudden-onset severe headache, focal neurological deficits, and seizures; associated with serotonin modulating drugs like SSRIs
Calot's triangle	the area bordered by the 1. cystic duct, 2. common hepatic duct, 3. lower edge of the liver; cystic artery, sometimes hepatic artery found here
Cameron lesions	erosions within incarcerated hiatal hernias, seen in 5.2% of patients with hiatal hernias
Campbell de Morgan spots	cherry angioma
Campbell diagram	used to determine the work of breathing, including the effects of chest wall compliance, lung compliance, and airway resistance
Campbell's sign	in chronic airway obstruction, downward motion of trachea during inspiration, perhaps due to downward pull of diaphragm
Camurati-Engelmann disease	progressive diaphyseal dysplasia; autosomal dominant, progressive diaphyseal dysplasia characterized by hyperosteosis and sclerosis of the diaphyses of long bones; associated with mutations in TGF beta 1
Canale-Smith syndrome	childhood disorder, first described in 1967, characterized by lymphadenopathy and autoimmunity; associated with mutations in Fas; implicates gene in accumulation of lymphocytes and the autoimmunity characteristic of the syndrome
Canavan disease	autosomal recessive infantile spongy degeneration of the brain with Alzheimer type II cells, increased prevalence among Ashkenazi from mutation in aspartoacylase leading to increased levels of N-acetylaspartic acid, described in 1931
Cantlie's line	separates the right and left lobes of the liver--a line drawn from the IVC to just left of the gallbladder fossa

Cantrell, pentalogy of	diaphragmatic defect (hernia), cardiac abnormality, omphalocele, pericardium malformation/absence, sternal cleft
Capgras syndrome	delusional belief that one or a few highly familiar people have been replaced by impostors who are physically very similar to the originals; seen rarely in schizophrenia.
Caplan's syndrome	coexistence of rheumatoid arthritis with a pneumoconiosis, leading to the development of distinctive pulmonary lesions that develop fairly rapidly; these nodular lesions have central necrosis surrounded by fibroblasts, macrophages, and collagen; can occur in asbestosis and silicosis
Carabello's sign	in severe aortic stenosis, during catheterization across aortic valve, an augmentation of the peripheral systolic pressure of more than 5 mm Hg during aortic pullback; from catheter further increasing the severity of the stenosis and removal resulting in a transient increase in stroke volume and systolic pressure (*Am J Cardiol* 1979;44:424–7)
Carey Coombs murmur	a blubbering apical mid diastolic murmur occurring in the acute stages of rheumatic mitral valvulitis and disappearing as the valvulitis subsides; described by Carey F. Coombs, English physician, 1879-1932
Carnett's test	head raise, tenderness persists in abdominal wall condition (rectus hematoma) whereas pain due to intraperitoneal disease lessens
Carney complex	autosomal dominant complex of cardiac myxomas with aggressive biologic behavior, spotty pigmentation, and association with endocrine tumors (causing e.g., Cushing syndrome); mutations in PRKAR1alpha gene (a protein kinase) account for half of the cases; unrelated to Carney syndrome
Carney syndrome (or triad)	nonfamilial disorder that includes combination of three rare tumors: gastric leiomyosarcoma, pulmonary chondroma, extraadrenal paraganglioma but no cardiac manifestations; unrelated to Carney complex
Caroli's disease	larger ducts of the intrahepatic biliary tree are segmentally dilated and may contain inspissated bile; pure forms are rare; this disease is usually associated with portal tract fibrosis of the congenital hepatic fibrosis type
Carpentier-Edwards valve	porcine valve, pressure-fixed, preserved in glutaraldehyde, mounted on a Teflon-covered Elgiloy strut
Carrión's disease	see Oroya fever
Carvajal syndrome	dilated cardiomyopathy, woolly hair, and keratoderma; associated with mutation in desmoplakin
Carvallo's sign	in tricuspid regurgitation, murmur increases with inspiration
Castellani's paint	topical fungicidal and bactericidal mixture of resorcinol (8 g), acetone (4 mL), magenta (0.4 g), phenol (4 g), boric acid (0.8 g), industrial methylated spirit 90% (8.5 mL), and water (to 100 mL); particularly effective against intertriginous tinea and acute *Candida* paronychia; named after Sir Aldo Castellani
Castle intrinsic factor	intrinsic factor secreted by parietal cells, which binds luminal B12 and permits its absorption in the ileum
Castleman's disease	lymphoproliferative disorder, either localized or multicentric; presents with massive lymphadenopathy, fever, splenomegaly, hepatomegaly; characterized by hyperplastic lymphoid follicles with capillary proliferation; associated with HHV 8; HIV-infected individuals at increased risk for multicentric Castleman's disease; multicentric can progressive to lymphoma
cat-scratch disease	tender regional lymphadenopathy persisting for 3 weeks or longer, frequently preceded by primary skin lesion after contact with kits, appears to be caused by *Bartonella* (formerly *Rochalimaea*) *henslae*, a small, pleomorphic gram-negative bacillus
Chaddock's sign	involuntary dorsiflexion of the toes when tapping from the lateral malleolus distally to the lateral dorsum of the foot in upper motor neuron defect
Chadwick's sign	blue-red passive hyperemia of the cervix that may appear after 7th week of pregnancy; may be seen in association with tumor; results from congestion of mucosa and most visible in anterior vaginal wall
Chagas's disease	zoonosis caused by protozoan parasite *Trypanosoma cruzi*; causes destruction of the myenteric plexus of the esophagus, duodenum, colon, and ureter, with resultant dilatation of these structures as well as cardiac disease; transmitted by excreta of hematophagous organisms of the family Reduiviidae
Chamberlain procedure	anterior mediastinotomy for staging lung cancer, direct approach into the mediastinum from the left second intercostals space; allows biopsy of AP window nodes which are the first N2 nodes for left upper lobe tumors
Chapman's sign	in left bundle branch block complicated by MI, notching of ascending limb of R in I, aVL, or V6
Char syndrome	heart-hand syndrome characterized by unusual facial features including long philtrum, down-slanting palpebral fissures, thick lips as well as patent ductus arteriosus and abnormal fifth digits
Charcot's disease	French eponym for ALS
Charcot's joints	neurogenic joint degeneration, can be secondary to syphilis, peripheral neuropathy
Charcot's triad	in 70% of patients with bacterial cholangitis, right-upper-quadrant pain, jaundice, and fever; see also Reynold's pentad
Charcot's triad	in multiple sclerosis, nystagmus, intention tremor, and staccato speech (or scanning speech)
Charcot-Bouchard aneurysms	minute brain aneurysms from chronic hypertension, occurring in vessels less than 300 microns in diameter (different from saccular aneurysms), most commonly found within basal ganglia
Charcot-Leyden's crystals	crystals in the shape of elongated double pyramids, formed from eosinophils (from lysophospholipase), found in the sputum in bronchial asthma and in other exudates or transudates containing eosinophils

Charcot-Marie-Tooth disease — most common inherited peripheral neuropathy, 1/2500, autosomal dominant, clinically heterogeneous disorder characterized by slowly progressive atrophy of the distal muscles, mainly those innervated by peroneal nerve; progressive weakness of varying intensity and atrophy of the muscles of the feet, hands, and legs, leading to pes cavus, clawhand, and stork-leg appearance, usually beginning in the 2nd or 3rd decade. Enlarged greater auricular nerves may be visible and enlarged ulnar and peroneal nerves may be palpated in some patients. Cranial nerves rarely involved

CHARGE assocation — coloboma, heart malformation, atresia choanae, retarded growth and development, and/or CNS anomalies, genital hypoplasia, ear anomalies and/or deafness

Charles Bonnet syndrome — visual deprivation hallucinations, generally occurring in visually-impaired individuals; patients realize unreality of hallucinations; first described by Swiss philosopher Charles Bonnet in 1760

Charlin's syndrome of neuralgia — severe pain in the inner corner of the eye disproportionate to the degree of ocular inflammation, pain in the root of the nose, tearing, and ipsilateral nasal watery discharge believed to be due to irritation of the ciliary ganglion

Chédiak-Higashi syndrome — recurrent pyogenic infections, partial albinism, multiple neurologic abnormalities; due to mutation in *LYST*; gene involved in intracellular protein transport to and from lysosome; neurologic abnormalities include photophobia, nystagmus, peripheral neuropathy, etc.; associated with neutropenia, defective degranulation, delayed microbial killing; abnormal giant granules in neutrophils

Cheyne-Stokes breathing — regularly irregular pattern characterized by a progressive increase in the depth and at times frequency of respiration with a crescendo-decrescendo shape that eventually culminates in an apneic phase; seen in CHF but also in meningitis, CVA, pontine damage

Chiari-Frommel syndrome — persistent galactorrhea-amenhorrhea after pregnancy

Chikungunya fever — alphavirus and arbovirus, cycles of activity every 5-10 years; resembles dengue fever, characterized by severe arthritis; spread by Aedes mosquitoes, speculated as risk factor for Burkitt's lymphoma

Chilaiditi syndrome — when redundant loops of transverse colon slip between the liver and diaphragm and cause vovulus

Child-Pugh classification system — classification for severity of liver disease according to degree of ascites, bilirubin, albumin, prothrombin time, and encephalopathy; initially used to predict mortality after surgery

Christmas disease — hemophilia B, deficiency in factor IX. First coagulation protein to be named after a patient, Stephen Christmas

Churg-Strauss syndrome — allergic angiitis and granulomatosis involvement in the lung, associated with eosinophilia; asthma is the cardinal feature (occurs in 95%) and precedes vasculitic phase by 8-10 years; 2/3 of patients have skin lesions which can appear as s.c. nodules on extensor surfaces; 70% have P-ANCA; 4/6 following criteria 85% sens and 99.7% spec: asthma; eosinophilia>10%; neuropathy; pulmonary opacities; paranasal sinus abnormality; biopsy of blood vessel showing eosinophils in extravascular area

Chuvash polycythemia — autosomal recessive disorder with features of both primary and secondary polycythemia, endemic to the mid-Volga River region of Russia (Chuvas); associated with homozygous mutation Arg200Trp in *VHL* gene (gene associated with von Hippel-Lindau syndrome); mutation impairs interaction of VHL with hypoxia-inducible factor 1 protein

Chvostek's sign — hypocalcemia, in latent tetany, tapping the facial nerve against the bone just anterior to the ear producing ipsilateral contraction of facial muscles

Civatte bodies — aka colloid bodies; in lichen planus; anucleate, necrotic basal cells becoming incorporated into the inflamed papillary epidermis

Clagett's procedure — for treating empyema, open drainage followed by instillation of antibiotic solution

Clara cells — cells found in the epithelium of terminal and respiratory bronchioles, devoid of cilia, present secretory granules in their apex and are known to secrete glycosoaminoglycans that probably protect the bronchiolar lining

Claude's syndrome — third nerve palsy combined with contralateral ataxia with midbrain infarction and injury to both red nucleus and superior cerebellar peduncle (includes signs of both Nothnagel's and Benedikt's syndrome)

clay shoveler's fracture — fracture of spinous process of C7

Clemmesen's hook — change in rates of breast cancer following menopause. First described in 1948.

Clerambault's syndrome — erotomania, delusional belief that someone (usually of higher social status) is in love with the person

Clichy criteria — criteria for liver transplantation in fulminant hepatic failure: hepatic encephalopathy, and factor V level < 20% in patients younger than 30 y.o. and factor V level < 30% in patients > 30 y.o.

Cloquet's node — first lymph node underneath the inguinal ligament; can be mistaken for femoral hernia when enlarged

Clutton's joints — in congenital syphilis, symmetrical arthrosis, especially of the knee joints

Cockayne's syndrome — a form of progeria characterized by dwarfism, pigmentary degeneration of the retina, optic atrophy, deafness, sensitivity to sunlight, and mental retardation; autosomal recessive inheritance defect in DNA repair

Cockcroft-Gault formula — for calculating clearance based on creatinine, age, weight: [(140-age) x lean body weight (in kg)]/[plasma creatinine (in mg/dL) x 72]; in women, multipled by 0.85 because of smaller muscle mass

Codman's triangle — in osteosarcoma, the triangular shadow between the cortex and raised ends of periosteum is known radiographically as Codman's triangle, and is characteristic but not diagnostic of this tumor

Cogan syndrome	multisystem inflammatory vascular disease particularly of the CNS characterized by eye (interstitial keratitis) and ear (vestibuloauditory) involvement; can be associated with aortitis; peak incidence third decade of life
Cole-Carpenter syndrome	craniosynostosis, ocular proptosis, associated with severe bone fragility and deformity
Coley's toxin	mixture of toxins from Strep and Serratia marcesens used by William Coley in the late 1800s to treat malignancy; active agent found to be lipopolysaccharide
Colles's fracture	fracture of the distal radius, extra-articular; classically occurs when persons fall with outstretched hands
Collet-Sicard syndrome	acquired palsy of all lower four cranial nerves (IX, X, XI, and XII); can occur in occipital fracture, burst fracture of cervical vertebra, carotid dissection, malignancy; described by Frederic Collet and Jean Sicard;
Collier's sign	lid retraction in lesion of posterior commissure
Conn's syndrome	primary hyperaldosteronism, caused by an aldosterone-secreting tumor, resulting in hypertension, hypokalemia, hypernatremia, metabolic alkalosis, and low plasma renin
Conradi-Hunermann syndrome	chondrodysplasia punctata, characterized by stippled epiphyses from abnormal accumulation of calcium salts and skeletal changes
Conradi-Hunermann-Happle syndrome	X-linked dominant variant of Conradi-Hunermann syndrome or chondrodysplasia punctata, associated with linear ichthyosis, cataract, and short stature; associated with mutation in delta8-delta7 sterol isomerase emopamil binding protein, a protein involved in cholesterol metabolism
Cooley's anemia	homozygous beta thalassemia; Mediterranean anemia
Coombs test	direct, ability of anti-IgG or anti-C3 antisera to agglutinate the patient's red blood cells; cold reacting antibodies react with anti-C3 (mostly drug-related antibodies, IgM antibodies (generally to polysaccharide), IgG antibodies of low affinity); indirect Coombs, serum of the patient is incubated with normal red cells, though IgM antibodies may agglutinate directly
Cooper's hernia	hernia through the femoral canal and tracking into the scrotum or labia majus
Coopernail's sign	ecchymosis of the perineum and scrotum or labia, reflecting pelvic fracture; after George Peter Coopernail, American physician (1876-1962).
Cope's syndrome	subacute milk-alkali syndrome; see also Burnett's syndrome
Cori's disease	glycogen storage disease type III, deficiency in debranching enzyme, amylo-1,6-glucosidase, leading to variable accumulation of glycogen in the liver, heart, or skeletal muscle, characterized by stunted growth, hepatomegaly, and hypoglycemia; also known as Forbes disease
Cornelia de Lange syndrome	complex developmental disorder consisting of characteristic facial features, upper limb abnormalities, hirsutism, ophthalmologic involvement, gastroesophageal dysfunction, hearing loss, as well as growth and developmental retardation
Corrigan's pulse	in aortic regurgitation, pulses are of the water-hammer or collapsing type with abrupt distension and quick collapse, can be exaggerated by raising the patient's arm
Costello's syndrome	syndrome of prenatally increased growth, postnatal growth retardation, coarse face, loose skin resembling cutis laxa, nonprogressive cardiomyopathy, developmental delay, and an outgoing, friendly behavior
Costen's syndrome	ear pain, tinnitus, impaired hearing, and dizziness from temporomandibular joint dysfunction
Cotard's syndrome	range of delusions from believing that one has lost organs to believe that one is dead or does not exist; seen in schizophrenia and also in organic lesions of the nondominant temporoparietal cortex as well as in migraine.
Coumel's sign	prolongation of the tachycardia cycle length in the presence of an ipsilateral bundle branch block and bypass tract
Coumel's triangle of arrhythmogenesis	three factors in arrhythmogenesis: 1. the arrhythmogenic substrate, 2. the trigger factor, and 3. the modulation factors of which the most
Councilman bodies	in apoptosis, hepatocytes that round up to form shrunken, pyknotic, and intensely eosinophilic bodies
Courvoisier's law	tumors that obstruct the common bile duct result in an enlarged bladder; obstructing stones do not, since the gallbladder is typically too scarred to allow enlargement; present in half of pancreatic CA
Cowden disease	autosomal dominant condition associated with multiple hamartomatous lesions, especially of the skin, mucous membranes, GI tract, breast and thyroid; associated with trichilemmomas (multiple skin-colored warty papules); associated with mutations in *PTEN/MMAC1*
Cowdry body	intranuclear inclusion seen in herpes virus infection
Cowper's gland	bulbourethral gland; small glands located beneath the prostate; involved in pre-ejaculatory secretions; homologous to Bartholin's gland; named after English anatomist William Cowper (1666-1709)
Cox maze procedure	surgical procedure developed in 1987 for atrial fibrillation where a tortuous path is created between the SA node to the AV node and incisions are placed so that atrial fibrillation cannot be sustained

Creutzfeld-Jakob disease	fatal transmissible disorder of the CNS characterized by rapidly progressive dementia and variable focal involvement of the cerebral cortex, basal ganglia, cerebellum, brainstem, and spinal cord, attributable to prions, characterized by myoclonus often induced by a startle, extrapyramidal signs, cerebellar signs; slowing and periodic complexes on EEG; 1 per 167,000 in U.S.; fatal within 1 year after onset of symptoms
cri du chat	5p-, severe mental retardation, microcephaly, catlike cry, low birth weight, hypertelorism, low-set ears, and epicanthal folds
Crigler-Najjar syndrome	type I, no hepatic glucoronyltransferase activity, kernicterus, requires liver transplantation; type II, moderate deficiency of glucoronyltransferase, phenobarb induces activity
Crohn's disease	transmural mucosal inflammation that may involve the entire GI tract from mouth to the perinanal area that often leads to fibrosis and obstructive clinical presentations; 80% small bowel involvement, usually distal ileum; 1/3 exclusively ileitis; 50% ileocolitis; and 20% disease limited to colon
Cronkhite-Canada syndrome	diffuse GI hamartoma polyps (i.e., no cancer potential) associated with malabsorption/weight loss, diarrhea and loss of electrolytes/protein; signs include alopecia, nail atrophy, and skin pigmentation
Crouzon syndrome	craniosynostosis correlated in mutations with the extracellular domain of FGFR2
Crowe's sign	axillary or inguinal freckling seen in 20-50% of neurofibromatosis
Crow-Fukase syndrome	POEMS (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, skin changes); known as Crow-Fukase syndrome in Japan
Cruveihiler-Baumgarten bruit	bruit heard over caput medusa in portal hypertension
Cullen's sign	a faintly blue coloration particularly of umbilicus as the result of retroperitoneal bleeding from any cause, but especially in ruptured ectopic pregnancy; also seen in acute pancreatitis (1-2%)
Curling's ulcers	stress erosions and ulcers occurring in the proximal duodenum and associated with severe burns or trauma, from ischemia of the gastric mucosa
Currarino syndrome	childhood familial idiopathic osteoarthropathy
Currarino triad	partial sacral agenesis with intact first sacral vertebra ('sickle-shaped sacrum'), a presacral mass, and anorectal malformation; associated with a mutation in a homeo box gene, *HLXB9*
Currarino-Silverman syndrome	premature obliteration of sternal sutures; associated with pectus carinatum appearance
Curschmann's spirals	spirally twisted masses of mucus plugs containing whirls of shed epithelium occurring in the sputum in bronchial asthma; Heinrich Curschmann, German physician, 1846-1910
Cushing reaction	increase in intracranial pressure causes compression of the cerebral blood vessels and cerebral ischemia, reaction of elevation in pressure with simultaneous reduction in heart rate, respiratory slowing
Cushing's disease	hypercortisolism from pituitary corticotropin-(ACTH)-secreting corticotroph tumors, almost always benign and usually microadenomas
Cushing's syndrome	hypercortisolism
Cushing's triad	signs of increased ICP 1. hypertension 2. bradycardia 3. irregular respirations
Cushing's ulcer	acute ulcer of the stomach, proximal duodenum, or esophagus, frequently leads to hemorrhage or perforation, associated with intracranial injury or increases in intracranial pressure, associated with gastric acid hypersecretion
D'Espine's sign	breath sounds louder over C7 than adjacent lung, suggests lesion in posterior mediastinum, e.g. lymphoma, tuberculosis, etc.
Da Costa syndrome	neurocirculatory asthenia, pain localized typically to the cardiac apex and consists of dull, persistent ache that lasts for hours, etc.
Dacie's syndrome	idiopathic hyperplastic enlargement of the spleen with anemia and neutropenia; progression to lymphoma in some cases
Dahl's sign	in COPD, protracted pressure applied by the elbows leads eventually to the formation of two patches of hyperpigmented calluses immediately above the knees
Dakin's solution	dilute solution of sodium hypochlorite (0.5%) used for cleansing wounds
Dallas criteria	for histologic diagnosis of myocarditis, active myocarditis if infiltrating lymphocytes and myocytolysis seen; borderline if only lymphocytic infiltration; and negative if both absent; introduced in 1986
Dalrymple sign	retraction of the upper eyelid in Graves's disease, causing abnormal wideness of the palpebral fissure
Daltonism	color blindness, from John Dalton (who proposed atomic theory), born 1766; felt that color blindness was from vitreous humor being blue, disproved his assistant Joseph Ransome who examined his eyeballs post mortem
Damus-Kaye-Stansel procedure	repair of congenital transposition of the great arteries of the heart by dividing the pulmonary artery and attaching the proximal section to the ascending aorta and connecting distal section to right ventricle
Dance's sign	empty right lower quadrant in children with ileocecal intussusception
Dandy-Walker	hydrocephalus resulting from failure of the foramina Luschka and Magendie to open; associated with an occipital meningocele and agenesis of the cerebellar vermis and splenium of the corpus callosum; associated with warfarin use during pregnancy

syndrome	
Dane particle	mature HBV virion, 42 nm, double-layered, genome is double-stranded circular DNA, all regions of genome encode stuff
Darier's sign	in mastocytosis (urticaria pigmentosa), is positive when a brown macular or a slightly papular lesion becomes a palpable wheal after being vigorously rubbed with the blunt end of an instrument such as a pen; wheal may not appear for 5-10 minutes
Darier-White disease	keratosis follicularis; autosomal dominant disorder characterized by warty papules and plaques in seborrheic areas (central trunk, flexures, scalp, and forehead), palmoplantar pits, and nail abnormalities; associated with mutation in SERCA2 Ca(2+)-ATPase.
Darkschewitsch, nucleus of	an ovoid cell group in the ventral central gray substance rostral to the oculomotor nucleus, receiving fibers from the vestibular nuclei by way of the medial longitudinal fasiculus; projections are not known, although some cross in the posterior commissure
Darwin's tubercle	completely benign and congenital nodule, located near the superior aspect of the auricle (approximately 2/3 of the way from the bottom of the helix), described by Charles Darwin
Dawson's fingers	in multiple sclerosis, perivascular demyelination creating the appearance of finger projections oriented transversely on an axial scan
de Musset's sign	in aortic regurgitation, head bobbing, named after 19th century French poet who had luetic aortic insufficiency
de Quervain's disease	a stenosing tenosynovitis of the thumb extensors and abductors; pain elicited with Finkelstein's test
de Quervain's thyroiditis	subacute granulomatous thyroiditis, viral etiology suspected
Degos disease	malignant atrophic papulosis; multisystem lymphocytic vasculitis characterized by thrombosis primarily of cutaneous small vessels as well as small vassels in GI tract, ocular, and CNS; has characteristic porcelain white atrophic lesion
Dejerine-Roussy syndrome	thalamic lesions causing sensory loss, spontaneous pain, and perverted cutaneous sensation described in 1906
Dejerine-Sottas disease	hereditary motor and sensory neuropathy III, recessive inheritance, slowly progressive demyelinating disorer, presents in infancy or childhood and progresses to cause severe disability by 30s; nerves typically enlarged
Delphian nodes	lymph nodes in midline of the thyrohyoid membrane; can be involved in thyroid cancer or subacute thyroiditis
Dennie's lines	in atopic dermatitis, an accentuated line or fold below the margin of the lower eyelid
Denonvillier's fascia	fascia that separates prostate and seminal vesicles from rectum
Dent's disease	X-linked syndrome characterized by renal proximal tubular dysfunction, proteinuria, hypercalciuria, nephrocalcinosis, nephrolithiasis, and rickets due to mutation in voltage-gated chloride channel
Denver shunt	for treating refractory ascites, peritoneovenous shunt connecting peritoneum and central venous system, compare with LeVeen shunt; Denver shunt uses valve that lies within a fluid-filled, compressible silicone chamber
Denys-Drash syndrome	gonadal dysgenesis (male pseudohermaphroditism) and nephropathy leading to renal failure (nephrotic syndrome); increased risk of Wilms's tumor; dominant negative missense mutation of *WT-1* gene
Dercum's disease	adipositas dolorsa, condition tending to affect obese women in middle age, mostly menopausal, consisting of multiple exquisitely tender lipomas
Desçemet's membrane	membrane that forms the deepest layer of the cornea and functions as thin basement membrane for endothelium; location where copper is deposited in Wilson's disease Kayser-Fleischer rings
Destot's sign	scrotal hematoma suggesting pelvic fracture
Deutschlander's fracture	fracture from overuse, e.g. marching
Devic's disease	neuromyelitis optica; relapsing-remitting demyelinating disorder characterizing by bilateral optic neuritis and transverse myelitis occurring in rapid succession; common in Asians; distinct from multiple sclerosis
Devon family syndrome	inherited condition characterized by inflammatory fibroid polyps
Diamond-Blackfan anemia	congenital pure red cell aplasia characterized by increased MCV, reticulocytopenia, bone marrow erythroblastopenia; can be diagnosed by increased erythrocyte adenosine deaminase; 40% associated with congenital abnormalities; associated with RPS19 ribosomal protein mutations in some; responds to steroids which increase Epo sensitivity
Dick test	injection of erythogenic toxin of *Strep. pyogenes*, positive result in those lacking antitoxin
Dieulafoy lesion	around 6 cm from the gastroesophageal junction, an uncommon cause of massive GI bleeding, a large submucosal artery which erodes the mucosa without any overlying ulceration or other obvious mucosal damage
DiGeorge syndrome	failure of 3rd and 4th pharyngeal pouches to differentiate into the thymus and parathyroid glands, facial abnormalities result primarily from abnormal development of the first arch components during formation of face and ears, 22q11 deletion, CATCH-22 (cardiac abnormality/abnormal facies, T-cell deficit owing to thymic hypoplasia, cleft palate, and hypocalcemia)
DiGuglielmo's disease	AML M6, erythroleukemia; felt to emerge from myelodysplastic state
dimple sign	in dermatofibroma, lateral compression with thumb and index finger produces a depression, or "dimple."

Disse, space of subendothelial space in liver separating endothelial cells from underdlying hepatocytes which contains hepatocyte microvilli

Dix-Hallpike test for testing benign paroxysmal positional vertigo, examiner stands at the patient's right side and rotates the patient's head 45 degrees to the right to align the right posterior semicircular canal with the sagital plane of the body; the examiner moves the patient, whose eyes are open, from the seated to the supine right-ear-down position and then extends the patient's neck slightly so that the chin is pointed slightly upward.

Dobrin's syndrome TINU syndrome (tubulointerstitial nephritis with uveitis), associated with bone marrow granulomas, first described in 1975

Dock's murmur in left anterior descending artery stenosis, diastolic murmur similar to that of aortic regurgitation

Doderlein's lactobacilli *Lactobacillus acidophilus*; metabolizes the glycogen in glycogen rich cells which are exfoliated in the vagina pre-menopause to lactic acid, resulting in a decrease in pH to 3.5-4

Döhle bodies irregularly shaped blue to gray-blue inclusions in neutrophil cytoplasm, consisting of ribosomes and/or rough ER; seen in severe bacterial infections

Doi's sign elicitation of diminished deep tendon reflexes after maximal voluntary contraction in Lambert-Eaton syndrome

Donath-Landsteiner antibody in paroxysmal cold hemoglobinuria, an antibody associated with syphilis and viral infections, directed against the P red cell erythrocyte antigen and can induce complement-mediated lysis; attacks precipitated by exposure to cold and are associated with hemoglobinemia and hemoglobinuria; chills and fever; back, leg, and abdominal pain; headache and malaise; recovery prompt; asymptomatic otherwise

Donnai-Barrow syndrome syndrome of diaphragmatic hernia, exomphalos, hypertelorism, agenesis of the corpus callousm, severe sensorineural deafness, and severe myopia

Donovan bodies in Calymmatobacterium granulomatis or granuloma inguinale, bodies characterized by multiple organisms filling large histiocytes

Dor procedure endoventricular circular patch plasty, a surgical procedure for treating postinfarction aneurysm where a purse string stich is sewed around the aneursym

Dorello's canal location where the sixth nerve penetrates the dura, an area where the nerve is liable to injury

Douglas, pouch of rectouterine pouch

Down syndrome trisomy 21 (in 95%), 1/700 births, 1% mosaics, 40% have congenital heart disease. 10-20 fold increased risk of acute leukemia, virtually all older than 40 develop Alzheimer's disease, abnormal immune system

Dressler's beat fusion beat seen in ventricular tachycardia

Dressler's syndrome pericarditis, possible autoimmune etiology, found to develop 2 weeks to several months after acute MI

Druckrey relationship relationship between carcinogen dose and tumor induction time: $dt^n = k$ where d is the dose of carcinogen, t is latency period, n is the slope of the double log [plot of carcinogen dose versus induction time, and k is a constant

Drummond, marginal artery of vessel which lies along mesenteric border of colon and supplies vasa recta; arises from right and left branches of middle colic artery and left and right colic arteries; also supplies collateral connection between superior and inferior mesenteric arteries

Duane's syndrome form of strabismus, congenital absence of cranial nerve 6, resulting in impaired abduction and/or adduction and eyeball retraction and narrowing of palpebral fissure during adduction of affected eye

Dubin-Johnson syndrome mostly conjugated hyperbilirubinemia from defect in transport of bilirubin and other organic anions across the canaliculus; other liver function tests normal; accumulation of dark pigment in liver lysosomes

Dubowitz syndrome malformation syndrome characterized by intrauterine growth retardation, short stature, microcephaly, mild mental retardation with behavior problems, eczema, and unusual and distinctive facies

Duchenne's muscular dystrophy X-linked, near or complete absence of dystrophin (normally stabilizes glycoprotein complex on cytoplasmic face of plasma membrane of muscle fibers and protects it from degradation), onset of weakness age 2-3, proximal weakness of limb muscles, Gower's maneuver, pseudohypertrophy of calves caused by fatty infiltration, cardiomyopathy, frequently mental retardation, CK elevated, wheelchair bound by age of 12

Duffy blood group system named after Mr. Duffy, a patient with hemophilia in whom anti-Fya was discovered by Cutbush in 1950; abbreviation Fy used because "Du" had already been taken; Fy6 antigen is the receptor for *Plasmodium vivax*

Dukes's disease fourth disease; see Filatov-Dukes's disease

Duncan's disease X-linked lymphoproliferative syndrome, normal response to childhood infections but develop fatal lymphoproliferative disorders after infection with EBV; most patients with this syndrome die of infectious mononucleosis

Dunnigan syndrome face-sparing partial lipodystrophy, loss of subcutaneous fat and increase in visceral fat, causing muscular appearance in arms and legs, due to mutation in laminin A/C

Dunphy sign increased pain with coughing in appendicitis

Dupuytren's contracture palmar fibromatosis

Durant's maneuver left lateral decubitus position, used in managing air embolism

Duret hemorrhage hemorrhage from uncal herniation

Duroziez's sign in aortic regurgitation, systolic murmur heard over the femoral artery when it is compressed proximally and a diastolic murmur when

	it is compressed distally
Dutcher bodies	PAS-positive inclusions containing Ig in the nucleus of lymphocytes, plasma cells, and intermediate lymphocytes in Waldenström macroglobulinemia and multiple myeloma
Eagle effect	failure of penicillin in streptococcal infection when bacteria aren't growing, i.e., beta-lactams being more effective against rapidly growing bacteria in the early stages and less effective as bacterial growth slows (described by Eagle in 1952)
Eagle-Barrett syndrome	prune-belly syndrome with triad of abdominal muscle deficiency, urinary tract abnormalities, and cryptorchidism
Eale's disease	isolated, peripheral retinal vasculitis
Ebstein's anomaly	malformation characterized by the downward displacement of the tricuspid valve into the right ventricle due to anomalous attachment of the tricuspid leaflets; associated with maternal exposure to Li
economy class syndrome	pulmonary embolism after travel, described by Symington and Stack in *Br J Dis Chest* 1977; 71:138-40
Edeiken pattern	normal variant ST segment elevation seen V2 or V3 where there is a shallow upward concavity; pattern disappears when leads are recorded one interspace lower than usual (*Am Heart J* 1954;48:331).
Edinger-Westphal nucleus	part of the cranial nerve III complex involved in direct and consensual light reflex involved in efferent limb of reflex arc
Edwards's syndrome	trisomy 18, 1/8,000 births, mental retardation, prominent occiput, micrognathia, low-set ears, rocker-bottom feet, flexion deformities of the fingers, and congenital heart disease
Ehlers-Danlos syndromes	clinically and genetically heterogeneous group of disorders that result from defect in collagen synthesis or structure, at least 10 variants, characterized by hyperextensible skin and hypermobile joints
Ehrlich's reaction	reaction where *p*-dimethylaminobenzaldehyde reacts with urobilinogen in a strong acid medium to produce a brown-orange color
Eisenmenger complex	a ventricular septal defect with right ventricular hypertrophy, severe pulmonary hypertension, and frequent straddling of the defect by a misplaced aortic root
Eisenmenger syndrome	cardiac failure with significant right to left shunt producing cyanosis due to higher pressure on the right side of the shunt; usually due to the Eisenmenger complex, any anomalous circulatory communication that leads to obliterative pulmonary vascular disease
Ekbom syndrome	restless leg syndrome
Ekiri syndrome	extremely rare, fatal encephalopathy described in Japanese children with *Shigella sonnei* or *Shigella flexneri* infections
Elejalde syndrome	neuroectodermal melanolysosomal disease characterized by silvery hair, CNS dysfunction, abnormal melanocytes and melanosomes, and abnormal inclusion bodies in fibroblasts and other cells; similar dermatologic features to Chédiak-Higashi syndrome and Griscelli syndrome but without defect in immune system
Ellis-van Creveld syndrome	chondroectodermdal dysplasia, short-limbed dwarfism, polydactyly, single atrium or atrial septal defect, autosomal recessive, reported most often in Amish
Ellsworth-Howard test	for diagnosis of pseudohypoparathyroidism by assessing response to exogenous PTH
Elschnig spots	yellow (early) or hyperpigmented (late) patches of retinal pigment epithelium overlying infarcted choriocapillaris lobules in hypertensive retinopathy
Emery-Dreifuss muscular dystrophy	distinctive form of muscular dystrophy with humero-peroneal weakness and quite pronounced muscle contractions and by severe cardiac arrhythmias which may cause sudden death; due to mutations in lamin A/C gene on 1q21.2-q21.3
Epley maneuver	in benign paroxysmal positional vertigo, a treatment maneuver for moving calcium carbonate debris to the common crus of the anterior and posterior canals and exit into the utricular canal; symptoms are improved in 90% of patients with no recurrence in 50-70% (also see Semont maneuver and Brandt-Daroff exercises)
Epping jaundice	outbreak of jaundice that occurred in 84 individuals after ingestion of bread made with flour contaminated with 4,4'-diaminodiphenylmethane in England in 1965
Epsom salts	magnesium sulfate, laxative
Epstein's pearls	small, white cysts along the median raphe of the hard palate
Epstein-Barr virus	EBV binds to CD21 found on epithelial cells and B cells; however a large number of T suppressor cells and EBV specific cells are seen as atypical lymphocytes
Erb's palsy	upper plexus palsy affecting C5 and C6 and +/- C7 nerve roots associated with weakness of shoulder and arm
Erdheim-Chester disease	Rare, rapidly fatal, multisystem histiocytosis syndrome that usually affects adults, characterized by bone pain, xanthomas, xanthelasma, exophthalmos, diabetes insipidus, retroperitoneal, and pulmonary disease; tropism for adipose and connective tissues
Erlenmeyer's flask deformity	seen in osteopetrosis, where the ends of long bones are bulbous

Esmarch bandage	rubber bandage applied around a part from distal to proximal in order to expel blood from it (Johannes Friedrich August von Esmarch, German surgeon, 1823-1908)
eustachian tube	a canal leading from the upper part of the pharynx to the middle ear; after Bartolomeo Eustachi, Italian anatomist (died 1574)
eustachian valve	in the fetus, a membranous fold at the opening of the IVC that directs blood flow from the IVC to the foramen ovale; after closure of the foramen ovale, no specific function; prominent eustachian valve more commonly found in patients with presumed paradoxical embolism; after Bartolomeo Eustachi, Italian anatomist (died 1574)
Evan's syndrome	ITP and immunohemolytic anemia
Ewart's sign	in large pericardial effusion, dullness to percussion of the left lung over the angle of the scapula may occur; due to compressive atelectasis by the large pericardial sac
Ewing's sarcoma	same tumor as primitive neuroectodermal tumor; onion skinning; small round cell tumor of bone, 85% of cases there is a t(11;22)(q24:q12) translocation; second most common cancer of bone in children and adolescents
Fabricius, bursa of	in birds, thymus-like gland that is an outgrowth of the cloaca and site for for B cell maturation
Fabry's disease	X-linked recessive sphingolipidosis (sulfatidose) alpha-galactosidase deficiency resulting in increased globosides, reddish-purple skin rash, kidney and heart failure, peripheral neuropathy, pain in lower extremities (acroparesthesias); 1:117,000 live births; most common clinical features are telangiectases and corneal dystrophy; can be trated with biweekly infusions of recombinant human alpha-galactosidase A
factor V Leiden	Arg(506)->Gln mutation, results in resistance to cleavage by activated protein C (an anticoagulant), found in 20% of patients with venous thromboembolism, 6% of U.S. population
Fahr's disease	neurodegenerative syndrome associated with symmetric intracerebral calcifications in basal ganglia, associated with cognitive and movement disorders including spastic paralysis, athetosis
Fallot, tetralogy of	1. ventricular septal defect; 2. infundibular, valvar, or supravalvar pulmonic stenosis; 3. an anteriorly displaced aorta that receives blood from both ventricles; 4. right ventricular hypertrophy
Fanconi's anemia	autosomal recessive, predisposal to aplastic anemia, progressive bone failure at age 5-7, congenital malformations (frequently defects in thumbs or forearms, kidneys, GI, skin); 20% develop cancer (primarily AML but also in skin, GI, GU); diagnosed by increased sensitivity of FA cells to bifunctional alkylating agents (e.g. diepoxybutane or mitomycin C)
Fanconi's syndrome	generalized dysfunction of proximal renal tubule leading to glycosuria, hyperphosphaturia, hypophasphatemia, aminoaciduria, and systemic acidosis; may be associated with out-dated tetracyclines
Fanconi-Bickel syndrome	type XI glycogen storage disease, hepatic glycogenosis with renal Fanconi syndrome; caused by defect in GLUT-2 transporter, characterized by proximal renal tubular dysfunction, impaired glucose and galactose utilization, liver and kidney glycogen accumulation; presents as failure to thrive in first year with hepatomegaly and kidney enlargement
Farber's disease	sphingolipidosis from ceraminidase deficiency leading to painful and progressively deformed joints, subcutaneous nodules, granulomas, fatal in early life
farmer's lung	hypersensitivity pneumonitis from exposure to antigens of microorganisms which colonize equipment used in farming, including from moldy hay, grain, silage (e.g. thermophilic actinomycetes, fungus such as Aspergillus umbrosus)
farmer's skin	cutis rhomboidalis nuchae
Fay sign	in carotid arteritis, pressure along the carotid causing pain to spread in distal branches of external carotid to jaw, ear, and temple
Fechtner syndrome	Alport syndrome with leukocyte inclusions and macrothrombocytopenia; associated with mutations in nonmuscle myosin heavy chain-9
Felty's syndrome	rheumatoid arthritis, splenomegaly, and neutropenia, and leg ulcers; associated with HLA-DR4
Ferguson's reflex	anesthesia in ob, interruption of oxytocin release in response to cervical dilatation may cause uterine inhibition
fetor hepaticus	"musty" or "sweet odor" from the formation of mercaptans by the action of GI bacteria on the sulfur-containing amino acid methionine and shunting of splanchnic blood from the portal into the system circulation (portosystemic shunting)
fifth disease	erythema infectiosum, associated with parvovirus B19 infection, characterized by "slapped cheeks" and erythematous lacy eruption on the trunk and extremities
Filatov-Dukes's disease	fourth disease, historically felt to be a separate form of rubella and then felt to be a non-entity but perhaps now is *Staph* scarlet fever Lancet 357:299 (2001); alternatively misdiagnosed cases of rubella or scarlet fever
Finkelstein's test	in de Quervain's tenosynovitis, dorsal thumb pain when the wrist is deviated in an ulnar fashion and the thumb is flexed across the palm
first disease	measles, aka rubeola, caused by paramyxovirus, described in 1627
Fisher's syndrome	see Miller-Fisher syndrome
Fitzgerald factor	high molecular weight kininogen; in intrinsic phase of coagulation, part of complex between factor XII, prekallikrein, and subendothelial collagen; facilitates conversion of XII to XIIa; deficiency not associated with bleeding
Fitz-Hugh-Curtis syndrome	associated with spread of gonococci or chlamydia: perihepatitis manifested by right upper quadrant or bilateral upper abdominal pain and tenderness and occasionally by a hepatic friction rib

Flatbush diabetes	GAD antibody negative diabetes in adult black subjects with diabetic ketoacidosis and increased frequency of human leukocyte antigen DR3 and DR4
Fletcher factor	prekallikrein; in intrinsic phase of coagulation, converts XII to XIIa; deficiency results in elevated PTT but with no clinical bleeding
Foix-Alajouanine disease	angiodysgenetic necrotizing myelopathy, venous angiomatous malformation of the spinal cord and overlying meninges associated with ischemic damage and worsening neurologic symptoms in lumbosacral cord
Fontan procedure	for treating tricuspid atresia, pulmonary atresia, or hypoplastic left heart syndrome, procedure developed in 1968 whereby IVC is connected to pulmonary artery; long term complications include arrhythmias and protein losing enteropathy because of edema in liver and small bowel
Forbes disease	glycogen storage disease type III; see Cori's disease
Forbes-Albright syndrome	galactorrhea-amenorrhea caused by a pituitary adenoma
Forchheimer spots	in 20% of rubella, small, red macules on the soft palate that may later expand and coalesce; not pathognomonic since they may also appear in scarlet fever, roseola, mono
Fordyce's spots	ectopic sebaceous glands seen in healthy mouth; may be confused with Koplik's spots which has an erythematous halo by comparison
Forrester classification	in myocardial infarction, I, PWP<18 and CI>2.2 L/min/m2, 2% mort; II, PWP>18 and CI>2.2, 10% mort; III, CI<2.2 and PWP<18, 12% mort; IV, PWP>18 and CI<2.2, 54% mort
Forscheimer spots	in rubella, punctate soft palate macules
Foster Kennedy syndrome	optic atrophy, contralateral papilledema, and anosmia; may be associated with olfactory groove meningioma
Fothergill's sign	in rectus sheath hematomas, tender mass that does not cross the midline and remains palpable when patient tenses the rectus muscle (bluish discoloration not usually seen until 3 or 4 days)
Fournier's gangrene	necrotizing infection of the perineal and genital fascia
fourth disease	Flatow-Dukes's disease, aka *Staph* scarlet fever, caused by *Staph aureus* epidermolytic toxin, described in 1900
Fouville's syndrome	dorsal pontine injury giving rise to lateral gaze palsy, ipsilateral facial palsy, contralateral hemiparesis
Fowler's syndrome	idiopathic voiding dysfunction and urinary retention
Fox-Fordyce disease	chronic, pruritic, papular eruption occurring in areas with apocrine glands, affecting mainly women
Fox's sign	in hemorrhagic pancreatitis, ecchymosis of the inguinal ligament due to blood tracking from the retroperitoneum and collecting at the inguinal ligament
Frank's sign	earlobe crease, associated with CAD (N Engl J Med. 1973;289:327-8)
Franklin's disease	gamma heavy chain disease, characterized by LAD, fever, anemia, malaise, HSM, and weakness, most distinctive symptom palatal edema
Frasier syndrome	female external genitalia in 46 XY patients, late renal failure, streak gonads, and high risk of gonadoblastoma; associated with mutation in WT1 gene
Freiberg disease	osteochondrosis of second metatarsal head; associated with avascular necrosis of metatarsal head; most patients are female
Frey syndrome	gustatory sweating or auriculotemporal syndrome seen in the context of parotid surgery; ipsilateral forehead becoming drenched with sweat; attributed to aberrant neuronal regeneration resulting in parasympathetic cholinergic innervation of cutaneous sympathetic receptors
Friedman curve	graph of dilataion of cervix v. fetal descent during active labor
Friedreich's ataxia	a spinocerebellar degeneration, autosomal recessive form manifesting at 11 years, less common autosomal dominant form 20 years; initial symptoms: gait ataxia, hand clumsiness, dysarthria, DTRs absent (extensor plantar present), joint position and vibratory sense impaired, sometimes loss of pain and temperature, paralysis over course of 20 years, high incidence of diabetes and hypertrophic cardiomyopathy (dilated less common) and arrhythmias
Friedreich's foot	seen in Friedreich's ataxia, pes cavus with hammer toe
Friedreich's sign	exaggerated y descent in patients with increased venous pressure, associated with an S3; also associated with constrictive pericarditis
Froin's syndrome	spinal block from tumor or inflammatory conditions, etc. causing increased protein in CSF, resulting in xanthochromia and increased coagulability in CSF
Froment's sign	diagnosis of ulnar nerve lesion; caused by flexor pollicus longus (median nerve) which comes into action when the patient attempts to grip a flat object between the thumb and the hand, and causes flexion at the interphalangeal joint
Fukuyama congenital muscular dystrophy	congenital muscular dystrophy in combination with cortical dysgenesis (micropolygyria); common autosomal recessive disorder in Japan
Gaisböck's syndrome	stress polycythemia, combination of modest elevation in hematocrit and normal red cell mass; unknown etiology, seen in hard-driving middle-aged males, usually smokers, who in addition tend to be overweight and hypertensive

Galeazzi fracture	fracture of the radius at the junction of the middle and distal thirds accompanied by disruption of the distal radioulnar joint
Galen, great vein of	great cerebral vein
Gallavardin dissociation	in aortic stenosis, high-velocity jet within the aortic root results in radiation of murmur upward to 2nd right intercostal space (in older patients, becomes harsh, noisy, and impure), whereas the murmur over apex is pure and often musical, mimicking mitral regurgitation.
Galloway-Mowat syndrome	autosomal recessive disorder characterized by early onset nephrotic syndrome, microcepahly, and hiatal hernia
gamekeeper's thumb	injury to ulnar collateral ligament of the thumb
Gandy-Gamna nodules	in congestive splenomegaly, organization of focal hemorrhages giving rise to foci of fibrosis containing deposits of iron and calcium salts encrusted on connective tissue and elastic fibers
Garcin syndrome	extensive unilateral cranial palsies associated with malignancy in nasopharynx or skull base
Gardner's syndrome	familial adenomatous polyposis (now realized all patients with FAP have extraintestinal manifestations), exhibiting intestinal polyps identical to those in FAP combined with 1. sebaceous cysts, 2. osteomas (particularly of the mandible, skull, and long bones), 3. desmoid tumors; mutation in adenomatous polyposis coli (APC) gene in 5q
Gardner-Diamond syndrome	autoerythrocyte sensitization; painful ecchymoses at site of trauma followed by progressive edema and erythema; associated with psychiatric disorders (also known as psychogenic purpura); can be demonstrated by intradermal injection of patient's own RBC stroma; described in 1955
Gardos channel	potassium-selective channel controlled by cytosolic calcium concentrations, one of two channels involved in red cell shrinkage playing a role in sickling; local tissue acidosis and deoxygenation-induced influx of calcium activate the Gardos channel resulting in shrinkage; clotrimazole potent blocker of this channel
Garland's triad	in sarcoidosis, triad of bilateral hilar lymphadenopathy, right paratracheal lymphadenopathy on chest radiograph
Garré, sclerosing osteomyelitis of	typically develops in the jaw, associated with extensive new bone formation that obscures much of the underlying osseous structure
gastrinoma triangle	a triangle where more than 90% of extrapancreatic gastrinomas are located bordered by 1. 3rd portion of duodenum, 2. Cystic duct, 3. Pancreatic neck
Gaucher's disease	autosomal recessive sphingolipidosis (sulfatidose), mutations in glucocerebrosidase gene on 1q21 (also known as acid beta-glucosidase), enzyme cleaves glucose residue from ceramide, frequently fatal; type I, chronic non-neuronopathic form, splenic and skeletal involvement; can be treated with infusions of macrophage-targeted human placental glucoerebrosidase, alglucerase (Ceredase), use described in 1991
Gelineau's disease	narcolepsy
Geraldi, fossa of	name of the fossa between the testicle and epididymus
Gerbode defect	ventricular septal defect communicating directly between the left ventricle and right atrium
Gerhardt's sign	in aortic regurgitation, pulsation of the spleen in the presence of splenomegaly; see also Sailer's sign
German measles	rubella
Gerota's fascia	fascia surrounding the kidney
Gerstmann syndrome	finger agnosia, agraphia, right-left disorientation, and dyscalculia
Gerstmann-Straussler-Scheinker syndrome	slowly progressive cerebellar ataxia, beginning in 50-60s; due to mutation in prion gene
Geschwind syndrome	personality syndrome in temporal lobe epilepsy characterized by viscosity, circumstantiality, hypergraphia (writing and drawing), and hyperreligiosity (*Arch Gen Psychiatry* 1975;32:1580)
Ghon lesion	primary area of tuberculosis infection
Gianotti-Crosti syndrome	reactive exanthem, first described in association with hepatitis B infection (papular acrodermatitis of childhood, papular eruption on face and limbs); also associated with EBV, CMV infection
Gibbs-Donnan effect	effect of charged particles on one side of a membrane on the distribution of other charged particles, when the former cannot diffuse through the membrane but the latter can.
Gilbert's syndrome	with the exception of hemolytic anemias, most common cause of mild unconjugated hyperbilirubinemia from mild decrease glucoronyltransferase activity, responds to phenobarbital, affects up to 7% of population
Gitelman's syndrome	variant of Bartter's syndrome where patients have hypomagnesemia and hypocalciuria due to mutations in thiazide-sensitive sodium-chloride transporter
Glanzmann's thrombasthenia	inherited disorder of platelet function where Gp IIb/IIIa receptor for fibrinogen missing
Glauber's salt	sodium sulfate, laxative

Gleason score prostate cancers stratified into 5 grades on basis of glandular patterns and degree of differentiation; grade 1 most differentiated and grade 5 no glandular differentiation; primary score and secondary score (for second most common histology) added together for score; therefore most differentiated tumor score of 2

Gleich syndrome episodic angioedema with eosinophilia characterzed by recurrent episodes of angioedema, urticaria, pruritus, fever, weight gain, elevated IgM, leukocytosis, and marked eosinophilia; blood eosinophilia parallels disease activity (*N Engl J Med* 1984;310:1621); no end-organ involvement or cardiac damage unlike other eosinophilc syndromes.

Glenn shunt In treatment of hypoplastic left heart syndrome. Unidirectional: SVC to right pulmonary artery anastomosis with ligation of the proximal RPA and cardiac end of the SVC. Bidirectional: SVC to right pulmonary artery anastomosis with flow to both the RPA and LPA, part of Norwood procedure

Glisson's capsule liver capsule

glycogen storage diseases type I, von Gierke's disease; type II, Pompe's disease; type III, Cori's disease; type V, McAdle's syndrome

Goetz sign in PDA, jet of unopacified blood from aorta into opacified blood of pulmonary artery

Goldblatt kidney atrophic kidney from vasoconstriction of renal artery; results in hypertension

Goldenhar syndrome syndrome of oculoauriculovertebral dysplasia characterized by unilateral deformity of the external ear and small ipsilateral half of the face with epibulbar lipodermoid and vertebral anomalies; autosomal dominant

Goldie-Coldman hypothesis malignant cells likely to acquire spontaneous resistance to cytotoxic drugs as they progressively grow and divide, even without any exposure to those drugs

Golgi apparatus membranous cell structure that processes proteins synthesized in the endoplasmic reticulum

Gonda's maneuver extensor plant response by flicking the little toe suggesting upper motor neuron defect

Good's syndrome immunodeficiency, hypogammaglobulinemia associated with thymoma (mainly thymoma of spindle cell type); associated with recurrent pulmonary infections; described by Good in 1954

Goodell's sign softening of cervix associated with pregnancy that occurs at around 8th week

Goodpasture's syndrome glomerulonephritis characterized by linear deposits of antibody along the glomerular basement membrane, antibodies interact with alveolar wall, leads to pulmonary hemorrhage and pulmonary fibrosis; Goodpasture antigen, resides in the noncollagenous portion of the alpha3 chain of collagen type IV; high prevalence of DRW15/DQW6, also see iron-deficiency anemia

Goodsall's rule anal fistulae course in a straight path anteriorly and take a curved path posteriorly

Gordon's maneuver extensor plant response by squeezing the calf muscle suggesting upper motor neuron defect

Gordon's syndrome type 2 pseudohypoaldosteronism associated with volume expansion, hypertension, and otherwise normal renal function; felt be to be due to enhanced distal chloride reabsorption

Gorham-Stout disease vanishing or disappearing bone disease; IL-6 has pathogenetic role

Gorlin's syndrome increased incidence of basal cell carcinoma, medulloblastoma, and rhabdomyosarcoma; attributed to heterozygous mutation in Patched, a negatively acting component of the Hedgehog receptor

Gorlin-Goltz syndrome focal dermal hypoplasia characterzied by widespread lesions of dermal hypoplsia with herniation of adipose tissue, streaks of pigmentary disturbance following lines of Blaschko, and severe absence deformities of bone, mental retardation, defects of optic nerve

Gottron's papules in dermatomyositis, scaling erythematous eruption or dark red patches over the knuckles, elbows, knees; may mimic psoriasis

Gower's maneuver Duchenne's muscular dystrophy, patient using hands to help himself get up

Graafian follicle small fluid-filled sac in ovary containing maturing egg; described by Regnier de Graaf (1641-1673), Dutch physician

Gradenigo's syndrome thrombosis of inferior petrosal sinus producing ipsilateral facial pain, lateral rectus muscle weakness

Grafenberg spot erogenous area in anterior vaginal wall

Graham patch piece of omentum used for repairing perforations

Graham Steell murmur early diastolic murmur of pulmonic regurgitation secondary to pulmonary hypertension, best heard in left 2nd interspace, may be indistinguishable from murmur of aortic regurgation; occurs in 12% of patients with tricuspid regurgiation; described by Graham Steell

Graham-Little syndrome end-stage lichen planus of the scalp resulting in scarring alopecia of the scalp

Graves's disease hyperthyroidism with diffuse goiter, ophthalmopathy, dermopathy from thyroid-stimulating Ig

Grawitz tumor renal cell adenocarcinoma

Gregg effect augmentation of left ventricular systolic performance with increase in coronary flow and perfusion pressure

Greig cephalopolydactyly syndrome rare autosomal dominant developmental disorder characterized by craniofacial abnormalities and post-axial and pre-axial polydactyly as well as syndactyly of hands and feet from mutation in GLI3

Grey Turner's sign	local areas of discoloration about the umbilicus and particularly in the region of the loins, in acute hemorrhagic pancreatitis (1-2%) and other causes of retroperitoneal hemorrhage
Griffith's point	watershed area in splenic flexure; see also Sudeck's point
Griffith's sign	in thyrotoxicosis, lag of the lower lids during elevation of the globes
Griscelli syndrome	primary immunodeficiency syndrome associated with partial "albinism"; presents similarly to Chédiak-Higashi syndrome but without the giant intracytoplasmic inclusions; defect in exocytosis of cytolytic granules; due to mutation in *RAB27A*, a member of the RabGTPase family or mutation in myosin-Va
Grover's disease	transient acantholytic dermatosis; most common in males over 40, sudden onset of pruritus across trunk, neck, and proximal limbs; a polymorphic, pruritic, papulovesicular dermatosis characterized histologically by acantholysis
Guam disease	Guam amyotrophic lateral sclerosis-parkinsonism-dementia linked to a plant excitant neurotoxin found in cycad plant eaten by Chamorro people, toxin believed to be excitotoxin beta-N-methylamino-L-alanine, a low-potency convulsant *Science* 1987;237:517-22
Guillain-Barré syndrome	acute idiopathic polyneuropathy following minor infective illnesses, inoculations, or surgical procedures (suggested association with *C. jejuni*) resulting in immunologically-mediated demyelination and leading to progressive weakness
Gull's disease	myxedema
Gunn's sign	tapering of veins on either side of AV crossing in hypertensive retinopathy (grade 3); see also Salus's sign and Bonnet's sign
Gunther's disease	AR congenital erythropoietic porphyria from decreased URO synthase activity, hemolytic anemia, cutaneous lesions
Guthrie test	for diagnosis of PKU, a bacterial assay for phenylalanine
Guyon's tunnel	ulnar tunnel
Haab's striae	lines in Descemet's membrane; associated with congenital glaucoma
Haff disease	unexplained rhabdomyolysis in people who have eaten fish in the 24 hours before onset of symptoms; first described in the summer and fall of 1924 when physicians near the Haff shores along the Baltic coast recognized an outbreak; attributed to an unidentified toxin
Hageman factor	factor XII, named after patient John Hageman. Interestingly, patient died of a pulmonary embolism after traumatic left hip fracture.
Hailey-Hailey disease	familial benign pemphigus; described by Hailey brothers in 1939
Hakim-Adams syndrome	normal pressure hydrocephalus
Haldane effect	deoxygenated hemoglobin having a greater affinity for CO2 than oxygenated hemoglobin
Hallermann-Streiff syndrome	autosomal recessive syndrome with bird-like facies with hypoplastic mandible and beaked nose, proportionate dwarfism, hypotrichosis, microphthalmia, and congenital cataract
Hallervorden-Spatz disease	autosomal recessive juvenile-onset generalized neuroaxonal dystrophy characterized by progressive movement alterations such as dystonia, tardive dyskinesia, rigidity, choreoathetosis, pyramidal signs, mental retardation, and iron accumulation in the brain; associated with mutation in pantothenate kinase 2; Hallervorden, Nazi pathologist involved in Aktion T-4 adult euthanasia program
Ham's test	for diagnosing paroxysmal nocturnal hemoglobinuria, uses increased sensitivity of PNH-affected RBCs to lysis by complement; introduced in late 1930s
Hamburger shift	in red blood cells, the conversion of carbon dioxide ultimately to bicarbonate, which diffuses out in exchange for chloride ions diffusing in
Hamman's sign	mediastinal crunch is a series of precordial crackles synchronous with the heart beat, not with respiration. Best heard in the left lateral position, it is due to mediastinal emphysema (pseudomediastinum), seen with Boerhaave's syndrome
Hamman-Rich syndrome	acute interstitial pneumonia, a rare fulminant form of lung injury that presents acutely (days to weeks from onset of symptoms), most commonly occurs in previously healthy individual, characterized by a temporally uniform lesion which reflects an episode of acute lung injury at a single point in time (different from usual interstitial pneumonia where lesions are different age); path shows diffuse alveolar damage (a nonspecific reaction pattern)
Hampton's hump	in pulmonary embolism/infarction, a wedge-shaped infiltrate that abuts the pleura; often associated with a small pleura effusion that is usually exudative and may be hemorrhagic; not specific
Hampton's line	radiolucent collar of granulation tissue across the base of an ulcer
Hand-Schüller-Christian triad	in multifocal Langerhans' cell histiocytosis, triad of calvarial bone defects, diabetes insipidus, and exophthalmos
hangman's fracture	fracture of pars interarticularis of C2, hyperextension injury
Hannington-Kiff sign	ipsilateral loss of the adductor reflex with preservation of patellar reflex in strangulated obturator hernia
Hansel's stain	special stain used to detect eosinophiluria on the urine sediment
Hansen's disease	leprosy

Hardy-Weinberg equilibrium	p^2 + 2pq + q^2 for describing the frequency of these genotypes; sexual reproduction doesn't cause a constant reduction in genetic variation in each generation but remains constant
harlequin syndrome	localized autonomic syndrome of heat or exercised-induced flushing and sweating limited to one side of the face with impairment of sweating and flushing on the contralateral side; possible link with Holmes-Adie syndrome
Harrison's groove	during active rickets, the protuberant rachitic abdomen pushes the plastic lower ribs outward on a fulcrum formed by the costal attachments of the diaphragm; the line of bending forms a groove or sulcus in the rib cage
Hartmann's procedure	proximal colostomy followed by stapled-off colon or rectum that is left in the peritoneal cavity
Hartnup disease	an autosomal recessive disorder in which there is a reduction of small-intestinal and renal transport of certain neutral amino acids, including (Trp, a precursor of niacin), causing elevated amino acid excretion in urine and feces
Hashimoto's encephalopathy	neurologic complication of Hashimoto's thyroiditis, mainly in people euthyroid, associated with a vasculitic condition with stroke-like episodes and cognitive impairment and diffuse progressive picture with dementia, seizures, movement disorders, psychosis, and sometimes delirium
Hashimoto's thyroiditis	first described in 1920, goitrous chronic autoimmune thyroiditis; in areas with sufficient iodine, elevated TSH is often viewed as evidence of chronic autoimmune thyroiditis as well as antithyroid antibodies; antithyroglobulin antibodies in 60% of patients and antithryoid microsomal antibodies in 95%
Hashimoto-Pritzker disease	pure cutaneous form of Langerhans cell histiocytosis
Hassall's corpuscles	spherical or ovoid bodies found in the medulla of the thymus, composed of concentric arrays of epithelial cells around central area of degenerated cells. Numbers increase until puberty, then decreases.
Hatchcock's sign	upward pressure applied to the angle of the mandible (ramus) produces tenderness with mumps but no tenderness with adenitis
Haw River syndrome	variant of dentalorubral-pallidoluysian atrophy (DRPLA) (ataxia and rigidity accompanied by choreoathetosis, myoclonic epilepsy, and dementia) but without the myoclonic epilepsy; described in ancestors born in Haw River, North Carolina; caused by same caused by the same expanded CTG-B37 repeat as as in DRPLA
Hawkin's sign	in shoulder impingement, with the arm in a throwing position and flexed forward about 30 degress, forcibly internally rotate the humerus; pain suggests impingement of the supraspinatus tendon against the coracoacromial ligament
Hawthorne effect	the effect (usually positive or beneficial) of being under study, upon the persons being studied; their knowledge of the study often influences behavior [city in Illinois; site of the Western Electric plant]
Heberden's nodes	characteristic in women, but not in men, represent prominent osteophytes at the DIP joints in osteoarthritis (enlargements of tubercles at the articular extremities of the distal phalanges
Hecht-Beals syndrome	trismus-pseudocamptodactyly syndrome, inability to open moth fully and problems with wrist dorsiflexion, producing involuntary flexion contracture of distal and proximal interphalangeal joints
Heck's disease	oral focal epithelial hyperplasia associated with HPV 13, 32 infection
Heerfordt-Waldenström syndrome	sarcoidosis associated with fever, parotid enlargement, anterior uveitis, and facial nerve palsy
Hegar's sign	softening of the uterus at the junction between the cervix and the fundus during the first trimester of pregnancy
Heimlich maneuver	abdominal thrust maneuver for clearing airway (blocked by foreign object, e.g. food) described in 1974 by Henry Heimlich
Heineke-Mukulicz pyloroplasty	longitudinal incision through all layers of the pylorus, sewed closed in a transverse direction to make the pylorus nonfunctional (used after truncal vagotomy)
Heiner syndrome	cow milk allergy associated with pulmonary infiltrates, pulmonary hemosiderosis, anemia, recurrent pneumonia and failure to thrive.
Heinz bodies	seen in unstable hemoglobin and oxidant stress; precipitates of denatured hemoglobin on red blood cells; only visible when blood is supravital stained (crystal violet); not seen on routine blood smears
Heister, spiral valves of	found in the neck of the gallbladder, where tiny folds of mucosal epithelium coalesce to form these valves, may assist in retaining bile between meals
HELLP syndrome	hemolysis, elevated liver function tests, and low platelets; microangiopathic process seen in third trimester of pregnancy; seen in 20% of preeclampsia
hemoglobin Bart's	four gamma globin chains, seen in hydrops fetalis (alpha thalassemia), very high oxygen affinity
hemoglobin Constant Spring	particularly common structural variant with alpha thalassemia in Asia, contains mutation which abolishes normal translation termination codon, so extra 31 residues read until another in-frame termination codon comees up
hemoglobin Gower	embryonic, primitive hemoglobin. Gower 1, zeta2 episilon2; Gower 2, alpha2 epsilon2
hemoglobin Lepore	no beta chain; delta chain by delta-beta hybrid
hemoglobin Portland	zeta2gamma2, primitive embryonic hemoglobin

HEMPAS	Hereditary Erythrocyte Multinuclearity and Positive Acidified Serum test, also known as congenital dyserythropoietic anemia type II. A congenital anemia characterized by multinucleated red cell precursors and hemolysis of red blood cells of the patient with autologous serum during the Ham's test
Henle, loop of	section of nephron between proximal and distal tubule located in the medulla and involved in reabsorbing 25-35% of filtered NaCl and reaborbing NaCl in excess of water
Henle's layer	the outer layer of cells of the inner root sheath of a hair follicle, lying between the outer root sheath and Huxley's layer
Henoch-Schönlein purpura	systemic hypersensitivity disease of unknown cause characterized by purpuric rash, colicky abdominal pain (presumably due to focal hemorrhages into the GI tract), polyarthralgia, and acute glomerulonephritis; may result from deposition of circulating immune complexes within stuff; hypersensitivity purpura, etiology group A streptococci
Hering, canals of	at the fringes of the portal tract, from the joining of bile canaliculi
Hering's nerve	carotid sinus nerve, CN IX, carries information to the vasomotor center in the brainstem
Hering-Breuer reflex	stretch receptor (in smooth muscle of airways) reflex, responsible for apnea, i.e. decreased breathing frequency, as a result of lung inflation
Herlitz syndrome	epidermolysis bullosa lethalis; mutation that prevents the folding of laminin 5 Gillis Herlitz, Swedish pediatrician, born 1902
Hermansky-Pudlak syndrome	autosomal recessive syndrome from defect in AP-3 adaptor complex beta3A subunit or HPS1; characterized by idiopathic pulmonary fibrosis, oculocutaneous albinism, a platelet storage pool deficiency, and ceroid lipofuscinosis from defects of multiple cytoplasmic organelles: melanosomes, platelet dense granules and lysosomes.
Herring bodies	neurosecretory granules consisting of either vasopressin or oxytocin in neurohypophysis
Hers disease	type VI glycogen storage disease, deficiency in liver phosphorylase; presents as hepatomegaly and growth retardation; hypoglycemic, hyperlipidemia, and hyperketosis usually mild if present; hepatomegaly and growth retardation improve with age and usually disppear at puberty
Heschl's gyrus	primary auditory cerebral cortex, transverse temporal gyrus
Hess test	see Rumpel-Leede sign; noticed phenomenon while treating children with scurvy.
Hesselbach's triangle	where direct inguinal hernias occur, bound by the inguinal ligament, inferior epigastric a., and rectus abdominus muscle
Heyde's syndrome	association between bleeding from gastrointestinal angiodysplasia and severe aortic stenosis which may resolve with aortic valve replacement; may also be explained by acquired type IIA von Willebrand's syndrome; first described by Heyde in *N Engl J Med* 1958;259:196.
Hickey-Hare test	in evaluating hypernatremia, 5% NaCl infused at a rate of 0.05 mL/kg/min followed by measurements of urine volume and Uosm
Hill's sign	in aortic regurgitation, refers to popliteal cuff systolic pressure exceeding brachial cuff pressure by more than 60 mm Hg
Hill-Sachs lesion	posterolateral humeral head indentation fracture from anterior shoulder dislocation; from humeral head impinging against glenoid of shoulder joint
Hinton test	test for diagnosing syphilis using a precipitin of glycerol, cholesterol, and beef heart extract; developed in 1927
Hippocratic fingers	clubbing
Hippocratica, facies	the mask of death following peritonitis, i.e. as a result of vomiting and depressed circulation, the face becomes pinched and anxious, the cheeks hollow, and the eyes dim and beringed with dark circles
Hirano bodies	in Alzheimer's, intracytoplasmic proximal dendritic eosinophilic inclusions consisting of actin
Hirata disease	insulin autoimmune syndrome; hypoglycemia associated with insulin autoantibodies; third leading cause of spontaneous hypoglycemia in Japan; rare in Western countries
Hirschberg test	corneal light reflex test
Hirschprung's disease	congenital aganglionic megacolon; colonic dilatation proximal to an aganglionic, contracted distal colon and rectum; caused by gestational failure of neural crest cells to migrate to distal colon; AD form associated with mutations of the RET gene, AR form associated with mutations of the endothelin-B-receptor gene
Hitzig zones	in tabes dorsalis (syphilis), regions of reduced sensation found in central face, nipples, ulnar forearms, and peroneal legs, leading to painless ulcers and joint damage
Hodgkin's disease	lymphoma characterized by arising in a single node or chain of nodes, spreading in anatomically contiguous nodes, Reed-Sternberg cells
Hoffman's sign	involuntary flexion of the digits when tapping or striking over the plamar digital aspects of the 2nd, 3rd, and 4th digits in upper motor neuron defect
Hoffman's syndrome	in hypothyroidism, diffuse muscle hypertrophy, accompanied by stiffness, weakness, painful muscle cramps, associated with elevated muscle enzymes
Hoigne reaction	pseudoanaphylactic reaction, complication associated with intramuscular procaine penicillin, occurs in 1/1000 patients; characterized by tachycardia, elevated blood pressure, fear of imminent death, violent combativeness, unusual taste sensation, auditory or visual disturbances; rarely lasts longer than 30 min; felt to be due to inadvertent intracapillary infusion resulting in microembolization
Hollenhorst plaques	cholesterol emboli visible as small bright flecks lodged in arterial bifurcations in retina

Holme's heart	single ventricle with normally-related great arteries
Holmes tremor	symptomatic tremor caused by lesions in the brainstem, cerebellum, or thalamus; postural and/or action in nature and worsen during movement and markedly increase during goal-directed movements; affect predominantly proximal limbs, low frequency; previously labeled rubral tremor, midbrain tremor, thalamic tremor, myorhythmia, and Benedikt syndrome
Holmes-Adie syndrome	frequently affects young women, large, often irregular pupils, unilateral at onset as well as segmental palsy and segmental spontaneous movement of the iris, delayed constriction in response to near vision, delayed redilation after near vision, impaired accommodation, absent light reflex, and absent deep tendon reflexes; may be caused by degeneration of ciliary ganglion, followed by aberrant reinnervation of the pupilloconstrictor muscles
Holter monitor	continuous ambulatory EKG monitoring; first described by Montana physician Normal Holter in 1949 (*Science* 1961;134:1214-1220)
Holt-Oram syndrome	autosomal dominant syndrome, dysplasia of the upper limbs and atrial septal defect, reduction anomalies of the upper limbs, heart-hand syndrome from mutation in TBX5
Homans's sign	in deep venous thrombosis, pain and tenderness on compression of the calf muscles (by either squeezing the calf muscles or forced dorsiflexion of the foot)
Homer-Wright pseudorosettes	in neuroblastoma, tumor cells arranged about a central space filled with fibrillar extensions of the cells
honeymooner's palsy	radial nerve palsy from compression of nerve between middle third of the humerus between the brachoradialis and forearm extensor muscles
Hoover's sign	in hysterical weaknes, patient does not push down into your hand when you ask patient to lift weak extremity
Hoover's sign	a modification in the movement of the costal margins during respiration, caused by a flattening of the diaphragm; suggestive of empyema or other intrathoracic condition causing a change in the contour of the diaphragm
Hopkins syndrome	poliomyelitis-like illness associated with acute asthma in childhood
Horner's syndrome	enophthalmos, ptosis, miosis, and anhidrosis, unilateral; if present since infancy, the ipsilateral iris is lighter and blue (heterochromia iridis); topical 4% cocaine will dilate normal pupil but not a desympathectomized pupil; associated with Pancoast tumor, due to involvement of paravertebral sympathetic chain and the inferior cervical ganglion
Horton's cephalalgia	cluster headache; also known as histaminic cephalalgia
Horton's disease	temporal arteritis
Hounsfield unit	scale named after inventor of CT, Sir Godfrey N. Hounsfield (d. Aug 12, 2004), water 0 H, -1024 for air to 3000-4000 H for bone, relative scale
Houston, valve of	rectal valves: superior, middle, and inferior
Howell-Evans syndrome	inherited tylosis (keratoderma of palms and soles), strongly associated with esophageal squamous cell carcinoma
Howell-Jolly body	in asplenia or malfunctional spleen, nuclear remnants on red blood cells as small, round, darkly-stained nuclear fragments; no special stains necessary; larger than Pappenheimer bodies
Howship's lacunae	resorption pits on bone formed by osteoclasts
Howship-Romberg sign	pain along the inner aspect of the thigh; seen with an obturator hernia due to nerve compression
Hughes syndrome	antiphospholipid antibody syndrome; described in 1983 by Graham Hughes (Clin Exp Dermatol 1984;9:535)
Hughes-Stovin syndrome	multiple pulmonary artery aneurysms with peripheral venous thrombosis
Hunner's ulcer	in 10% of interstitial cystitis, lesions that involve all layers of the bladder wall and appear as brownish-red patches on the bladder mucosa
Hunter's ligament	round ligament of uterus
Hunter's syndrome	mucopolysaccharidosis (II), iduronate sulfatase deficiency, X-linked, affecting degradation of dermatan sulfate and heparan sulfate, resulting in physical deformity and mental retardation, no corneal clouding
Huntington's disease	autosomal dominant from expanded CAG trinucleotide repeat 4p16.3 associated with chorea initially as restlessness, psychiatric symptoms, and dementia; Westphal variant, atypical variant where there is progressive rigidity and akinesia with little or no chorea
Hurler's syndrome	AR mucopolysaccharidosis (I H) caused by deficiency of alpha-L-iduronidase, with consequent accumulations of the mucopolysaccharides heparan sulfate and dermatan sulfate in the heart, brain, liver, and other organs; progressive deterioration, hepatosplenomegaly, dwarfism, gargoyle-like facies, stubby fingers, corneal clouding, progressive mental retardation, and death by age 10
Hurler-Scheie syndrome	mucopolysaccharidosis I caused by deficiency of alpha-L-iduronidase (severe form is Hurler's syndrome), have same problems as Hurler's syndrome but progression is lower, little or no mental retardation, and they die in teens or 20s
Hurst's disease	acute hemorrhagic leukoencephalitis

Hürthle cells	in Hashimoto's thyroiditis, deeply stained colloid or clusters of these oncocytes having an abundant, brightly eosinophilic granular cytoplasm, thought to represent a degenerated state of the follicular epithelium
Hutchinson freckle	lentigo maligna, a nonfamilial precursor to lentigo maligna melanoma
Hutchinson pupil	blown third pupil in uncal herniation
Hutchinson's sign	pigment in the paronychial area suggesting melanoma
Hutchinson's sign	in herpes zoster ophthalmicus, vesicular rash at nasal tip indicating involvement of the external nasal branch of CN V associated with increased incidence of ocular zoster
Hutchinson's teeth	smaller and more widely spaced than normal and are notched on their biting surfaces; sign of congenital syphilis
Hutchinson's triad	congenital syphilis manifesting late at around 2 years old with triad of interstitial keratitis, notched incisors, and eighth nerve deafness
Hutchinson-Guilford syndrome	aka progeria a condition in which the normal development of the first year is followed by gross retardation of growth, with a senile appearance characterized by dry wrinkled skin, total alopecia, and bird-like facies; genetics unclear
Huxley's layer	a layer of the inner root sheath of a hair follicle, lying between Henle's layer and the inner sheath cuticle
I-cell disease	lysosomal disorder, from deficiency of ability to phosphorylate mannose, causing an incorrect targeting of potential lysosomal enzymes to extracellular sites instead of lysosomes, characterized by skeletal abnromalities, restricted joint movement, coarse facial features, severe pscyhomotor impairment, death by 8 y.o.
Imerslund-Grasbeck syndrome	juvenile megaloblastic anemia, an autosomal recessive condition involving mutation in cubulin, receptor that binds IF-B12 complex; also associated with proteinuria
Irish's node	left axillary adenopathy associated with metastatic disease, e.g. gastric cancer
Isaacs's syndrome	neuromyotonia; continuous muscle stiffness, rippling muscle movements (myokymia), delayed relaxation following muscle contraction believed to be due to autoantibodies to presynaptic potassium channels
Ishihara plates	pseudoisochromatic plates (plates with color dots for numbers) for testing color vision
Ishikawa's sign	in cystic adventitial disease of the popliteal artery, disappearance of foot pulses on knee flexion or after exercise in patients
itai-itai disease	bone disease from cadmium toxicity, characterized by multiple fractures, mixed pattern of osteoporosis and osteomalacia, and renal damage. Means "ouch-ouch" in Japanease, cadmium toxicity noted in exposed individuals living in Jinzu river basin in Japan who used river water contaminated with cadmium for irrigating rice fields
Ito cells	vitamin A fat-storage cells, of mesenchymal origin found in the space of Disse; during development of cirrhosis, they become activated, transform into fibroblast-like cells
Ivor Lewis procedure	esophagectomy through right thoracotomy and intrathoracic esophagogastric anastomosis; described in 1946 by Ivor Lewis
Jaccoud's arthritis	in SLE, ulnar deviation of the fingers, swan neck deformities, and subluxations, initially reversible since not joint problem per se but can become fixed over time
jacksonian march	in simple partial seizures, clonic movements of a single muscle group spreading to involve contiguous regions of the motor cortex
Jackson-Weiss syndrome	craniosynostoses as well as limb defects, mutations if FGFR2, broad great toes
Jamaican vomiting sickness	poisoning from hypoglycin A in unripe ackee fruit; hypoglycin A is potent hypoglycemic agent that decreases rate of fatty-acid beta oxidation probably by inhibition of acyl dehydrogenase flavin-dependent oxidation; causes liver damage indistinguishable from Reye's syndrome
James fibers	tracts of atrial tissue running from the atria and inserting into the low AV node; functional significance not well established, controversial if associated with abnormal AV conduction
Janeway lesions	in infective endocarditis; nonpainful, small, erythematous or hemorrhagic macules or nodules of palms or soles; more common in acute bacterial endocarditis but occur in subacute bacterial endocarditis
Jansen metaphyseal chondrodysplasia	an autosomal dominant form of dwarfism resulting from an activating mutation in the PTHrP receptor, premature ossification from acceleration of the transition from proliferative to hypertrophic chondrocytes
Jansky-Bielschowsky disease	late infantile neuronal ceroid lipofuscinosis, CLN2 disorder generally characterized by visual loss, epilepsy, and psychomotor deterioration
Jarisch-Herxheimer reaction	sudden fevers, rigors, and persistent hypotension following antimicrobial treatment of louse-borne relapsing fever (spirochete *Borrelia recurrentis* infection) or syphilis; treatment with anti-TNF-alpha Fab before penicillin suppresses this reaction (NEJM 335:311)
Jatene's arterial switch	for treating transposition of great vessels, pulmonary artery and aorta are transected above valves and switched, and coronary arteries are moved from old aortic root to new aorta (former pulmonary root)
Jefferson fracture	fracture of C1 at more than one site; rarely associated with neurological deficits because of size of spinal canal at this level
Jendrassik maneuver	for increasing sensitivity of deep tendon reflexes, have patient clasp hands together or grab wrists tightly while checking reflexes
Jerusalem syndrome	psychosis affecting visitors to Jerusalem whereby afflicted develop psychotic religious delusions; affects a handful of visitors each year
Jervell-Lange-Nielsen	long QT syndrome (autosomal recessive) associated with congenital deafness

syndrome	
Job's syndrome	immune deficiency where neutrophils fail to respond to chemotactic stimuli, associated with high levels of IgE; patients susceptible to cold staphylococcal abscesses
Jod-Basedow phenomenon	thyroid hyperfunction induced by excess iodine ingestion in patients with various thyroid disorders; *Jod* German for iodine; K. A. Von Basedow
Joffroy sign	in thyrotoxicosis, absence of forehead wrinkling with upward gaze, the head being tilted down; disorder of the arithmetic faculty in the early stages of organic brain disease
John Thomas sign	positive sign when penis inclined to side of the disorder radiographically (e.g., fractured hip); 70% sens 67% specific for JT sign in hip fracture, *Med J. Aust.* 1998;169:670
Johnson-McMillin syndrome	autosomal dominant neuroectodermal syndrome characterized by anosmia, hypogonadotropic hypogonadism associated with conductive deafness, alopecia, congenital heart defects
Jolly test	in myasthenia gravis, a sequence of repetitive nerve stimulation (RNS) studies specifically designed to look for neuromuscular junction disease. A positive test is a >10% decremental response with 3 Hz repetitive stimulation, 50-80% sensitive. After Friedrich Jolly, German neurologist, 1844-1904
Jones criteria	for diagnosing rheumatic fever, major criteria: carditis, polyarthritis, chorea, erythema marginatum, and subcutaneous nodules. minor criteria: arthralgia, fever, elevated ESR, CRP, prolonged PR. in setting of antecedent GAS infection, two major or one major and 2 minor suggests RF. throat cultures negative by the time rheumatic fever appears
Jones's fracture	fracture at the base of the fifth metatarsal diaphysis
Jumping Frenchmen of Maine syndrome	unusually extreme startle reaction that occurs in selected populations with reactions including echolalia, echopraxia; first described in French Canadian lumberjacks in the Moosehead Lake Region of Maine in the late 19th century; also described as *latah* in Malaysia
kabuki syndrome	congenital mental retardation syndrome; postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids (reminiscent of the make-up of actors of Kabuki, a Japanese traditional theatrical form), a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy; also known as Niikawa-Kuroki syndrome
Kahler's disease	multiple myeloma
Kallman's syndrome	anosmia; hypogonadotropic hypogonadism stemming from failure of LHRH-expressing neurons to migrate, etc.
Kanavel's sign	four signs of tenosynovitis: 1. affected finger held in slight flexion; 2. pain over volar aspect of affected finger tendon upon palpation; 3. swelling of affected finger; 4. pain on passive extension of affected finger
Kandinsky-Clerambault's syndrome	alienation from or loss of one's own mental processes which are attributed to somebody else, combined with delusions of external influences, such as stealing or insertion of thoughts, described independently by Russian psychiatrist Victor Khrisanfovich Kandinsky (1849–1889) and French psychiatrist Gaëtan Gatian de Clerambault (1872–1934)
Kaplan-Meier curve	curve that estimates the probability of survival of a defined group at a designated time interval (conditional probability) based on a non-parametric survival function
Kaposi's sarcoma	low-grade vascular tumor associated with HHV-8, four forms, classic (older men of Mediterranean or Jewish extraction), African, organ transplant-associated, and AIDS; differential for skin lesions includes bacillary angiomatosis from Bartonella
Kartagener's syndrome	a triad of sinusitis, bronchiectasis, and situs inversus; also associated with ciliary dysfunction; Manes Kartagener, Swiss physician, 1897-1975
Kasabach-Merritt syndrome	capillary hemangioma associated with thrombocytopenic purpura and extensive and progressively enlarging vascular malformations which may involve large portions of their extremities; bleeding commonly develops in the first year of life, secondary to chronic DIC triggered by stagnant blood flow through the tortuous abnormal vessels; anemia caused by red cell damage as blood passes through deformed vessels of the tumor
Kasai procedure	for treating biliary atresia, hepatoportoenterostomy, biliary drainage created from small intestine
Kashin-Beck disease	deforming arthritis associated with selenium deficiency, identified in an area of China and Tibet where soil is extremely low in selenium; iodine deficiency risk factor
Katayama fever	acute schistosomiasis, after Katayama valley in Japan where S. japonicum endemic; seen in travelers but not in native people because of exposure in utero; symptoms of prostration, fever, sweats, myalgia, and diarrhea
Kawasaki's disease	acute febrile illness of infants and children, characterized by cutaneous and mucosal erythema and edema with subsequent desquamation, cervical lymphadenitis, and complicated by coronary artery aneurysms (20%); associated with antiendothelial antibodies
Kayser-Fleischer rings	a greenish yellow pigmented ring encircling the cornea just within the corneoscleral margin, seen in hepatolenticular degeneration, due to copper deposited in Desçemet's membrane (posterior limiting layer of cornea); seen in Wilson's disease (with neurologic involvement) and other cholestatic hepatic diseases
Kearns-Sayre syndrome	a mitochondrial disease with progressive ophthalmoplegia, retinal pigmentation degeneration, heart block; caused by various mitochondrial DNA mutations

Kegel exercises	exercises for strengthening pelvic muscles in order to increase urethral closure mechanism in urinary incontinence
Kehr's sign	pain in the left shoulder associated with splenic rupture
Kell blood group system	red blood cell antigen, named after Mrs. Kelleher, pregnant woman in whom anti-K was discovered by Coombs in 1946; function of antigens in this system are unknown.
Kellgren's arthritis	severe form of primary osteoarthritis, erosive osteoarthritis, associated with severe osteoporosis of hands
Kelly's sign	visible peristalsis of the ureter in response to squeezing or retraction; used to identify the ureter during surgery
Kennedy syndrome	X-linked spinobulbar muscular atrophy; from CAG repeat expansion encoding androgen receptor; leads to distal limb amyotrophy, bulbar signs, fasiculations prominent, androgen insensitivity with gynecomastia, testicular atrophy, and oligospermia
Kent, bundle of	AV bypass accessory tract in Wolf-Parkinson-White syndrome that directly connects atrial and ventricular myocardium
Kerandel's hyperesthesia	in African trypanosomiasis, excruciating pain after minor soft-tissue injury, e.g. in palms and ulnar region
Kerckring's valves	plicae circulares (lining of small intestine)
Kerley A lines	in pulmonary edema, thin nonbranching lines several inches in length, radiating from hila, and not following the course of vessels or airways, attributed to thickening of connective tissue sheets which contain communicating perivenous and bronchoarterial lymphatics
Kerley B lines	in pulmonary edema, transverse lines 1-3 cm in length and 1-2 mm in width, arranged in a horizontal stepladder pattern, 0.5-1 cm apart along the lower lateral lung margins, extending to the pleura; lines visible in this area because pulmonary lobules are well-developed in this area and lines reflect thickening of, or fluid in, lymphatic vessels in interlobular septa; also seen in lymphangitic spread of malignancies.
Kerley C lines	in pulmonary edema, fine interlacing lines throughout the lung base producing a spider web appearance; controversial if unique lines v. crossing lines
Kernig sign	first flex patient's leg at both hip and knee, and then straighten knee; positive sign is pain and increased resistance to extending knee, suggesting meningeal irritation
Kernohan notch	focal impression against the cerebral peduncle, pressure against notch in uncal herniation
Kerr incision	low transverse uterine incision for caesarian section
Keshan disease	endemic cardiomyopathy in China associated with selenium deficiency
Keutel syndrome	malformation syndrome characterized by multiple pulmonary stenoses, neural hearing loss, short terminal phalanges, and calcifications and/or ossification of the cartilage in ears, nose larynx, trachea, and ribs; due to mutation in gene encoding human matrix Gla protein
Kew Gardens spotted fever	Rickettsialpox due to Rickettsia akari, from Kew Gardens, New York
Kidd blood group system	named after Mrs. Kidd, pregnant woman in whom anti-Jka was discovered by Allen in 1951; antigens in this system are located on a red cell urea transporter
Kienböck disease	osteonecrosis of the carpal lunate that most commonly affects men between ages 20-40; sometimes history of trauma; may lead to end-stage arthritis of wrist with progression
Kiesselbach's plexus	vascular plexus on the anterior nasal septum, bleeding from this plexus associated with most common form of epistaxis
Kikuchi's disease	histiocytic necrotizing lymphadenitis, characterized by cervical lymphadenopathy with tenderness, fever, and night sweats; tends to occur in young women; histology can be confused with lymphoma
Killian-Jamieson area	region between oblique and transvere fibers of the cricopharyngeus muscle; potential area for muscular dehisence and Zencker's diverticulum
Killip class	described in 1967, in myocardial infarction, I, no heart failure, 0.5% mortality; II, S3 and/or basal lung crepitations, 2.2% mortality; III, acute pulmonary edema, 19.2% mortality; IV, cardiogenic shock, 61.3% mortality (mortality rates current)
Kimmelstiel-Wilson disease	intercapillary glomerulosclerosis from diabetes; lesion is PAS-positive material deposited at periphery of glomerular tufts
Kimura's disease	large subcutaneous masses on head or neck of Asian males associated with eosinophilia
Klatskin tumors	tumors arising from the part of the common bile duct between the cystic duct junction and the confluence of the right and left hepatic ducts at the liver hilus; notable for their slow growing behavior, marked sclerosing characteristics, and the infrequent occurrence of distal metastases
Kleihauer-Betke test	testing for the presence of fetal blood cells in maternal circ
Kleine-Levin syndrome	a rare form of periodic hypersomnia, occurring in males aged 10 to 25 years, characterized by periods of ravenous binge eating alternating with prolonged sleep (as long as 18 hours), along with behavioral disturbances, impaired thought processes, and hallucinations; acute illness or fatigue may precede an episode, which may occur as often as several times a year; thought to be a disorder of hypothalamically mediated satiety
Klein-Waardenburg	see Waardenburg syndrome type III

syndrome	
Klinefelter syndrome	XXY, first described in 1942 as constellation of enlarged breasts, sparse facial and body hair, small testes, long bone abnormality, and an inability to produce sperm; occurs in 1/500 to 1/1000 births
Klinger disease	Wegener's disease; first described by Heinz Klinger in 1931 as a medical student and then described by Friederic Wegener in 1936
Klippel-Feil syndrome	congenital fusion of two or more cervical vertebrae, producing a low posterior hairline, decreased movement, and a short neck that displaces the head anteriorly and inferiorly
Klippel-Trenaunay syndrome	congenital condition characterized by port-wine stain (cutaneous capillary hemangiomas), soft tissue and bony hypertrophy, and venous malformations and lymphatic abnormalities, usually limited to one limb
Klippel-Trenaunay-Weber syndrome	association of Klippel-Trenaunay syndrome with AV fistula; affects Casey Martin, golfer in disability case brought to U.S. Supreme Court 2001 against PGA
Klumpke's palsy	lower plexus palsy affecting C8-T1 nerve roots
Klüver-Bucy syndrome	1937 bilateral removal of temporal lobe (including amygdala and hippocampal formation) in monkeys, animals became tame, showed a flattening of emotions, exhibited remarkable oral tendencies (they put all manner of objects into their mouths), enormous increase in sexual behavior, including mounting of inappropriate objects and species, compulsive tendency to react to every object, failed to recognize familiar objects
Kniest dysplasia	autosomal dominant metatropic dwarfism associated with mutation in COL2A1
Kobberling's syndrome	familial partial lipodystrophy affecting adipose tissue in extremities and normal adipose tissue elsewhere
Koch's bacillus	tubercule bacillus
Koch's postulates	criteria for linking a specific microorganism to a disease 1. organism must be found in lesion of disease; 2. organism can be isolated in single colonies on a single medium; 3. inoculation of culture causes lesion in experimental animals; 4. organism can be recovered from lesions in these animals
Kocher's incision	right subcostal incision for open cholecystectomy
Kocher's maneuver	dissection of the duodenum from the right-sided peritoneal attachment to allow mobilization and visualization of the back of the duodenum and pancreas
Kocher's maneuver	for closed reduction of anterior shoulder dislocation, traction to the elbow with external rotation of the humerus and adduction of elbow towards chest; however not generally recommended because of association with neurovascular complications and proximal humerus fractures
Kocher's test	compression of lateral lobes of thyroid causing stridor; associated with thyroid carcinoma, goiter, or thyroiditis
Koebner's phenomenon	referring to physical trauma (rubbing or scratching) as a major factor in eliciting psoriasis lesions; also depigmented, sharply demarcated papules in vitiligo following minor trauma; may also be seen in lichen planus
Koeppe's nodules	projections into the pupil seen in sarcoidosis, tuberculosis, and other uveitides
Köhler's disease	aseptic necrosis or osteochondrosis of the navicular bone
Kohn, pores of	connections between alveoli
Kojewnikoff syndrome	epilepsia partialis continua (simple partial status epilepticus)
Kommerell diverticulum	anomaly where left subclavian artery arises from this diverticulum on the aortic arch as the 4th branch and passes behind the esophagus to the left arm; can be rarely associated with dysphagia
Koplik's spots	small, white spots (often on an reddened background) that occur on the inside of the cheeks early in the course of measles; considered to be pathognomonic exanthem in measles
Korotkoff sounds	sounds heard in between systolic and diastolic pressure; origin related to the spurt of blood passing under the cuff and meeting a static column of blood; I, first appearance of low-frequency tapping sounds; II, softer and longer sounds; III, crisper and louder sounds; IV, muffled and softer sounds; V, complete disappearance of sounds
Korsakoff's syndrome	profound memory loss; mammillary bodies as well as portions of medial thalamus believed to be destroyed from thiamine deficiency accompanying alcoholism
Kostmann syndrome	inherited neutropenia, responds to G-CSF, may be due to environmental insults in bone marrow, characterized typically by a granulopoeisis impairment at the promyelocyte stage
Kounis syndrome	allergic or hypersensitivity reaction associated with acute coronary syndrome
Krabbe's disease	AR sphingolipidosis (sulfatidose), from beta-galactosidase deficiency, resulting in increased galactocerebrosides, mental retardation, blindness, deafness, paralysis, convulsions, total absence of myelin, globoid bodies in white matter of brain, fatal in early life
Krause's end-bulbs	sensory receptors for cold
Krebs cycle	citric acid cycle or tricarboxylic acid cycle; central metabolic pathway that takes place in mitochondria involved in catabolism of carbohydrates and fat and generation of ATP; after Sir Krebs who proposed pathway in 1937
Krukenberg's tumor	ovrian metastasis of stomach cancer; characterized by signet-ring cancer cells; also seen in colon, breast, and other mucin-gland organs

Kübler-Ross dying stages	1. denial and isolation, 2. anger, 3. bargaining, 4. depression, 5. acceptance; described in 1969 by Elisabeth Kübler-Ross
Kufs disease	adult neuronal ceroid lipofuscinosis
Kugelberg-Welander disease	see under Wohlfart-Kugelberg-Welander disease
Kulchitsky's cells	neuroendocrine argentaffin cells present along the bronchial epithelium, particularly in the fetus and neonate; small cell carcinoma has granules similar to Kulchitsky's cells; origin of carcinoid tumors
Kupffer cells	phagocytic cells of the mononuclear phagocyte series found on the luminal surface of endothelial cells in hepatic sinusoids; they metabolize old RBCs, digest hemoglobin
Kussmaul's respiration	hyperpnea, associated with acidosis, especially DKA but also in uremia
Kussmaul's sign	jugular vein distension during inspiration (reversal of normal pattern of decreasing jugular venous pressure during inspiration); classically seen in constrictive pericarditis because of normal increase in venous return with increased abdominal pressure from diaphragmatic contraction leading to increase in right atrial pressure from non-complaint right ventricle, though most common contemporary cause is severe right-sided heart failure (JAMA 1996: 275:632); generally negative in cardiac tamponade
Kussmaul-Maier disease	old name for polyarteritis nodosa
Kveim antigen	saline suspension of human sarcoid tissue prepared from spleen of an individual with active sarcoidosis; used in Kveim-Sitzbach test
Kveim-Siltzbach test	an intradermal test for the detection of sarcoidosis, done by injecting Kveim antigen (see elsewhere) and examining skin biopsies after 3 and 6 weeks; positive test, a noncaseating granulomatous reaction; sensitivity 35-88%, specificity 75-99% Morton A. Kveim, Norwegian dermatologist, born 1892
Kyrle disease	chronic generalized dermatosis, formation of large papules with central keratin plugs; associated with diabetes mellitus and renal failure
La Crosse encephalitis	transmitted by tree hole mosquito, *Aedes trisereiatus* and causes most common arboviral infection in children in N. America; hyponatremia and fever associated with worse outcome
Lachman maneuver	for diagnosing ACL tear, pull on tibia in anterior direction with knee flexed at 20-25 degrees (if tibia slides anteriorly >2 mm, positive)
Ladin's sign	an area of elasticity on the anterior face of the uterus just above the cervix; felt on palpation through the vagina as early as the 5th or 6th week
Lady Windermere syndrome	mycobacterium avium complex pulmonary disease described in fastidious elderly women in the dependent portion of the lingula or the right middle lobe due to proposed habitual voluntary suppression of cough leading to the development of nonspecific inflammatory processes in these poorly draining lung regions, upon which MAC-PD engrafted. Chest 1992;101:1605-9
Laennec's cirrhosis	in alcoholic cirrhosis, residual parenchymal nodules that protrude like "hobnails" from the surface of the liver
Lafora's disease	progressive myoclonus epilepsy, autosomal recessive, onset teenage years, characterized by seizures and cumulative neurological deterioration, death usually within ten years of first symptoms; associated with intracellular inclusions (Lafora bodies); associated with mutation in gene *EPM2A*
Laimer triangle	area defined by cricopharyngeus muscle and esophageal muscles
Laki-Lorand factor	factor XIII
Lambert, canals of	direct accessory bronchioalveolar connections
Lambert-Eaton myasthenic syndrome	disease of neuromuscular junction associated with paraneoplastic process with proximal muscle weakness that improves with repetitive stimulation; associated with P/Q type presynaptic voltage-gated calcium channel antibodies; 70% have malignancy, usually small cell cancer
Lambl's excrescences	small filiform or lamellar lesions on aortic valve leaflets, first described in 1856
Lance-Adams syndrome	post hypoxic monoclonus
Lancisi's sign	giant v waves in tricuspid regurgitation
Landau-Kleffner syndrome	acquired epileptic aphasia; childhood disorder characterized by auditory verbal agnosia, aphasia, and seizures
Landolfi's sign	in aortic regurgitation, systolic contraction and diastolic dilation of the pupil
Landsteiner-Weiner blood group system	after Landsteiner and Weiner who who discovered this antigen system; injected Rhesus monkey red blood cells into guinea pigs producing an antibody thought to recognize the antigen D described by Levine in 1939. Later discovered that these antibodies did not recognize D but rather an antigen that is strongly expressed on Rh positive red cells and weakly expressed on Rh negative red blood cells; antigens located on an intercellular adhesion molecule for CD11/CD18.
Langer's lines	lines used for elective incisions, developed by Karl Langer, an anatomy professor. from cadavers in rigor mortis
Langerhans cell	member of class of bone marrow-derived dendritic cells; antigen-presenting cell; differs from other histiocytes by being CD1a-positive

Langerhans cell histiocytosis	encompasses histiocytosis-X, eosinophilic granuloma, Letterer-Siwe disease, and Hand-Schüller-Christian disease
Langerhans granules	see Birbeck granules
Laplace's law	wall tension = pressure x radius (thus colon perforates preferentially at the cecum because of the increased radius and resultant increased wall tension)
Laron dwarfism	autosomal recessive, dwarfism from growth hormone receptor defect leading to failure to produce IGF-1
Lasègue's sign	when patient is supine with hip flexed, dorsiflexion of ankle causing pain or muscle spasm in the posterior thigh indicates lumbar root or sciatic nerve irritation
Lassa fever	hemorrhagic fever caused by Lassa virus, first isolated in 1969 in Nigeria, an arenavirus, spread through contact with secretions (e.g., urine) from infected rats; fatality 15-20% of hospitalized patients; can be treated with ribavirin
Laterjet, nerve of	branch of vagus nerve thought to carry nerves specifically to antrum of stomach
Laurence-Moon syndrome	mental retardation, pigmentary retinopathy, hypogenitalism, and spastic paraplegia; distinct from Bardet-Biedl syndrome
Lawrence-Seip syndrome	two forms: congenital lipodystrophy and acquired generalized lipodystrophy; general disappearance after birth (generally during childhood and may occur following infections); associated with acanthosis nigricans, excess body hair, enlargement of genitalia, diabetes
Leber hereditary optic neuropathy	mitochondrial DNA disease, progressive bilateral loss of central vision, presents 15-35 y.o, associated with cardiac conduction defects and minor neurological manifestations
LeFort I fracture	transverse maxillary fracture above dental apices, which also traverses pterygoid plate; nasal complex stable
LeFort II fracture	fracture through frontal process of maxilla, through orbital floor, and pterygoid plate; midface is mobile
LeFort III fracture	complete craniofacial separation; different from LeFort II in that it extends through the nasofrontal suture and frontozygomatic sutures
Legg-Calvé-Perthes disease	self-limiting hip disorder of children, 4-8 y.o. (M:F 8:1) involving vascular compromise of the capital femoral epiphysis; perhaps some aseptic necrosis action too
Legionnaire's disease	pneumonia caused by Legionella species, aerobic gram-negative bacilli that do not grow on routine bacteriologic media; first identified in 1976 during outbreak at American Legion Convention in Philadelphia
Leigh's disease	autosomal recessive disorder from defects in mitochondrial pathway for converting pyruvate to ATP leading to lactic acidemia, developmental problems, seizures, involvement of periaqueductal gray area, extraocular palsies, weakness, hypotonia, death within 1 to 2 years
Leiner's disease	seborrheic erythroderma associated with diarrhea and failure to thrive and to generate C5a chemotactic factor
Lemierre's syndrome	oropharyngeal infection (usually by anaerobe *Fusobacterium necrophorum*) complicated by internal jugular vein thrombosis and metastatic abscesses in lung, characterized by spiking fevers, tenderness of sternocleidomastoids; see ring enhancement with central lucency in internal jugular vein on CT
Lenègre's disease	sclerodegenerative disease of conduction system, particularly of right bundle branch and left anterior fasicle in people over 50; associated with slow progression to complete heart block
Lenks triad	in retroperitoneal hemorrhage, flank pain, a palpable tender mass, and signs of internal bleeding (e.g. hematuria)
Lennox-Gastaut syndrome	childhood seizure disorder associated with multiple seizure types (including drop attacks, atypical absences general tonic clonic seizures, and myoclonus), slow generalized spike-and-slow-wave EEG, mental retardation, and resistant to standard anti-epileptic medications
Leopold's maneuver	at 28 weeks, four sequential palpations of gravid abdomen to determine the position of the fetus
leprechaunism	1 in 4 million births, associated with elfin-like facies, decreased s.c. adipose tissue, acanthosis nigricans, and growth retardation, failure to thrive, and early death, insulin resistance
Leriche's syndrome	aortoiliac occlusive disease producing distal ischemic symptoms and signs, e.g. pulseless femoral artery: 1. claudication of buttocks, 2. impotence, 3. atrophy of buttocks (seen with iliac occlusive disease)
Leri-Weill syndrome	dyschondrosteosis; a short stature syndrome characterised by mesomelic shortening of the forearms and lower legs and by bilateral Madelung deformity of the wrists
Lesch-Nyan syndrome	a complete lack of hypoxanthine guanine phosphoribosyl transferase (HGPRT, involved in salvage pathway in purine synthesis), X-linked, hyperuricemia, severe neurologic deficits with mental retardation, self-mutilation, and in some cases gouty arthritis
Leser-Trélat sign	the sudden appearance and rapid increase in the number and size of seborrheic keratoses with pruritus; associated with internal malignancy
Letterer-Siwe disease	acute disseminated Langerhans' cell histocytosis; associated with development of cutaneous lesions that resemble a seborrheic eruption secondary to infiltrations of Langerhans' histocytes over the front and back of the trunk and on the scalp; concurrent hepatosplenomegaly, lymphadenopathy, pulmonary lesions, and eventually destructive osteolytic bone lesions
Lev's disease	fibrosis or calcification extending from cardiac fibrous structures into the conduction system

LeVeen shunt	for treating refractory ascites, peritoneovenous shunt connecting peritoneum and central venous system, compare with Denver shunt; LeVeen shunt uses disc valve in firm polypropylene casing
Levine's sign	when describing angina, defining the constricting discomfort with a clenched fist over sternum; q.v. angina for sens and spec; Samuel A. Levine, U.S. cardiologist, 1891-1966; designated hand movements 80% sens 49% spec; Levine sign itself 14% sens for cardiac pain (*BMJ* 1995;311:1660)
Levinthal paradox	magnitude of conformational search problem in protein folding; an unbiased search for all possible protein conformations would take a lifetime, yet proteins fold in microseconds to minutes
Lewis blood group system	named after Mrs. Lewis, pregnant woman in whom anti-Lea was discovered by Mourant in 1946; function of antigens in this system are unknown
Lewy bodies	eosinophilic intracytoplasmic bodies of alpha synuclein in neurons of substantia nigra and locus ceruleus present in Parkinson's disease
Lewy body dementia	most common dementia associated with Parkinson's disease characterized by Lewy bodies found in brain stem and cortex, visual hallucinations which may be exacerbated by the treatment for parkinsonism
Leydig cell tumors	testicular tumor derived from the stroma, may find Reinke crystals (q.v.)
Lhermitte's sign	sudden electric-like shocks extending down the spine on flexing the head; may result from posterior column lesion, toxic effects of radiation; (may be found in vitamin B_6 toxicity); (may be found in vitamin B_{12} deficiency); seen in 3% of multiple sclerosis
Lhermitte-Duclos disease	dysplastic cerebellar gangliocytoma; associated with germ line PTEN mutations; felt to be a part of multiple hamartoma syndrome or Cowden's syndrome
Libman-Sacks disease	in systemic lupus erythematosus, intense mitral and tricuspid valvulitis with development of small, sterile vegetations
Lichtenberg's figures	branching pattern of cutaneous marks pathognomonic for lightning injury
Liddle's syndrome	severe low renin hypertension, hypokalemia, and metabolic alkalosis that mimicked hyperaldosteronism but found to have low aldosterone levels; due to constitutively activating autosomal dominant mutation in beta subunit of epithelial sodium channel
Lieberkühn, crypts of	intestinal glands found between villi
Li-Fraumeni syndrome	mutant p53 allele inherited, predisposition toward breast carcinomas, sarcomas, leukemia, brain tumors, and adrenocortical carcinoma; half have tumors before age 30, 70% before age 90
Light's criteria	for separating transudate v. exudate; if at least one of the criteria is present, fluid virtually always an exudate: 1. pleural fluid/serum protein ratio > 0.5; 2. pleural fluid LDH/serum LDH ratio > 0.6; 3. pleural fluid LDH > 2/3 upper limit of normal serum LDH. Ann Intern Med 1972; 77:507
Likoff's syndrome	in young women (30s-50s), presence of chest pain with abnormal EKG but with normal coronary angiograms (*N Engl J Med*1967;276:1063)
Linton test	modification of Perthes test, elevation of leg with patient supine and tourniquet in place; if veins fail to drain, implies venous obstruction
Lipschultz ulcer	vulvar ulcer
Lisch nodules	in type I neurofibromatosis, pigmented iris hamartomas
Lisker's sign	in thrombophlebitis or deep vein thrombosis, tenderness to percussion of the medial anterior tibial surface
Lissauer's tract	small diameter primary sensory axons (presumably mediating pain and temperature senses) on their way into the dorsal horn
Litten's sign	diaphragmatic movement seen on one side but not the other side in unilateral phrenic nerve palsy
Little's disease	cerebral palsy; specifically congenital spastic diplegia; described by English surgeon Little in 1843
Littre's hernia	hernia involving a Meckel's diverticulum
Löffler's cardiomyopathy	a form of restrictive cardiomyopathy; endocarditis parietalis fibroplastica
Löffler's syndrome	simple pulmonary eosinophilia, characterized by transient pulmonary lesions, eosinophilia in the blood, and a benign clinical course; lungs show alveoli whose septa are thickened by an infiltrate composed of eosinophils and occasional interspersed giant cells, but there is no vasculitis, fibrosis, or necrosis; associated with ascaris, strongyloides
Löfgren's syndrome	sarcoidosis associated with triad of erythema nodosum, bilateral hilar adenopathy, and polyarthritis; 10-15% of sarcoidosis present this way
Looser's zones	radiolucent narrow lines that lie either at right angles or obliquely to the cortical outlines of bones and often transect them; bilateral and symmetric, found at the axillary margins of the scapula, lower ribs, neck of the proximal femurs, and posterior regions of the proximal ulnas; related either to stress fractures or to mechanical erosion by penetrating nutrient arteries; aka Milkman's fractures
Lorenzo's oil	after Lorenzo Odone, diagnosed with adrenoleukodystrophy in 1984, the triglycerides of monounsaturated oleic acid and erucic acid 4:1, felt to reduce very long chain fatty acids to normal levels in ALD
Lou Gehrig's disease	amyotrophic lateral sclerosis (in France, Charcot's disease)
Louis, angle of	sternal angle

Louvel's sign in deep vein thrombosis, venous pain induced by coughing, prevented by pressing over proximal end of vein

Lovibond's angle the angle made by the proximal nail fold and the nail plate

Lowe's syndrome X-linked, oculocerebrorenal syndrome characterized by congenital cataracts, mental retardation, renal tubular acidosis type 2; from defect in lipid phosphatase, phosphatidylinositol 4,5 bisphosphate [PtdIns(4,5)P2]5-phosphatase, which localizes to the Golgi apparatus and is suspected to play a role in Golgi vesicular transport

Lowenberg's sign in deep vein thrombosis, two calves are wrapped with cuffs to see if there is assymetry in tolerance to pressure of 180 mm Hg

Löwenstein-Jensen's medium for growing out *M. tuberculosis*; contains malachite green, a triphenlyamine die like crystal violent, inhibits growth of unwanted organisms during 6 week incubation period as well as complex nutrients

Lown-Ganong-Levine syndrome enhanced AV node pathways, <0.12 s, no QRS widening, no delta waves

Ludwig's angina aggressive infectious process of the submandibular, sublingual, and submental fascial spaces frequently occurring as a result of infection from 2nd and 3rd lower molar; 54% mortality in preantibiotic era, now 4% described in 1836 by Wilhelm Frederick von Ludwig

Lugol's solution 5% iodine and 10% KI, a dose of 6.3 mg of I per drop, for giving iodine

Lund's node lymph node found in Calot's triangle, aka Calot's node

Lundberg A wave in ICP monitoring, waves that have a duration of 5-20 minutes and an amplitude of 50 mm Hg over the baseline ICP, after which ICP is reset to a higher baseline level; sign of severely compromised intracranial compliance and can result in significant decrease in cerebral perfusion pressure and herniation

Luria's law three antibiotics = 1 fungal infection

Luschka, ducts of small tubular channels found buried within the gallbladder wall adjacent to the liver, communicates with the biliary tree, rarely patent accessory bile secretory ducts

Luschka, foramina of two laterally placed foramens exiting out of fourth ventricle

Lutembacher's syndrome atrial septal defect with mitral stenosis; though mitral stenosis is often of rheumatic origin

Lutheran blood group system named after Mrs. Luteran, pregnant woman in whom anti-Lua was discovered by Cutbush in 1945; Cutbush assumed that the name on the sample had been misspelled, hence "Lutheran" instead of "Luteran"; function of antigens in this system unknown

Lyell's syndrome toxic epidermal necrolysis

Lyme disease inflammatory disease caused by spirochete *Borrelia burgdorferi*, spread by Ixodes tick, characterized by early local disease with erythema migrans, myalgias, followed by early disseminated disease characterized by carditis, neurologic findings (lymphocytic meningitis, cranial nerve palsies, also see Bannwarth's syndrome), and late disease characterized by arthralgias and arthritis

Lynch syndrome I cancer family syndrome with multiple colon cancers 2 to 3 decades earlier, predilection for proximal colon; Lynch syndromes most common forms of familial colon cancer, 5-10% of all cases of colon cancer; better prognosis than sporadic colon cancer; deficiency in mismatch repair genes in 85% of Lynch syndromes; autosomal dominant

Lynch syndrome II cancer family syndrome, all features of Lynch I with early onset of carcinoma at other sites including endometrium, ovaries, and stomach, also upper tract TCC

Lyon hypothesis only one of two X chromosomes is genetically active

MacCallum's plaques irregular thickenings, usually in the left atrium, from subendocardial lesions, usually exacerbated by regurgitant jets

MacConkey medium medium for growing nonfastidious Gram-negative rods

Macewen's sign in hydrocephalus before closure of sutures, cracked pot sound when percussing over dilated ventricles

Machado-Joseph disease spinocerebellar ataxia 3, named for affected families of Azorean origin, autosomal dominant disorder caused by a CAG expansion in gene on 14q, onset after age 40, associated with diabetes

Mackler triad in esophageal rupture, triad of vomiting, chest pain, and subcutaneous emphysema

Macleod's syndrome unilateral hyperlucent lung, see Swyer-James syndrome

Maddrey discriminant function for predicting mortality in patients with alcoholic hepatitis, Maddrey DF = 4.6 x (PT-control PT) + serum bilirubin; DF > 32 associated with high short-term mortality, 1 month mort 35% in absence of encephalopathy and 45% if encephalopathy present

Madelung deformity growth disturbance in the volar-ulnar distal radial that results in bowing of the distal end of the radius, resulting in radius shorter than the ulna; presents mainly in adolescent females; also associated with variant of dwarfism, dyschondroestosis

Madelung's disease benign symmetric lipomatosis; characterized by numerous, symmetrically distributed, non tender, poorly demarcated, fatty tumors; distributed mainly in vicinity of neck, sub-occipital region, proximal extremities, and upper part of trunk; can result in "horse-collar appearance."

Madura foot actinomycetoma of the foot; chronic bacterial infection of subcutaneous tissue often due to traumatic injury (walking barefoot); *Nocardia* usual organism (Madura is a city in southern India)

Maffucci's syndrome enchondromatosis associated with soft tissue hemangiomas; associated with ovarian carcinomas and brain gliomas; essentially Ollier's

	disease with hemangiomas
Magendie, foramen of	midline foramen exiting out of fourth ventricle
Maisonneuve fracture	spiral fracture of the proximal third of the fibula associated with ankle fracture (rupture of the distal tibiofibular syndesmosis associated with fracture of the tibia and rupture of the deltoid ligament); usually from ankle eversion injury
Majocchi granuloma	infection of hair follicle from dermatophyte; foreign body granuloma most commonly due to *Trichophyton rubrum* infection, e.g. in young women who frequently shave their legs
Majocchi's disease	purpura annularis telangiectodes
Mallory body	"alcoholic hyalin," an eosinophilic intracytoplasmic inclusion in liver cells that is characteristic of alcoholic liver disease but seen in many other conditions as well (e.g., primary biliary cirrhosis, Wilson's disease, chronic cholestatic syndromes, focal nodular hyperplasia, and hepatocellular carcinoma); inclusions composed largely of intermediate filaments of prekeratin
Mallory-Weiss tears	small defects in gastroesophageal junction, from violent retching and beef with alcoholic gastritis, can bleed like crazy, 87% occur below gastroesophageal junction; bleeding stops 90% without intervention
Malone procedure	creation of a continent appendicostomy or neoappendix as a conduit for irrigation of the colon as treatment for fecal incontinence due to pediatric anorectal malformations, spina bifida, chronic constipation, etc. as an alternative to rectal enemas.
Malta fever	brucellosis
Maltese cross	fat droplets in urine
Mantoux test	test for tuberculosis with intradermal injection of purified protein derivative of tuberculin, 0.1 mL of 5 tuberculin units
maple syrup urine disease	deficiency in branched-chain alpha ketoacid dehydrogenase resulting in increased levels of branched chain alpha amino acids and alpha-keto analogs in plasma and urine, 1:200,000, high mortality
Marburg's disease	acute form of multiple sclerosis, fulminant and progressive
Marchiafava-Bignami syndrome	seen in malnourished alcoholics, necrosis of corpus callosum and subcortical white matter leading acutely to chronically to dementia, spasticity, dysarthria, gait disorder, and coma
Marcus Gunn pupil	afferent pupillary defect, pupil dilates instead of constricts because of optic nerve defect; named after Scottish ophthalmologist Robert Marcus Gunn
Marfan's syndrome	1/10,000-1/20,000 autosomal dominant, mutation in fibrillin-1 gene, 50% of children have dilatation of aorta
Marie's sign	fine tremor in hyperthyroidism
Marie-Strümpell disease	refers to ankylosing spondylitis in Europe
Marine-Lenhart disease	co-existence of TSH-dependent nodules and Graves's disease
Marion's disease	congenital obstruction of the posterior urethra
Marjolin's ulcer	squamous cell carcinoma ulceration overlying chronic osteomyelitis or burn scar
Markesbery distal myopathy	distal myopathy associated with rimmed vacuoles
Markle sign	jar tenderness in abdomen from heel drop as a localizing sign of peritoneal irritation; described in 1973
Maroni sign	erythema of the skin overlying thyroid in hyperthyroidism
Maroteaux-Lamy syndrome	mucopolysaccharidosis (VI) from arylsulfatase B deficiency leading to accumulation of dermatan sulfate characterized by skeletal dysplasia, corneal clouding, coarse facies, valvular heart disease
Marshall syndrome	pediatric condition characterized by periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis (PFAPA); origins of syndrome unknown
Marshall syndrome	dominantly inherited chondrodysplasias characterized by midfacial hypoplasia, high myopia, and sensorineural hearing deficit, from mutation in COL11A1; similar to Stickler syndrome
Marshall, vein of	oblique vein of left atrium
Marshall-Smith syndrome	accelerated skeletal maturation, failure to thrive, and dysmorphic facial features with death in early infancy or childhood from pulmonary infections
Martin-Gruber anastomosis	anatomic anomaly where there is a branch from the median to ulnar nerve in the forearm, innervating 1st dorsal interosseus, adductor pollicis, and abductor digiti minimi, seen in 10-44% of population
Martin-Lewis agar	variant of chocolate agar medium for growing *Neisseria*
Martorell's ulcer	associated with poorly controlled hypertension, punched out ulcers, sharply demarcated, with surrounding halo of erythema, very painful, often crusted, relieved by placing leg in dependent position, found on anterior external aspect of leg between and middle and lower third of limb

Mary Walker effect	effect of physostigmine reversing the effects of myasthenia gravis; described in 1934; Mary Walker, an assistant medical officer at St. Alfege's Hospital in Greenwich England 1920-1936
Masson body	granulation tissue plug found in lumen of small airways in BOOP
Mattox maneuver	for gaining access to the retroperitoneal structures (including aorta), left medium visceral rotation
Matuhasi-Ogata phenomenon	non-specific binding of IgG to antibody-coated red cells; binding of an antibody to a red blood cell that lacks the antigen toward which the antibody is directed due to the presence of antibodies directed toward antigens present on the red blood cell surface.
Maurer's dots	seen in *P. falcipirum*, stippling of erythrocytes
Mayer's reflex	basal joint reflex; adduction of the thumb in response to flexion of the MCP joint of the ring finger in a person with a relaxed hand, a normal finding, a normal finding, but may be absent in pyramidal lesions
Mayer-Rokitansky-Küster-Hauser syndrome	absence of vagina from abnormality of müllerian development
May-Hegglin anomaly	neutrophils with large pale blue inclusions resembling Döhle bodies, giant platelets (30-80 fL v. 7-10 normally), variable thrombocytopenia; rare AD asymptomatic trait
Mayne's sign	in aortic regurgitation, decrease in diatolic pressure of 15 mm Hg when arm is held above the head; of questionable utility since observed in 65% of normal people
Mayo, veins of	vein overlying pylorus
May-Thurner syndromne	iliofemoral deep vein thrombosis from impaired venous return because of compression of the left common iliac vein by overlying right common iliac artery
Mazzotti reaction	reaction to proteins released by dying onchocerca, including fevers, rashes, ocular damage, joint and muscle pain, and lymphangitis as well as hypotension, pyrexia, respiratory distress, and prostration
McArdle's syndrome	glycogen storage disease V, deficiency in muscle phosphorylase, with consequent glycogen accumulation in skeletal muscle, produces painful muscle cramps and muscle weakness following exercise
McBurney's point	one-third the distance from the ASIS to the umbilicus
McBurney's sign	tenderness at McBurney's point in appendicitis
McConnell's sign	in acute pulmonary embolism, distinct regional pattern of RV dysfunction, with akinesia of the mid-free wall but normal motion at the apex, 77% sensitive, 94% specific for PE
McCune-Albright syndrome	triad of irregular café au lait spots, fibrous dysplasia of long bones with cysts, and precocious puberty
McDonald's sign	uterine corpus and cervix can be easily flexed on each other due to Hegar's sign
McGinn-White sign	S1Q3T3 pattern in pulmonary embolism; described in 1935
McLeod phenotype	occurs from the absence of the Kx red blood cell antigen; characterized by acanthocytes; compensated hemolytic anemia; decreased Kell system antigens; increased CKMM isoenzymes; and progressive neurologic changes including areflexia, choreiform movements, dysarthria, wasting of muscles, and cardiomyopathy; may be associated with X-linked chronic granulomatous disease
McMurray test	for diagnosing meniscal tear, passively flex hip and knee until heel touches buttock, steady the knee with one hand, externally rotate foot, then extend the knee to 90 degrees, return ot the beginning and then internally rotate the foot, then passively extend the knee to 90 degrees, feeling for clicks, PPV 83% for tears
McRoberts's maneuver	flexion of legs onto maternal abdomen for shoulder dystocia, causeing a significant cephalad rotation of the symphysis pubis and subsequent flattening of the sacrum.
Means-Lerman scratch	in hyperdynamic heart (as in hyperthyroidism), a systolic scratch occasionally heard in the second left intercostal space during expiration, presumed to be secondary to rubbing together of normal pleural and pericardial surfaces
Meckel scan	^{99}Tc pertechnetate scan that selectively tags acid secreting cells (gastric mucosa); it is used most often for unexplained bleeding in infants and young adults
Meckel's cartilage	branchial arch 1
Meckel's cave	located in skull base, site of trigeminal ganglion and its divisions; grandson described Meckel's diverticulum.
Meckel's diverticulum	persistence of vitelline duct, contains all 3 layers of bowel (mucosa, submucosa, muscularis propria), antimesenteric, present in 2% of population, usually within 30 cm of ileocecal valve; the five 2s: 2" long, 2 feet from ileocecal valve, 2% of population, commonly persists in first 2 years of life, may have 2 types of epithelia
Meckel-Gruber syndrome	sloping forehead, posterior encephalocele, polydactyly, and polycystic kidneys
Mees's lines	horizontal white bands of the nails seen in chronic arsenic poisoning, and occasionally in leprosy; usually appears 6 weeks after exposue to arsenic; R.A. Mees, 20th century Dutch physician
Meige's disease	lymphedema praecox (in contrast to Milroy's disease)

Meige's syndrome	blepharospasm with facial dystonia and lip smacking
Meigs's syndrome	unusual combination of hydrothorax, ascites, and ovarian fibroma
Meissner's corpuscle	mechanoreceptor in the skin, sensitive to light touch, asymmetrical, lamellated
Meissner's plexus	submucous plexus; innervates glandular epithelium, muscularis mucosa, intestinal endocrine cells, and submucosal blood vessels
MELAS	mitochondrial encephalopathy with acidosis and stroke, onset in childhood, stroke-like episodes with hemiparesis, hemianopia, or cortical blindness; full expression of the disease leads to death often before age 20; 80-90% have point mutation in leucine tRNA in mtDNA
Meleney's ulcer	necrotizing fascitis
Melkersson-Rosenthal syndrome	triad of recurrent facial paralysis, recurrent and eventually permanent facial, especially labial edema, and plication of the tongue; etiology unknown
Meltzer's traid	in cryoglobulinemia, triad of palpable purpura, arthralgia and myalgia
Mendelson's syndrome	chemical pneumonitis after aspiration of gastric contents
Ménétrier's disease	giant cerebriform enlargement of the rugal folds of the gastric mucosa, results from profound hyperplasia of the surface mucous cells with accompanying glandular atrophy, most often encountered in men (3:1), 40s-60s, sometimes in children, may produce epigastric discomfort, weight loss, and sometimes bleeding related to superficial rugal erosions, gastric secretions mostly mucous, little HCl, may be sufficient protein loss to produce hypoalbuminemia
Ménière's disease	1. fluctuating sensorineural loss, classically involving low frequencies; 2. vertiginous episodes; 3. aural pressure; 4. tinnitus (frequently roaring); pathologic changes consist of dilation of the endolymphatic system that leads to degeneration of vestibular and cochlear hair cells
Menkes's disease	X-linked, problem in distribution of copper in the body, amount of copper and ceruloplasmin in serum reduced, excess of copper in intestinal mucosa, muscle, spleen, and kidney, associated with brittle hair; usually fatal by age 3; believed to be due to defect in copper-transporting-ATPase (ATP7A)
Mentzer index	MCV/RBC ratio; >13 iron def, <13 thalassemia (*Lancet* 1973 Apr 21;1:882)
Merkel cell	cells located in the skin associated with sensory neurites in the dermal papillae, which are the mechanoreceptors of the skin; speculated to be the cell of origin of Merkel cell carcinoma
Merkel cell carcinoma	rare aggressive neuroendocrine tumor involving the skin, predominantly affects elderly Caucasians with a propensity for local recurrence and regional lymph node metastases
MERRF	myoclonic epilepsy and ragged red fibers, syndrome of mitochondrial myopathy, myoclonus, generalized seizures, intellectual deficits, ataxia, and hearing loss; extraocular movements normal; associated with point mutations in lysine tRNA in mtDNA
Meyer's loop	a portion of the optic radiations subserving vision from the superior field coursing rostrally within the temporal lobe before heading caudally to the primary visual cortex; lesions produce contralateral upper quadrantanopia (pie in the sky)
Meynert, basal nucleus of	degenerates in Alzheimer's; uses acetylcholine
Mibelli, porokeratosis of	classic porokeratosis, localized, chronically progressive, hyperkeratotic, irregular plaques with central atrophy and prominent peripheral keratotic ridge; characterized pathologically by presence of cornoid lamella
Middlebrook media	medium used for growing mycobacterium
Mikulicz's syndrome	bilateral inflammatory enlargement of the parotid, submaxillary, and sublingual, and lacrimal glands and xerostomia, secondary to sarcoid, leukemia, lymphoma, etc.
Milkman's fractures	in osteomalacia, radiolucent narrow lines that lie either at right angles or obliquely to the cortical outlines of bones and often transect them; bilateral and symmetric, found at the axillary margins of the scapula, lower ribs, neck of the proximal femurs, and posterior regions of the proximal ulnas; related either to stress fractures or to mechanical erosion by penetrating nutrient arteries; aka Looser's zones
Millard-Gubler syndrome	ventral pontine injury causing symptoms similar to Fouville's syndrome except lateral rectus weakness only, instead of gaze palsy
Miller Fisher syndrome	variant of Guillain-Barré where cranial nerves are affected, leading to triad of ataxia, areflexia and ophthalmoplegia; cross-reacting antibodies to GQ1b ganglioside have been found; triggered by certain strains of *C. jejuni*
Miller Fisher test	in normal pressure hydrocephalus, objective gait assessment before and after 30 cc CSF removed reflecting prognosis for shunting
Milroy's disease	congenital lymphedema with swelling present from time of birth (in contrast to Meige's disease), mainly below the waist, associated with mutation in VEGF C/D receptor (VEGFR-3)
Milwaukee brace	brace used for treating scoliosis of the back
Milwaukee shoulder	basic calcium phosphate disease associated with large joint destructive arthropathy (especially shoulder) and rotator cuff tears
Mirizzi's syndrome	extrinsic obstruction of the common bile duct from a cystic duct gallstone

Mississippi mud	vancomycin, referring to brownish, muddy appearance of early preparations
Mitchell disease	erythromelalgia
mitral facies	malar flush with pinched and blue facies in mitral stenosis
Mobitz type I AV block	Wenckebach pattern, second degree block with intermittent conduction failure resulting in progressive PR prolongation before dropped beat; may be seen in ischemic heart disease, digitalis, beta blockers, calcium channel blockers, inferior wall MI
Mobitz type II AV block	second degree block with intermittent conduction failure with sudden non-conducted sinus P wave without progressive prolongation of PR interval; not seen with digitalis excess or inferior wall MI generally but may be seen with anterior wall MI
Möbius syndrome	congenital facial paralysis with or without limb defects associated with maternal misoprostol use
Möbius's sign	in Graves's ophthalmopathy, failure of ocular convergence following close accommodation at a distance of 5 inches
Modigliani syndrome	thyroid in normal position but people with long curving necks enhance prominence and palpation of thyroid
Moersch-Woltmann syndrome	stiff-man syndrome; associated with antibodies to glutamic acid decarboxylase (GAD)
Mohs's surgery	surgery involving microscopic examination of tissue being removed; generally used for skin cancers; developed by Frederic Mohs in the 1930s
Mollaret's meningitis	benign recurrent aseptic meningitis characterized by large monocytic cells in CSF; significant percentage due to HSV-2
Mönckeberg's arteriosclerosis	ring-like calcifications within the media of medium-sized to small muscular arteries (femoral, tibial, radial, and ulnar arteries, genital arteries), occurs almost exclusively in individuals over 50 years old; doesn't narrow lumen, distinct from atherosclerosis
Mondini malformation	cause of sensorineural hearing loss due to developmental anomaly of otic capsules with developmental arrest of cochlea in seventh fetal week; increased risk of developing recurrent meningitis or perilymphatic fistula
Mondor's disease	thrombophlebitis of superficial breast veins
Monge's disease	chronic mountain sickness, loss of high altitude tolerance after prolonged exposure, characterized by extreme polycythemia, exaggerated hypoxemia, and reduced mental and physical capacity; relieved by descent
Monod's sign	in aspergillomas, radiolucent crescent seen around solitary 3-5 cm lesion on chest X-ray
Monro, foramina of	connects each of the lateral ventricles with the third ventricle
Monro-Kellie doctrine	cranial cavity is a closed rigid box, change in the amount of intracranial blood can occur only through changes in CSF quanitity
Monsel's solution	ferric subsulfate, solution used for its styptic properties (stopping bleeding)
Monteggia fracture	fracture of the proximal third of the ulna with a dislocation of radial head
Montgomery, glands of	sebaceous glands of areola
Mooren corneal ulcers	chronic, painful ulcers, involves circumference of peripheral cornea and may progress to vision loss; associated with hep C
Morgagni, hydatids of	fallopian tube cysts found near the fimbriated end or in the broad ligaments
Morgagni's hernia	anterior parasternal diaphragmatic hernia, right more common than left
Morquio's syndrome	mucopolysaccharidosis (IV) from N-acetylglucosamine-6-sulfate sulfatase deficiency leading to accumulation of keratan sulfate and chondroitin-6 sulfate, characterized by distinctive skeletal deformity, corneal clouding, odontoid hypoplasia, and aortic valve disease but with normal intelligence
Morrison's pouch	hepatorenal recess; the most posterior cavity in the peritoneal cavity
Morton's neuroma	interdigitial palmar neuroma
Morvan's syndrome	fibrillary chorea; neuromyotonia, pain, weight loss, severe insomnia and hallucinations, excess secretions (sweating, lacrimation, and salivation); associated with voltage-gated potassium channel antibodies
Moschowitz syndrome	thrombotic thrombocytopenic purpura, described in 1924
Moses's sign	see Bancroft's sign
Mott cells	cytologic variant of plasma cells with globular cytoplasmic inclusions that contain immunoglobulin proteins (blue, grapelike cytoplasmic droplets); seen in multiple myeloma and trypanosomiasis
Mounier-Kuhn syndrome	tracheomegaly, tracheobronchomegaly associated with recurrent pneumonia, copious purulent sputum production, hoarseness, load cough; presents in 30s-40s, more common in males; tracheal lumen increases with Valsalva; autosomal recessive
moyamoya disease	occlusive disease involving large intracranial arteries, especially the distal ICA and stem of the MCA and ACA; "puff of smoke" (in Japanese, *moya moya*) appearance on angiography from lenticulostriate arteries developing rich collateral circulation around the middle cerebral occlusion that; anticoagulation risky because of occurrence of SAH from rupture of the transdural anastomotic channels
Mucha-Habermann disease	pityriasis lichenoides et varioliformis acuta, scattered necrotic papules and vesicles that can resemble insect bites but usually are more generalized and symmetric

Muckle-Wells syndrome	syndrome of urticaria, progressive perceptive deafness, and amyloidosis; from mutation in cryopyrin; autosomal dominant; of note familial cold autoinflammatory syndrome, commonly known as familial cold urticaria, characterized by intermittent episodes of rash, arthralgia, fever and conjunctivitis after generalized exposure to cold is also associated with mutations in cryopyrin.
Muehrcke's nails	paired narrow horizontal white bands that are immobile as nail grows; seen in hypoalbuminemia, nephrotic syndrome
Muenke syndrome	nonsyndromic coronal craniosynostosis, associated with a specific mutation of the fibroblast growth factor receptor-3 gene (FGFR3), Pro250 to Arg
Muir-Torre syndrome	patients with hereditary nonpolyposis colon cancer (HNPCC) who also develop benign or malignant sebaceous skin tumors (often basal cell or squamous cell)
Mulder's sign	palpable click in Morton's neuroma when compressing the forefoot and pushing up in the distal third intermetatarsal space
Müller's maneuver	reverse Valsalva
Müller's sign	in aortic regurgitation, systolic pulsations of the uvula
Mullerian duct	in female embryonic development, duct that gives rise to fallopian tubes, uterus, and upper vagina
Mullerian inhibiting substance	in embryonic development, gonadal peptide hormone that initiates regression of Mullerian duct during male sexual development
Müller-Lyer illusion	visual illusion involving two horizontal lines with arrows where line with inward facing arrows appears shorter than line with outward facing arrows
Munchausen's syndrome	factitious disorder, eponym given by Asher in 1951 after Baron Karl Munchausen (1720-1797), retired German cavalry officer with a reputation for preposterous stories about his activities as a soldier, hunter
Munro's microabscesses	in psoriasis, when neutrophils form small aggregates within the parakeratotic stratum corneum
Murphy drip	saline infusion in the rectum for treating peritonitis; also refers to continuous irrigation of the bladder; after John Benjmain Murphy surgeon (1896-1982) of "Murphy's sign."
Murphy eye	small hole in the side of the endotracheal tube that allows for ventilation if the distal end becomes occluded
Murphy's punch sign	tenderness over costovertebral angle suggesting pyelonephritis
Murphy's sign	a sharp increase in tenderness with a sudden stop in inspiratory effort, sign of acute cholecystitis; 27-97.2% sensitive, 48.3% specific
Mustard procedure	for treating transposition of great vessels, now rarely used; an atrial inversion procedure which connects RA to LV, which pumps out to pulmonary arteries, and connects LA to RV, which becomes systemic pump to aorta; variant uses pericardial or prosthetic intraatrial baffles.
Myerson's sign	repetitive tapping (about twice per second) over the bridge of the nose producing a sustained blink response in parkinsonism and as frontal release sign
Naclerio, V sign of	in esophageal rupture, V-shaped collection of air along mediastinum and diaphragm
Nägele's rule	means of estimating date of delivery by counting back 3 months from the first day of the last menstrual period and adding seven days; full term 38 weeks after fertilization, 40 weeks after LNMP
Nardi test	in relapsing pancreatitis, narcotic-induced stimulation or spasm reproducing abdominal pain and amylase elevation (for inferring sphincteric disease in any pancreatic or biliary ductal system without a gallbladder), presumably accurate in the diagnosis of perisphincteric disease
Naxos disease	initially described on Greek island of Naxos, synrome of arrhythmogenic right ventricular cardiomyopathy, non-epidermolytic palmoplantar keratoderma, and woolly hair, due to deletion in plakoglobin, protein in adherens and desmosomal junctions
Neer sign	in shoulder impingement, forcibly flexing the arm to an overhead position to roduce pain from impingement of humerus against coracoacromial arch
Negri bodies	in rabies virus-infected brain neurons, eosinophlic cytoplasmic inclusions
Nelson's syndrome	enlargement of a pituitary adenoma in a patient with Cushing's disease whose adrenals have been removed from loss of feedback inhibition of cortisol
nerve endings	free includes Merkel cell associated: found in the epidermis as small aggregates called tactile corpuscles (Merkel cell has neural crest and squamous properties); encapsulated includes Meissner's: asymmetrical, lamellated; Pacinian: symmetrical, lamellated; Ruffini: no lamellation
Nicolau's syndrome	livedo-like dermatitis from acute arterial thrombosis occurring immediately after intravascular injection of an insoluble drug substance
Niemann-Pick disease	a sphingolipidosis where there is lysosomal accumulation of sphingomyelin and cholesterol, type A and B (deficiency of sphingomyelin-cleaving enzyme sphingomyelinase) and type C and D (enzyme normal or nearly normal, defect in esterification and transport); type A, 75-80% of all cases, extensive neurologic involvement, marked visceral accumulations of sphingomyelin, and progressive wasting and early death within the first 3 years of life); half have cherry red spot in macula as in Tay-Sachs
Niikawa-Kuroki syndrome	see kabuki syndrome
Nijmegen breakage	primary immunodeficiency autosomal recessive disorder that presents as microcephaly, a distinctive "bird-like" facies, growth

syndrome	retardation, recurrent pulmonary infections, chromosomal instability, radiation hypersensitivity, and increased incidence of malignancies, especially lymphomas from mutation in nibrin protein, a protein involved in DNA repair; cellular defect similar to ataxia-telangiectasia
Nikolsky's sign	associated with pemphigus vulgaris; apparently normal epidermis may be separated at the basal layer and rubbed off when pressed with a sliding motion
Nissen fundoplication	for treating gastroesophageal reflux disease, surrounding lower end of esophagus with cuff of gastric fundal muscle to increase lower esophageal sphincter competence
Nissl bodies	in neurons, rough endoplasmic reticulum
Nissl stain	stains cell bodies dye binds to acid groups, in particular the RNAs of the ribosomes located within the cell body; Franz Nissl, German medical student at time
Noonan's syndrome	Turner's phenotypic characteristics without any sex chromosome abnormalities, males or females with congenital heart defect (usually pulmonic stenosis and hypertrophic cardiomyopathy), pectus carinatum, short stature, mild mental retardation, hypertelorism, and webbed neck; autosomal dominant; associated with mutation in nonreceptor protein tyrosine phosphatase SHP2 (*PTPN11*); most common non-chromosomal cause of congenital heart disease
Norman-Landing disease	GM1 gangliosidosis, a lysosomal storage disorder
Norwalk virus	calcivirus associated with gastroenteritis; named after outbreak in a school in Norwalk, Ohio in 1969
Norwegian scabies	crusted scabies, highly contagious, psoriaform dermatosis of the hands and feet with dystrophy of the nails and an erythematous scaling eruption that may be generalized; itching minimal; seen in institutionalized patients, mentally retarded, and in patients with HIV, HTLV-1, or adult T cell leukemia,
Norwood procedure	for treating hypoplastic left heart syndrome, has 3 stages. Stage 1, anastomosis of proximal main pulmonary artery to aorta, with aortic arch reconstruction and transection and patch closure of distal main pulmonary artery; modified right Blalock-Taussig shunt (subclavian artery to right PA) to provide pulmonary blood flow. ASD created to allow for adequate left to right flow. Stage 2, Bidirectional Glenn shunt to reduce volume overload of single right ventricle. Stage 3, modified Fontan procedure to correct cyanosis.
Nothnagel's syndrome	injury to superior cerebellar peduncle causing ipsilateral oculomotor palsy and contralateral cerebellar ataxia
Ober's test	provocative test for iliotibial band syndrome: patient lies on unaffected side and affected leg is abducted while knee is flexed to 90 degrees and leg is then released; positive for iliotibial band syndrome when thigh is released and thigh remains abducted
Occam's razor	described in 14th century, "plurality must not be posited without necessity," in other words, parsimony in diagnosis
Oddi, sphincter of	sphincter around opening of common bile duct into the duodenum; Oddi, Italian anatomist and surgeon (1864-1913)
Ogilvie's syndrome	massive idiopathic non-obstructive dilatation of the colon
Oguchi disease	congenital night blindness; asymptomatic during day; found to be due to mutations in arrestin (in Japanese Oguchi disease) and rhodopsin kinase (in European Oguchi disease)
Oliver's sign	in aortic aneurysm, pulsation of aorta felt through cricoid cartilage when chin is elevated
Ollier's disease	syndrome of multiple enchondromas
Omenn's syndrome	combined immunodeficiency associated with hypereosinophilia, erythroderma, hepatosplenomegaly, lymphadenopathy, alopecia; autosomal recessive; associated with mutations in Rag-1 and Rag-2 proteins and decreased V(D)J recombination efficiency; also associated with mutation in ARTEMIS protein (involved in opening coding end hairpins)
Ommaya reservoir	device implanted under the scalp that communicates with the CSF and allows intrathecal delivery of therapeutic agents (e.g., chemotherapy)
Ondine's curse	faiulure of the automatic control of ventilation, named after mythological tale in which the suitor of Neptune's daughter was cursed to lose automatic control over all bodily functions; associated with mutation in ret proto-oncogene; associated with Hirschsprung's disease
Onufrowicz, nucleus of	parasympathetic neurons in the sacral spinal cord that innervate the sphincters of the bowel and bladder
Opitz syndrome	hypospadias-dysphagia syndrome or telecanthus with associated abnormalities; associated with midline abnormalities such as cleft lip, laryngeal cleft, heart defects, hypospadias, and agenesis of the corpus callosum
Oppenheim's sign	involuntary dorsiflexion of the toes when stroking the medial/anterior tibial surface superiorly to inferiorly, indicating upper motor neuron defect
Ormond's disease	retroperitoneal fibrosis
Oroya fever	from *Bartonella bacilliformis* (transmitted by sandfly vector Phlebotomus found in valleys of the Andes mountains, 600-2500 m): profound intravascular hemolytic anemia of a few weeks duration, associated with lesions called verruga peruana resembling Kaposi's sarcoma; aka Carrión's disease
Ortner's syndrome	hoarseness from compression of left recurrent laryngeal nerve by a greatly dilated left atrium (e.g., in mitral stenosis), enlarged tracheobronchial lymph nodes, and dilated pulmonary artery

Ortolani's sign	in congenital hip dislocation, rotate hip with patient in supine position and hip abducted; a "clunk" or "click" represents congenitally dislocated hip
Osborn wave	EKG with distinctive convex "hump" at J point associated with hypothermia and temperature around 32C
Osgood-Schlatter's disease	epiphysitis of tibial tubercle resulting from repeated powerful contractions of the quadriceps seen in adolescents with open physis
Osler's nodes	tender to painful, purplish, split pea-sized, subcutaneous nodules in the pulp of the fingers and/or toes and thenar and hypothenar eminences; transient, disappearing within several days (5% of patients); in acute bacterial endocarditis, associated with minute infective emboli; aspiration may reveal the causative organism; in subacute bacterial endocarditis, associated with immune complexes and small-vessel arteritis of skin
Osler's sign	palpable brachial or radial artery when cuff > systolic pressure
Osler-Weber-Rendu disease	hereditary hemorrhagic telangiectasia, larger lesions can be a source of chronic blood loss, systemic emboli, hypoxemia, hepatic dysfunction, and a high-output cardiac failure; important risk factor for brain abscess, especially in affected patients with clubbing, cyanosis, and/or polycythemia; use aminocaproic acid (an antifibrinolytic agent)
Ota, nevus of	pigmentation mostly involves the skin and mucous membranes innervated by the first and second branches of the trigeminal nerve
Ottawa ankle rules	decision guide for managing acute ankle injury and ordering ankle x-ray and foot x-ray series. Ankle x-ray series required only if there is any pain in the malleolar zone and any of these findings: bony tenderness at posterior edge or tip of lateral malleolus, base of fifth metatarsal, or inability to bear weight both immediately and in emergency department. Foot x-ray is required only if there is any pain in the midfoot zone and any of these findings: bone tenderness at base of fifth metatarsal, bone tenderness at navicular, or inability to bear weight both immediate and in emergency department
Ouchterlony reaction	test based on the precipitin reaction where antigen and antibody are placed in separate wells cut in agar; precipitate forms a band between wells at the optimal concentration; described by Ouchterlony in 1948
Owren's disease	factor V deficiency; parahemophilia
Pacinian corpuscles	mechanoreceptor located in skin, sensitive for vibration and touch; symmetrical, lamellated
Paget's disease of bone	osteitis deformans, characterized by initial osteolytic stage followed by a mixed osteoclastic-osteoblastic stage, which ends with osteoblastic activity and evolves into a burnt-out osteosclerotic stage; increased alkaline phosphatase and increased urine hydroxyproline
Paget's disease of breast	breast cancer involving nipple and areola, appears as eczematous rash over breast with crusting, scaling, or erosion
Paget's disease of vulva	intraepithelial adenocarcinoma, less than 1% of all vulva malignancies
Paget-Schroetter syndrome	effort thrombosis of the subclavian vein
Palla's sign	in pulmonary embolism, enlarged right descending pulmonary artery
Panayiotopoulos syndrome	benign childhood partial seizure characterized by vomiting, deviation of the eyes and seizures lasting for more than 30 min (without permanent injury)
Pancoast's tumor	apical lung cancers in the superior pulmonary sulcus which invade neural structures around the trachea, including the cervical sympathetic plexus, leading to severe pain in distribution of the ulnar nerve and Horner's syndrome on same side of the lesion
PANDAS	Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infections. Consists of tics, movement disorders, emotional-lability, and obsessive-compulsive behavior associated with streptococcal infection. Caused by IgG antibodies which recognize group A streptococcal cell wall proteins as well as neuronal cytoplasmic antigens of the basal ganglia. Related to Sydenham's chorea
Paneth cells	in small intestine crypts, cells with apically oriented bright eosinophilic granules and which appear to play a role in the mucosal immune system
Panner disease	avascular necrosis or osteochondrosis of the capitellum (head of humerus)
Pap smear	developed by Papanicolaou, reported in 1941, used for screening cervical cancer
Papez circuit	pathway from subiculum to the mamillary body and back to the cingulate gyrus believed to play a role in emotion (initially) and memory
Papillon-Lefevre disease	early development of severe periodontal disease, palmoplantar keratosis; autosomal recessive; associated with mutation in cathepsin C gene
Pappenheimer bodies	iron-containing granules, small dark blue irregularly shaped granules often in clusters, smaller than Howell-Jolly bodies, commonly seen in sideroblastic anemias, thalassemias, megaloblastic anemias, and in post-splenectomy states
Pardee waves	symmetrically inverted T waves in myocardial ischemia or infarction
Parinaud's oculoglandular	preauricular node enlargement associated with chronic granulomatous conjunctivitis; atypical form of cat scratch disease from inoculation near eye

syndrome	
Parinaud's syndrome	dorsal midbrain syndrome with supranuclear vertical gaze disorder from damage to posterior commissure with loss of upgaze, convergence-retraction nystagmus, downward ocular deviation, lid retraction, due to hydrocephalus from aqueductal stenosis, pineal region tumors
Parkinson's disease	degenerative disorder of central nervous system due to degeneration of dopaminergic neurons of the substantia nigra which project to the striatum; characterized by tremor, rigidity, akinesia, and gait disturbance
Parkland formula	total body surface area % burned x kg x 4; 1/2 in first 8 hours, second 1/2 given next 16 hours
Parry-Romberg syndrome	acquired progressive hemifacial atrophy
Parsonage-Turner syndrome	acute brachial neuritis
Passy-Muir valve	speaking valve for patients with tracheostomy developed by Patricia Passy and David Muir
Pastia's sign	associated with scarlet fever (GAS or *S. aureus* rarely); finely punctate erythema has become confluent (scarlatiniform) on the lower trunk and thighs with petechiae having a linear configuration in the inguinal regions
Patau's syndrome	trisomy 13, 1/15,000 births, mental retardation, microcephaly, microphthalmia, brain abnormalities, cleft lip and palate, polydactyly, rocker-bottom feet, and congenital heart disease
Patrick sign	hip pain on external rotation of the hip in hip joint disease that may refer pain to back and thighs
Patterson-Brown-Kelly syndrome	Plummer-Vinson syndrome
Paul-Bunnell test	agglutination of sheep red blood cells by heterophile antibodies
Paul-Bunnell-Davidsohn test	extension of classic Paul-Bunnell test for heterophile antibody; antibodies not absorbed by guinea pig kidney cells but cause sheep erythrocytes to agglutinate
Pautrier's microabcesses	q.v. Sézary-Lutzner cells
Pearson syndrome	refractory sideroblastic anemia with vacuolization of marrow precursors and exocrine pancreatic dysfunction from mitochondrial DNA mutation
Pel-Ebstein fever	in Hodgkin's disease, unusual systemic manifestation of a periodic fever that is present for some days, remits, and then returns
Pelger-Hüet anomaly	seen in blood of AML (e.g., M2) or myelodysplastic syndromes or inherited as autosomal recessive trait and maybe sideroblastic
Pelizeus-Merzbacher disease	mutation in proteolipid protein on X chromosome, a major protein in CNS myelin; results in hypomyelination confined to the CNS, whitespread white matter dysfunction, leading to seizures, mental retardation, and death in childhood; see "tigroid" appearance on tissue sections stained for myelin
Pellegrini-Stieda syndrome	calcification of medial collateral ligament at its femoral insertion; usually due to trauma
Pemberton's sign	in SVC (superior vena caval) obstruction, development of facial plethora, inspiratory stridor, and non-pulsatile elevation of the JVP when patient lifts arms over head
Pendred's syndrome	autosomal recessive goiter and congenital sensorineural deafness from mutation in pendrin, a transport protein that affects organification of thyroglobulin; also see malformations of cochlea and hypothyroidism (40%); pendrin involved in transport of chloride and iodide
Percheron, artery of	anatomic variant where artery arises from posterior communicating artery and then gives rise to bilateral medial thalamic perforators; occlusion results in bilateral paramedian thalamic infraction; an example where cerebral vessel supplies structures on both sides of the midline
Perl's stain	stain for iron; tissue section treated with hydrochloric acid to release ferric ions from binding proteins; section then treated with potassium ferrocyanide to produce an insoluble blue compound (the Prussian blue reaction)
Perlia's nucleus	cell group located between motor neuron groups innervating left and right medical rectus muscles; felt to be involved in ocular convergene
Perthes test	testing for competence of deep femoral veins; tourniquet is applied at mid-thigh with patient standing and patient then walks for 5 min with tourniquet pin place; reduction in size of veins indicates patent valves and lumens
Petersen's hernia	hernia involving the space between the mesentery of the Roux limb and the transverse colon; can occur following gastric bypass surgery
Peutz-Jeghers syndrome	rare AD syndrome characterized by multiple hamartomatous polyps scattered throughout the entire GI tract and melanotic mucosal and cutaneous pigmentation around the lips, oral mucosa, face, genitalia, and palmar surfaces; patients have increased risk of carcinomas of pancreas, breast, lung, ovary, uterus, and malignant transformation in small bowel polyps, colon cancer, sex cord tumors with annular tubules of the ovary; associated with mutation in *STK11/LKB1* gene and dysregulation of mTOR
Peyrone's salt	cisplatin; first synthesized by Peyrone in 1845

Peyronie's disease penile fibromatosis, a palpable induration or mass appears on the dorsolateral aspect of the penis. It may cause eventually abnormal curvature of the shaft or constriction of the urethra, or both

Pfannenstiel's incision low transverse abdominal incision with retraction of the rectus muscles laterally, used in ob/gyn procedures

Pfeiffer bacterium *H. influenza*, found in respiratory tracts of people ill with flu in 1890

Pfeiffer syndrome craniosynostoses as well as limb defects, mutations in FGFR1, broad thumbs, broad great toes

Phalen's maneuver in carpal tunnel syndrome, palmar flexion of the wrist to 90 degrees for 1 minute exacerbates or reproduces symptoms, paresthesias; 75% sens 47% spec

Philadelphia chromosome seen in 95% of chronic myelogenous leukemia (210 kD tyrosine kinase), 2-5% childhood ALL (180 kD TK), higher percent in adult; reciprocal and balanced translocation between chr22 (*bcr*, breakpoint cluster region) and chr9 (*c-abl*); *c-abl-bcr* encodes a chimeric protein with tyrosine kinase activity; genomic imprinting, chr9 paternal and chr22 maternal

Pick bodies in neurons, cytoplasmic, round to oval, filamentous inclusions that strongly stain with silver, weakly eosinophilic; composed of neurofilaments, vesiculated endoplasmic reticulum, and paired helical filaments that are immunocytochemically similar to those found in Alzheimer's; don't survive death of host neuron like they do in Alzheimer's

Pick cells characteristic swelling of neurons in Pick's disease

Pick's disease subtype of frontal lobe dementia, characterized by language abnormalities such as logorrhea, echolalia, and palilalia (compulsive repetition of phrases), Pick bodies, Pick cells; occurs 1-5% as often as Alzheimer's

Pickwickian syndrome obesity hypoventilation syndrome defined by extreme obesity and alveolar hypoventilation during wakefulness, characterized by hypersomnolence, dyspnea, hypoxemia (cyanosis, polycythemia, and plethora), and pulmonary hypertension leading to RV failure and edema; based upon Charles Dickens' book "The Posthumous Papers of the Pickwick Club" and the character Joe who was a "wonderfully fat boy, standing upright with his eyes closed"

Pierre Robin syndrome micrognathia and abnormal smallness of the tongue, often with cleft palate, severe myopia, congenital glaucoma, and retinal detachment; French pediatrician, 1867-1950

pink disease acrodynia, occurs from exposure to high concentrations of mercury vapor, characterized by a body rash, swelling and irritation of palms and feet followed by skin desquamation, irritability, photophobia, fever, insomnia and profuse sweating, which may also follow oral exposure to mercury compounds

Pisa syndrome pleurothotonus, rare side-effect of classic neuroleptic medication, characterized by dystonia with flexion of body and head to one side and axial rotation of the trunk; has been seen in cholinesterase inhibitors as well; originally described in 1972

Piskacek's sign asymmetry of the uterus with a well-defined soft prominence of the cornu, due to implantation near one of the cornua

Pittsburgh pneumonia agent *Legionella micdadei*

Plummer's disease toxic multinodular goiter

Plummer's nail onycholysis as a sign of hyperthyroidism, especially when it affects the ring finger

Plummer-Vinson syndrome from iron deficiency, a microcytic hypochromic anemia, atrophic glossitis, and esophageal webs (upper esophagus); 10% develop squamous cell carcinoma ; also known as Patterson-Brown-Kelly syndrome

POEMS syndrome polyneuropathy, organomegaly, endocrinopahty, M protein, and skin changes which may be seen in plasma cell dyscrasias; also known as Crow-Fukase syndrome in Japan

Poiseuille's law flow proportional to fourth power of radius, inversely proportional to length

Poland syndrome amastia associated with hypoplasia of ipsilateral musculature (microsyndactyly or lack of one hand) and chest wall (atrophy of the ipsilateral pectoralis major) and GU abnormalities; 10% have dextrocardia or dextroversion

Polle syndrome Munchausen syndrome by proxy; after daughter Polle from second marriage of Munchausen to 17 y.o. Bernhardine at age 74, daughter died 1 year later (though paternity apparently suspect) (*Lancet* 1977;2:456); but daughter Polle apparently doesn't exist but name of town where Bernhardine came from (*Pediatrics* 1984;74:554)

Pompe's disease type II glycogen storage disease, deficiency in alpha-1,4-glucosidase (lysosomal enzyme) with consequent accumulation of glycogen, especially in the liver, heart, and skeletal muscle, characterized by cardiomegaly, muscle hypotonia, and splenomegaly, death from cardiorespiratory failure before age 3

Poncet's disease in acute tuberculosis infection, aseptic inflammatory polyarthritis

Pontiac fever nonpneumonic legionellosis

Porter-Silber test measurement of urinary 17-OH corticosteroid

Posner-Schlossman syndrome glaucomatocyclitic crisis; recurrent episodes of markedly elevated intraocular pressure with mild idiopathic anterior chamber inflammation

Pott's disease tuberculous involvement of the spine; occurs in about 2% of TB cases

Pott's fracture fracture of distal fibula

Pott's puffy tumors extension of frontal sinusitis anteriorly into frontal bone causing a distinct swelling

Potter's sequence from oligohydramnios (from e.g. renal agenesis, amniotic leak) leading to amnion nodosum, fetal compression which leads to pulmonary hypoplasia, altered facies, positioning defects of feet, hands, and breech presentation

Potts shunt for treating tetralogy of Fallot, anastomosis of direct descending aorta to left pulmonary artery; now rarely performed

Poupart's ligament inguinal ligament

Prader-Willi syndrome deletion of 15q11-q13, paternally derived; uncontrollable hyperphagia after 12 months

Pratt's sign in deep vein thrombosis, presence of three dilated veins or sentinel veins over the tibia; dilatation persists when legs are elevated to 45 degrees

Prausnitz-Küstner reaction passive transfer of cutaneous anaphylaxis; transferring serum of affected patient intradermally into a recipient and then challenging recipient with antigen 24 hours later at the same site and checking for wheal and flare; known since 1920s

Prehn's sign elevation of painful testicle decreases pain of epididymitis

Preiser disease spontaneous, non-traumatic osteonecrosis of the scaphoid bone

Pringle maneuver clamping of porta hepatis to control hemorrhage

Prinzmetal's angina variant angina occurs at rest, manifests on EKG as episodic ST segment elevations, caused by coronary artery spasms with or without superimposed coronary artery disease; patients more likely to develop ventricular arrhythmias

Proteus syndrome congenital condition characterized by generalized, unilateral, or localized overgrowth of any tissue type, hemihypertrophy, lymphangiomas, lipomas, hemangiomata macrocephaly; thought that Joseph Merrick, the "Elephant Man" may have had this condition rather than neurofibromatosis; associated with mutation in *PTEN*

psammoma bodies papillary thyroid cancer

psoas sign pain elicited by extending the hip with the knee in full extension, seen with appendicitis and psoas inflammation

Puestow procedure for chronic pancreatitis, side-to-side anastomosis of the pancreas and jejunum, for decompressing dilated main pancreatic duct and providing pain relief

Pulfrich phenomenon misperception of trajectory of moving objects (an optical illusion that can be elicited in normal subjects), can be associated with optic neuritis

Purtilo's syndrome X-linked lymphoprolierative syndrome, results in pathologic response to EBV infection that may result in lymphoma

Purtscher's angiopathic retinopathy in acute pancreatitis, sudden and severe loss of vision due to posterior retinal artery occlusion with aggregated granulocytes; discrete flame-shaped hemorrhages with cotton-wool spots; also seen in fat embolization; first described in 1919 by Othmar Purtscher

Quebec platelet disorder from excessive production of u-PA (urine plasminogen activator) within alpha granules of platelets; autosomal dominant; characterized by mild thrombocytopenia and moderately severe bleeding 12-24 hours after surgery or trauma; also known as factor V Quebec (as 20% of factor V carried within alpha granules of platelets; u-PA actvates plasminogen, degrades intraplatelet stores of factor V)

Queckenstedt's maneuver applying pressure on the internal jugular vein to dilate cranial veins and increase incranial pressure; can be used to see if there's block in CSF flow by at lumbar puncture pressures in response

Queen Anne's sign in hypothyroidism, sparse eyebrows laterally; apparently it was fashionable to shave the lateral third of the eyebrow during the reign of Queen Anne (1707-1714) in Great Britain

Quellung reaction swelling of bacterial capsule when exposed to antibody; used for diagnosis of *S. pneumoniae*, *H. influ* type B, *N. meningitidis* groups A and C

Queyrat, erythroplasia of carcinoma in situ or invasive squamous cell cancer of the penile glans described by Auguste Queyrat, French dermatologist, born 1872

Quilty lesion endocardial infiltrates; associated with cyclosporine and waxing and waning levels of immunosuppresion; of unclear clinical significance; named after patient with this lesion

Quincke's disease angioedema

Quincke's sign in aortic regurgitation, capillary pulsations detected by pressing a glass slide on the patient's lip or by transmitting a light through the patient's fingertips; of questionable utility since seen in normal people

Rabson-Mendenhall syndrome congenital syndrome characterized by insulin resistance, acanthosis nigricans, and growth retardation; associated with developmental abnormalities of bones and teeth, PCOD, genitomegaly, and pineal gland hyperplasia; associated with mutation in insulin receptor

Raeder's syndrome also Raeder's paratrigeminal neuralgia, condition characterized by severe, unilateral headache, facial pain in distribution of ophthalmic division of trigeminal nerve combined with ipsilateral Horner's syndrome; also associated with nasal stuffiness or rhinorrhea; felt to reflect pathology in location where oculosympathetic fibers exit the internal carotid artery to join the ophthalmic division of the trigeminal nerve; first described in 1918 by George Raeder, Norwegian neurologist

Raisinghani sign paradoxical motion of the infero-posterior left ventricular wall in patients with liver disease and high intraabdominal pressures from hepatomegaly, ascites, splenomegaly or a combination of these

Ramirez sign in deep vein thrombosis, sphygmomanometer cuff placed above knee inflated to 40 mm Hg causing pain at site of thrombosis

Ramsay Hunt syndrome	herpes zoster infection of the geniculate ganglion; facial nerve involvement (ear, palate, pharynx, or neck); pain and vesicles appear in external auditory canal along with hyperacusia, and patients lose their sense of taste in anterior 2/3 of tongue while developing ipsilateral facial palsy; described by James Ramsay Hunt, American neurologist (1872-1937)
Randall disease	monoclonal light chain deposition disease; distinct from amyloidosis; associated with kappa light chains
Randle cycle	glucose-free fatty acid cycle; inverse relationship between glucose and free fatty acid use
Ranke complex	combination of Ghon lesion and involved lymph nodes in tuberculosis
Ransons' criteria	for evaluating acute pancreatitis at presentation, age>55; WBC>16,000; glucose>200; AST>250; LDH>350. during initial 48 hrs, base deficit>4; BUN increase>5; fluid sequestration>6L; Ca<8; Hct decrease>10; pO2<60. mortality 0-2, <5%; 3-4, 15%; 5-6, 40%; 7-8, 100%
Rapoport-Luebering shunt	in red blood cells, pathway converting 1,3-diphosphoglyceric acid to 2,3-DPG and then to 3-phosphoglyceric acid; enzyme is diphosphoglycerate synthetase; 2,3-DPG reduces affinity of hemoglobin for oxygen; 2,3-DPG rises with alkalosis and decreases with acidosis, result of effect of pH on enzyme
Rapunzel syndrome	small bowel obstruction by trichobezoar, named after Rapunzel, the German princess in Grimms' fairytales who let her golden hair down from her tower to facilitate a tryst with her lover
Rashkind's atrial septostomy	for treating transposition of great arteries, tricuspid atresia, and mitral atresia; catheter-based creation of atrial septal defect via foramen ovale
Rasmussen's aneurysm	aneurysm of the PA or pulmonary arteriole within or adjacent to a TB cavity
Rasmussen's encephalitis	progressive childhood disease characterized by severe epilepsy, hemiplegia, dementia, and inflammation of the brain potentially from autoantibodies to GluR3 antigen
Rastelli procedure	for treating transposition of great vessels with pulmonary stenosis, patching ventricular septal defect such that LV outflow passes through VSD into aorta, and a valved conduit or graft is placed between the RV and pulmonary arteries
Rathke's pouch	a divertic involved in development of pituitary gland, vestigial remnants lead to craniopharyngioma
Raynaud's phenomenon	exaggerated vascular response to cold temperatures or emotional stress, manifested by symmetrical, sharply demarcated color changes of the skin of the digits due to abnormal vasoconstriction of digital arteries and cutaneous arterioles
Rebuck skin window	dermal abrasion technique for testing tissue penetration of neutrophils, scraping forearm, then putting coverslip over it, checking glass for neutrophils
Reed-Sternberg cells	in Hodgkin's lymphoma, giant macrophage-like cells with two nuclei
Refsum's disease	autosomal recessive; phytanic acid accumulates as a result of an absence of the enzyme alpha-phytanic acid alpha-hydroxylase involved in its catabolism; give patient's large amounts of nicotinic acid or triparanol, chemicals that inhibit lipid synthesis; dryness and scaling similar to the appearance of icthyosis develop, associated with neuropathy
Reichert's cartilage	branchial arch 2
Reid index	comparing the relative thickness of the mucous glands with the total thickness of the airway wall; increased in patients with chronic bronchitis (normally < 0.4)
Reifenstein syndrome	partial androgen insensitivity; from mutation in androgen receptor gene; male pseudohermaphroditism characterized by hypospadias, hypogonadism, gynecomastia, normal XY karyotpe, X-linked recessive
Reinke crystals	in 25% of Leydig cell tumors, intracytoplasmic rod-shaped crystalloids
Reinke's edema	vocal cord polyposis in female smokers, 50s-70s
Reitan trail test	timed connect-the-number test for detecing alterations in mental status; time >60 s pathologic in all age groups
Reiter's syndrome	triad of arthritis, urethritis, and conjunctivitis described by Reiter in 1916 (Nazi sympathizer); 80% possess HLA-B27; associated with *Shigella flexneri*; ReA develops in 20% of exposed B27+ individuals; subset of reactive arthritis; triad present in 1/3 of patients; incidence estimated as 3.5/100K in males under age of 50; balanitis circinata (penis) and keratoderma blenorrhagica
Renshaw cells	inhibitory cells in the ventral horn of the spinal cord
Rett syndrome	childhood neurodevelopmental disorder almost exclusively affecting girls who develop normally for the first few months of life before undergoing a period of regression with loss of purposeful hand use and speech. Patients develop stereotypic hand-wringing movements, with ataxia and episodes of hyperventilation; from mutation in MECP2 on X chr which binds to single methylated CpG base pairs and "silences" other genes; Rett syndrome thought to arise because of excessive transcriptional noise due to failure of gene silencing by MECP2
Retzius, space of	the preperitoneal space anterior to the bladder
Retzius, veins of	numerous small veins in the retroperitoneum that connect the retroperitoneal viscera to the posterior abdominal wall; dilated in portal hypertension
Reye's syndrome	rare disease characterized by fatty change in liver and encephalopathy that in its most severe forms may be fatal; associated with VZV and influenza virus B in children given aspirin
Reynold's pentad	Charcot's triad plus altered mental status and shock in cholangitis

Rich foci	small subpial or subependymal foci of metastatic tuberculosis lesions in meninges or brain parenchyma
Richter syndrome	the evolution of chronic lymphocytic leukemia to a large cell lymphoma with high fever, weight loss, enlarging lymph nodes, and hepatosplenomegaly
Richter's hernia	incarcerated or strangulated hernia involving only one sidewall of the bowel, which can spontaneously reduce, resulting in gangrenous bowel and perforation within the abdomen without signs of obstruction
Riddoch's sign	in cerebellar disease, with outstretched hands, the hand on the affected side begins to hyperpronate, so that the palm faces outward, and rises above the level of the other hand
Riedel's lobe	in some persons, especially those with a lanky build, the liver tends to be somewhat elongated so that its right lobe is easily palpable as it projects downward toward the iliac crest; such elongation, called Riedel's lobe
Riedel's thyroiditis	unknown etiology, marked by glandular atrophy, hypothyroidism, and replacement of the thyroid by fibrous tissue with adhesion to surrounding structures
Riesman's sign	bruit over closed eyes in thyrotoxicosis
Riggler's sign	see bowel wall in perforation on plain films
Riley-Day syndrome	hereditary sensory and autonomic neuropathy type III (familial dysautonomia), recessive disorder that commences in infancy and is characterized by conspicuous autonomic dysfunction (absent tearing, labile temperature, and blood pressure), and accompanied by absent taste sensation, absent fungiform papillae on tongue, impaired pain and temperature sensation, and areflexia; occurs among Ashkenazi; associated with mutation in *IKBKAP* gene
Rinne test	vibrating tuning fork placed against mastoid and then next to ear; in sensorineural loss, air conduction is better than bone conduction; in conduct loss, BC>AC
Ritgen maneuver	for delivery of the fetal head by pressing with the tips of the fingers upon the perineum via the anus; alternatively, protection of the perineum by providing support of the perineum with pressure over the coccygeal area
Ritter's disease	*Staph* scaled skin syndrome
Roberts syndrome	autosomal recessive syndrome characterized by absence of leg bones, hypoplastic arms, bilateral cleft lip and cleft palate, prominent eyes
Robertsonian translocation	joining of two acrocentric chromosomes at the centromeres with loss of their short arms to form a single abnormal chromosome; acrocentric chromosomes are the Y chromosome and chromosome numbers 13, 14, 15, 21, and 22
Robinow's syndrome	short stature syndrome, mesomelic shortening, hemivertebrae, genital hypoplasia, and "fetal facies"
Rocky Mountain spotted fever	rickettsial illness caused by *Rickettsia rickettsii*, spread to human by ixodid ticks; characterized by sudden onset o fever, headache, myalgias, purpura. First recognized in 1896 in Snake River Valley of Idaho, name is a misnomer as it occurs throughout the U.S.
Roger's disease	small congenital VSD defect <0.5 cm in diameter (most are muscular); Henri L. Roger, French physician, 1809-1891
Rokitansky-Aschoff sinuses	small outpouchings of the gallbladder mucosa that may penetrate into and through the muscle wall; their prominence in inflammation and gallstone formation (e.g. chronic cholecystitis) suggests that they are acquired herniations
Romaña's sign	in the first week of Chagas disease, unilateral periorbital edema and swelling of the eyelid associated with reduviid bug of eye
Romano-Ward syndrome	long QT syndrome without deafness, inherited as autosomal dominant
Romberg test	patient stands feet together, eyes open and then closes both eyes for 20 to 30 sec without support; positive test with eyes open suggestive of cerebellar ataxia; with eyes closed suggestive of impaired proprioception
Roos sign	in thoracic outlet syndrome, abduction of the shoulders to 90 degrees, flexion of the elbows to 90 degrees, and opening and closing the hands slowly for 3 minutes causing hand pallor, ulnar dysesthesias, diminished pulse
Rosai-Dorfman disease	sinus histiocytosis with massive lymphadenopathy (mainly neck), fever, polyclonal hypergammaglobulinemia; extranodal sites involved in 43%; first described in 1969
Rosenbach's sign	in aortic regurgitation, hepatic pulsations
Rosenbach's sign	in thyrotoxicosis, tremor of the closed eyelids
Rosenmuller fossa	a pharyngeal fossa behind the Eustachian orifice where nasopharyngeal carcinoma most commonly occurs; named after Italian anatomist
Rosenthal fibers	inclusions that develop in astrocytes in chronic reactive and neoplastic proliferations; abundant in Alexander's disease
Rosenthal syndrome	factor XI deficiency; described by Rosenthal in 1953; also referred to as plasma thromboplastin antecedent deficiency; predominantly in patients of Ashkenazi background
Ross procedure	aortic valve replacement involving translocation of native pulmonary valve into aortic position with coronary artery relocation followed by reconstructing RV outflow tract with pulmonary homograft
Ross's syndrome	tonic pupils (generally bilateral), anhydrosis, and areflexia which may appear in a different pattern distribution; possible link with Holmes-Adie syndrome
Roth's spots	in bacterial endocarditis and other retinal hemorrhagic conditions, a round white spot surrounded by hemorrhage (secondary to

	microemboli in endocarditis)
Roth-Bielschowsky syndrome	internuclear ophthalmoplegia; caused by lesions in medial longitudinal fasiculus; bilateral lesions almost always due to multiple sclerosis; unilateral lesion often due to vascular occlusion
Rothmann-Makai syndrome	a variant of idiopathic lobular panniculitis seen in infants, tends to be localized and spontaneously resolves; associated with autoimmune diseases such as juvenile rheumatoid arthritis, diabetes, and Hashimoto's thyroiditis
Rothmund-Thomson syndrome	autosomal recessive disorder characterized by poikilodermatous skin changes that develop in infancy, premature aging, juvenile cataracts, sparse hair, short stature, skeletal defects, dystrophic nails, and predisposition for malignancies including osteosarcoma
Rotor's syndrome	poorly defined defects in hepatic uptake and storage of bilirubin; resembles Dubin-Johnson syndrome, but liver pigment missing
Rotter's lymph nodes	lymph nodes between the pectoralis minor and pectoralis major
Roussy-Levy syndrome	hereditary areflexic dystasia; phenotypic variant of Charcot-Marie-Tooth (CMT-1B) associated with postural tremor and ataxia; autosomal dominant inheritance
Rovsing's sign	pain in the right lower quadrant during left-sided pressure suggests appendicitis; so does right lower quadrant pain on quick withdrawal (referred rebound tenderness)
RSH syndrome	see Smith-Lemli-Opitz syndrome
Rubinstein-Taybi syndrome	congenital condition characterized by mental and growth retardation, short broad thumbs and/or halluces, and typical facial features
Ruffini's corpuscles	sensory receptors for heat; not lamellated
Rumpel-Leede sign	test for capillary fragility carried out by incresing venous pressure in forearm with BP cuff and then inspecting the skin for petechial eruptions. Also called Hess test; associated with scurvy
Russell bodies	endoplasmic reticulum of plasma cells engaged in active synthesis of immunoglobulins may become hugely distended, producing large, homogenous eosinophilic inclusions called Russell bodies; also seen in Waldenstöm macroglobulinemia
Russell viper venom time	sensitive screening test for lupus anticoagulant activity; directly activates factor X to Xa; from the venom of *Vipera russelli*
Russell's sign	lanugo, dry skin, hand calluses, associated with purging and bulimia
Ruvalcaba-Myhre-Smith syndrome	association of macrocephaly, intestinal polyposis, and pigmentation of the penis; related to Bannayan-Zonana syndrome
Sabin-Feldman dye test	serum titer rises in toxoplasma infection
Sabouraud's agar	for growing fungi, low pH of medium and chloramphenicol and cycloheximide
Saethre-Chotzen syndrome	common craniosynostosis syndrome characterized by craniofacial and limb anomalies; associated with mutation in TWIST transcription gene
Sailer's sign	in aortic regurgitation, pulsation of spleen in the setting of splenomegaly
Saint's triad	the coexistence of hiatal hernia, gallbladder disease, and diverticulosis in a patient; named after Saint, a South African surgeon; no pathophysiologic basis for the coexistence of all three processes, emphasizing that more than one disease process may be responsible for a patient's symptoms
Salisbury effect	reduction in left ventricular diastolic distensibility with increase in coronary flow and perfusion pressure
Salus's sign	deflection of veins at AV crossings in hypertensive retinopathy (grade 2); see also Bonnet's and Gunn's sign
Sampson's artery	artery found within and nourishes round ligament of uteru
Samter's triad	nasal polyps, bronchial asthma, aspirin sensitivity
San Joaquin valley fever	coccidiomycosis (from infection with Coccidiomycosis imites)
Sandhoff's disease	GM2 gangliosidosis, from mutation in beta subunit of both hexosaminidase A and B; clinically similar as Tay-Sachs but more rapid progression of disease
Sandifer syndrome	association of torsional dystonia, mainly involving the neck and upper extremities, with either esophageal reflux or hiatus hernia; presents in childhood; at times mistaken for seizures
Sanfilippo's syndrome	mucopolysaccharidosis, four types (A-D) from deficiency of enzymatic steps necessary for removal of N-sulfated or N-acetylated glucosamine residues from heparan sulfate, leading to mental retardation, CNS disorders, coarse facies
Santavuori-Haltia disease	infantile neuronal ceroid lipofuscinosis, CLN1
Santorini, duct of	dorsal duct of pancreas, embryologically, the biggest duct, clinically the smaller pancreatic duct
Santorini's fissures	fissures in anterior aspect of cartilaginous portion of ear canal through which neurovascular tissues pass; may allow spread of ear

	disease to parotid joint, TMJ, soft tissues of upper neck
Santos's syndrome	Hirschsprung's disease with renal agenesis, polydactyly, hypertelorsim, and deafness
Sappey, veins of	retroperitoneal veins located around liver and diaphragm connecting epigastric and internal mammary veins with azygos and which become dilated in portal hypertension
Saturday night palsy	radial neuropathy from compression at the spiral groove
Saxon test	for diagnosing Sjogren's, measurement of whole mouth saliva production (analogous to Schirmer test for tears) by asking patients to chew a gauze sponge; normal is increase in weight of 2.75 g over 2 min
Scarpa's fasica	fascia around edge of subcutaneous inguinal ring
Schafer's maneuver	extensor plantar response by squeezing Achilles tendon suggesting upper motor neuron defect
Schamberg's disease	idiopathic capillaritis in which inflammation weakens capillaries, causes petechial lesions like cayenne pepper
Schamroth's window	normal diamond-shaped window formed by the nail bases when ends of opposing ends of fingers are placed together; not seen in clubbing
Schatzki's ring	esophageal rings and webs in lower esophagus, located at or just above the squamocolumnar junction; most common cause of intermittent solid food obstruction
Schaumann's bodies	laminated concretions composed of calcium and proteins, seen in granulomatous diseases (e.g. sarcoidosis)
Scheie's syndrome	mucopolysaccharidosis (I S), from alpha-L-iduronidase deficiency (same as Hurler's syndrome), milder than Hurler's syndrome, resulting in corneal clouding, stiff joints, aortic valve disease, normal intelligence and potentially normal life span
Scheuermann's disease	juvenile kyphosis characterized by end-plate irregularities and wedging of the vertebral body without loss of bone density, develops in adolescence
Schick's test	for testing immune status to *Cornyebacterium diphtheriae*, intradermal injection of 0.1 mL of purified standardized toxin; if no inflammation, antitoxin present
Schilder's disease	adrenoleukodystrophy, X-linked disorder, presents in childhood with weakness, spasticity, leading to dementia, blindness, and quadriparesis, associated with adrenal insufficiency; from defect in beta-oxidation of fatty acids in peroxisomes leading to accumulation of very long-chain saturated fatty acids
Schiller-Duval body	in endodermal sinus tumor (yolk sac tumor), tumor cells arranged around a capillary
Schilling test	first stage, measuring cobalamin (B12) absorption by determining how much p.o. radioactive cobalamin is excreted in urine over 24 to 48 hours (after i.m. inj inj B12 to saturate the transcobalamines and to "flush" orally absorbed radiolabeled B12 into the urine). Second stage, test repeated with oral intrinsic factor, should normalize B12 absorption in pernicious anemia but not in intestinal malabsorption
Schimke immuno-osseous dysplasia	autosomal-recessive pleiotropic disorder with the diagnostic features of spondyloepiphyseal dysplasia, renal dysfunction and T-cell immunodeficiency; associated with mutation in SMARCAL1 (SWI/SNF2-related, matrix-associated, actin-dependent regulator of chromatin, subfamily a-like 1)
Schirmer's test	measures quantity of tears secreted in 5 minutes in response to irritation from a filter strip placed under each lower eyelid; normal young person moistens 15 mm; 33% of elderly wet only 10 mm in 5 minutes; in Sjögren's syndrome, <5 mm in 5 min, 85% sensitivity, 85% specificity
Schlesinger's solution	morphine and scopolamine
Schmid metaphyseal chondrodysplasia	mutation in collagen X, mechanical pressure reduces growth
Schmidt's syndrome	type II polyglandular autoimmune syndrome, coexistent adrenal and thyroid disease, sometimes accompanied by diabetes
Schmorl's nodule	herniated nucleus pulposus looking like osteomyelitis
Schnitzler's syndrome	association of IgM monoclonal protein (e.g. as with Waldenström's macroglobulinemia) with erythematous, urticarial skin lesions
Schober test	measures distraction between 2 marks on the skin during forward flexion; distance decreased in ankylosing spondylitis
Schüffner's granules	pigment in trophozoite, seen in *Plasmodium vivax*
Schultz-Charlton phenomenon	was observed after intradermal inj of 0.1 mL of antitoxin into an area of scarlet fever rash that produced "blanching" at the site of injection within 12-24 h. The test has to be performed during the very early phase of the eruption before exudation into the lesion made skin changes irreversible.
Schwachman-Diamond syndrome	combination of neutropenia, metaphyseal dysplasia, and exocrine pancreatic insufficiency, associated with recurrent infections beginning in the first year of life in the sinuses, bones, lungs, skin and urinary tract, fatty replacement of liver; associated with increased risk of aplasia, myelodysplasia, and leukemia; life expectancy is 35 years
Schwartz's dictum	no acid, no ulcer
Schwartz-Bartter syndrome	SIADH
Scott syndrome	defect in primary homeostasis, prolonged PT, deficiency in platelet coagulant activity characterized by a failure to expose

	phosphatidylserine (PS) to the outer leaflet of the platelet plasma membrane
Seckel syndrome	rare autosomal recessive disorder characterized by "bird-headed" facial appearance, growth retardation, and microcephaly with mental retardation; associated with mutation in gene encoding ataxia-telangiectasia and RAD3-related protein (ATR).
second disease	scarlet fever, aka scarlatina, caused by *Strep pyogenes* exotoxin, first described in 1626
Segawa syndrome	hereditary progressive dystonia with marked diurnal fluctuaion from defect in GTP cyclohydrase I, cofactor for synthesis of dopamine; dystonia treatable with levodopa
Seldinger technique	guide wire-assisted vascular cannulation, i.e., small-bore needle first used to enter vessel, then wire passed through needle, needle removed, leaving wire in place for guiding cannulation of vessel, developed in 1953
Seligmann's disease	alpha heavy chain disease, characterized by infiltration of the lamina propria of the small intestine with lymphoplasmacytoid cells that secrete truncated alpha chains
Sellick's maneuver	cricoid pressure during intubation
Semmes-Weinstein nylon test	nylon monofilament developed in 1950s for sensation testing; can be used in diabetic foot screening with a 5.07 monofilament which delivers 10 g of force
Semont maneuver	in benign positional vertigo, a liberatory maneuver where the patient is moved rapidly from side to side; not favored in the U.S. (also see Epley maneuver and Brandt-Daroff exercises)
Senear-Usher syndrome	pemphigus erythematosus; a localized variety of pemphigus foliaceus confined to seborrheic sites
Sengstaken-Blakemore tube	a double-balloon system tube, one for stomach, one for esophagus, for tamponade of bleeding varices
Sennetsu fever	human ehrlichiosis from *Ehrlichia sennetsu*, generally not fatal, occurring mainly in Japan
Senning procedure	variant of Mustard procedure
Sertoli cell tumors	testicular tumor derived from the sex cord
Sever's disease	apophysitis or osteochondrosis of the calcaneus, common (but frequently source of heel pain), condition occurs before or during the peak growth spurt, often resolves two weeks or two months after initiation of conservative treatment
Sézary's syndrome	rare special variant of cutaneous T-cell lymphoma characterized by generalized or universal erythroderma, peripheral lymphadenopathy, and cellular infiltrates of atypical lymphocytes (Sézary cells) in the skin and blood
Sézary-Lutzner cells	found in cutaneous T-cell lymphoma, T-helper cells that characteristically form band-like aggregates within the superficial dermis and invade the epidermis as single cells and small clusters (Pautrier's microabcesses)
Shah-Waardenburg syndrome	see Waardenburg syndrome type IV
Shapiro's syndrome	agenesis of the corpus callosum associated with spontaneous recurrent hypothermia and hyperhidrosis
Sheehan's syndrome	postpartum pituitary necrosis, syndrome results from sudden infarction of the anterior lobe precipitated by obstetric hemorrhage or shock (pregnancy, pituitary enlarges to almost twice its normal size, compressing blood supply)
Shelley's sign	in aortic regurgitation, pulsation of the cervix (*J Indiana State Med Assoc* 1959;52:1283-89)
Shenton's line	a radiological line drawn between the medial border of the femoral neck and the superior border of the obturator foramen; interrupted in dislocations and fractures
Shiraz dwarfism	zinc deficiency, in Iran, short stature and aspermia
Shohl's solution	alkalinizing citrate solution in hyporeninemic hypoaldosteronism associated with renal tubular acidosis associated with diabetic nephropathy
Shone's complex	series of four obstructive or potentially obstructive left-sided cardiac lesions: supravalvular mitral ring, parachute deformity of mitral valve, subaortic stenosis, and coarctation of the aorta
shoshin syndrome	acute fulminant cardiac beriberi (in Japanese, *sho* is acute damage, *shin* is heart)
Shprintzen syndrome	velo-cardio-facial syndrome, associated with cleft palate, cardiac anomalies, typical facies, and learning disabilities; may result from mutation in same gene as DiGeorge syndrome given clinical overlap between the two syndromes
Shulman's syndrome	eosinophilic fasciitis, characterized by acute onset of erythema, swelling, induration of the extremities, and eosinophlia, often following exercise; epidermis and dermis normal
Shwachman's syndrome	pancreatic insufficiency and bone marrow dysfunction
Shwartzman reaction	two iv injections of sublethal lipopolysaccharide, 24 hrs apart, causing DIC in rabbits from widespread intravascular thrombus formation on surfaces of endothelial cells; TNF obligatory mediator
Shy-Drager syndrome	degenerative disorder characterized by parkinsonian features (leading to postural hypotension, anhidrosis, disturbance of sphincter control, impotence, etc.) and signs of more widespread neurologic involvement (pyramidal or lower motor neuron signs and often a cerebellar deficit)

Siegrist streaks	linear hyperpigmented areas over choroidal vessels in hypertensive retinopathy
Siewert syndrome	see Kartagener's sydnrome
silk glove sign	indirect hernia sac in the pediatric patient; the sac feels like a finger of a silk glove when rolled under the examining finger
Silk Road disease	see Behçet's disease
Silver-Russell syndrome	syndrome characterized by lateral asymmetry and low-birth-weight dwarfism
Simmond's disease	pituitary cachexia (e.g. from Sheehan's syndrome)
Simmond's test	for testing Achilles tendon, lack of plantar flexion after squeezing calf muscles suggests rupture of tendon; see Thompson's test
Simon focus	in tuberculosis, nodule in subapical region which develops from hematogenous spread from initial infection in lower half of lung
Sims-Huhner test	post-coital test, done 2-4 hrs after intercourse to assess number and motility of sperm that have entered the cervical canal
singers's nodule	is a small, benign laryngeal polyp, usually induced by chronic irritation, such as excessive use of the voice, and is associated most commonly with heavy cigarette smoking; is usually localized to the true vocal cords
Sipple's syndrome	MEN type IIa; pheochromacytoma, medullary carcinoma of the thyroid, and hyperparathyroidism due to hyperplasia or tumor
Sippy powders	for treating peptic ulcer disease in early 1900s, powder containing 600 mg of magnesium carbonate and 600 mg sodium bicarbonate alternating with a powder containing 600 mg of bismuth subcarbonate and 1200 to 1800 mg of sodium bicarbonate; associated with milk alkali syndrome
Sister Mary Joseph nodule	abdominal carcinoma, especially gastric, may metastasize to the navel; Sister Joseph, in the early days of the Mayo Clinic, noted periumbilical nodules in patients with intraabdominal cancer
sixth disease	roseola infantilis, also known as exanthem subitum, caused by HHV 6, described in 1910
Sjögren's syndrome	lymphocytic infiltration affects salivary and lacrimal glands and is associated with dry mouth and dry eyes (keratoconjunctivitis sicca); may have either interstitial pulmonary fibrosis or a lymphocytic infiltration of the alveolar walls (may have a malignant transformation with the development of a lymphoma)
Skene's glands	paraurethral glands in women
Skirrow's medium	contains vancomycin, trimethoprim, cephalothin, polymixin, and amphotericin B; for growing e.g. *Campylobacter*
Skoda's sign	with large pleural effusion or area of consolidation, increased resonance above effusion
Sluder's disease	lower-half headache, characterized by unilateral pain involving maxilla, teeth, ear, mastoid, and base of nose; tendency for long remissions and serial attacks
Sly's syndrome	mucopolysaccharidosis (VII), from beta-glucoronidase deficiency, resulting in hepatosplenomegaly, physical deformity from defect in degradation of dermatan sulfate and heparan sulfate
Smith's fracture	opposite of Colle's fracture; fracture of the distal radius, but from falling on the dorsum of the hand
Smith-Lemli-Opitz syndrome	autosomal recessive disorder in cholesterol synthesis caused by mutation in the sterol delta-7-reductase gene; characterized by constellation of congenital anomalies: microcephaly, mental retardation, hypotonia, incomplete development of the male genitalia, short nose with anteverted nostrils, failure of masculinization; also known as RSH syndrome after initials of surnames of three families with syndrome
Smith-Magenis syndrome	brachycephaly, growth retardation; associated with hair and nail pulling, reversed sleep pattern, and polyembolokoilamania (insertion of foreign bodies into body orifices); associated with deletion in 17p11.2
Sneddon's syndrome	triad of livedo reticularis, stroke-like episodes, and hypertension; associated with antiphospholipid antibodies
Sneddon-Wilkinson disease	subcorneal pustular dermatosis; rare, chronic , recurrent pustular eruption characterized by subcorneal pustules that contain abundant neutrophils
Somogyi phenomenon	rebound hyperglycemia following an episode of hypoglycemia due to counterregulatory hormone release
Soret band	absorption band for heme
Sotos syndrome	cerebral gigantism; associated with mutation in *NSD1*
Spanish flu	1918 influenza with 20-100 million deaths worldwide with 2.5% mortality (influenza normally 0.1% mortality)
Spigelian hernia	hernia through the linea semilunaris, aka spontaneous lateral ventral hernia
Sprengel scapula	congenital elevation of the scapula
Spurling's test	for diagnosing cervical radiculopathy, exerting downward pressure on the head while rotating the head towards the symptomatic side creating pain radiating into affected extremity
St. Anthony's fire	erysipelas; also refers to ergotism
St. Anthony's fire	chronic ergotism; disease caused by excess ergot alkaloid; classically an epidemic in the Middle Ages caused by consumption of rye contaminated by the ergot fungus, leading to chronic ergotism characterized by intense burning pain and gangrene of feet, hands, and whole limbs due to vasconstrictive properties of ergot; acute ergotism characterized by manic episodes and hallucinations due to

	serotonin antagonism of ergot related to LSD; also refers to erysipelas
St. Jude valve	prosthetic valve, bileaflet, with two semicircular discs that pivot between open and closed positions without supporting struts, first used in 1977
St. Vitus's dance	see Sydenham's chorea
Starr-Edwards valve	ball and cage valve, oldest prosthetic valve in continuous use, first used in 1965
Stauffer's syndrome	elevation of LFTs due to cholestasis in renal cell carcinoma
Steele-Richardson-Olszewski syndrome	aka progressive supranuclear palsy
Steinert's disease	myotonic muscular dystrophy
Stein-Leventhal syndrome	polycystic ovarian disease
Stellwag's sign	incomplete and infrequent blinking in Graves's disease
Stensen's duct	parotid duct, enters oral cavity opposite crown of second maxillary molar tooth
Stevens-Johnson syndrome	extensive and symptomatic febrile form of erythema multiforme, more common in children; 1-6 cases/million person-years
Stewart's granuloma	lethal midline granuloma, T-cell lymphoma that gradually ulcerates nose and mid-face cartilage and bone
Stewart-Treves syndrome	lymphedema following mastectomy leading to lymphangiosarcoma
Stickler syndrome	mild spondyloepiphyseal dysplasia, osteoarthritis, and sensorineural hearing loss, some forms associated with a dominant negative mutation in the human COL11A2 gene encoding the alpha 2(XI) chain; another form of Stickler syndrome from mutation in COL2A1
stiff-man syndrome	chronic disorder of involuntary stiffness, painful muscle spasms, and rigidity, predominantly in the axial muscles; often occurs in conjunction with autoimmune diseases, particularly type I diabetes, associated with circulating antibodies to glutamic acid decarboxylase
Still's disease, adult onset	polyarthritis associated with sudden onset of high spiking fever, sore throat, and an evanescent erythematous salmon-colored rash
Still's murmur	described by George Still in 1909; normal vibratory midsystolic murmur; innocent murmur
Stockholm syndrome	victims sympathizing with victimizer, e.g., kidnapper; described after incident in summer of 1973 in Stockholm where hostages were taken in a failed bank robbery and after end of their captivity in six days, the hostages resisted rescue
Stokes collar	in superior vena caval obstruction, enlargement of neck with non-pitting edema
Stokes-Adams attacks	syncope associated with complete heart block (or other types of bradycardia)
Stoppa repair	hernia repair using tension-free intraparietal prosthetic mesh
Stransky's sign	involuntary dorsiflexion of the toes after firmly abducting the 5th digit for 2 seconds, and then acutely letting it go in upper motor neuron defects
Stroop test	stress test, also used for checking frontal function, where patients are tested for the ability to read off the text color of text spelling potentially different colors
Strümpell's sign	involuntary pronation of the forearm/wrist with passive extension and flexion of the arm at the elbow in upper motor neuron defect
Stuart factor	factor X; also known as Stuart-Prower factor. Named after patients Audrey Prower and Rufus Stuart
Stuart-Prower factor	see Stuart factor
Sturge-Weber syndrome	association of port-wine stain with vascular malformations in the eye (glaucoma) and leptomeninges and superficial calcifications of the brain; attributed to faulty development of certain mesodermal and ectodermal elements, and associated with mental retardation, seizures, hemiplegia, and radiopacities in the skull
Sudeck's point	watershed region between sigmoid and superior rectal artery in ileocecal region; see also Griffth's point
Sudeck's syndrome	reflex sympathetic dystrophy syndrome occurring in older people characterized by cystic changes and subchondral erosion in bone, diffuse osteoporosis, and muscle atrophy, but not necessarily associated with trauma
Sugiura procedure	for treating esophageal varices, esophageal and gastric devascularization, esophageal transection
Susac syndrome	microangiopathy of the inner ear, retina, and brain leading to deafness, retinal artery occlusion, and encephalopathy
Sutton's disease	major aphthous stomatitis; recurrent aphthous stomatitis with large deep ulcers >1 cm, may leave scarring; unknown etiology
Swedish porphyria	acute intermittent porphyria
Sweet's syndrome	acute febrile neutrophilic dermatosis or Sweet syndrome, initially described in 1964 by Robert Sweet. It is characterized fever, neutrophilic leucocytosis, abrupt appearance of erythematous, painful, cutaneous plaques and dense dermal infiltrate consisting of

	mature neutrophils without vasculitis signs.
Swyer's syndrome	XY gonadal dysgenesis
Swyer-James syndrome	in pediatrics, acquired hypoplastic lung that develops after severe obliterative bronchiolitis associated with bronchiolar obstruction, bronchiectasis, and distal air-space destruction
Sydenham's chorea	Postinfectious chorea appearing several months after a streptococcal infection with subsequent rheumatic fever; chorea typically involves the distal limbs and is associated with hypotonia and emotional lability; improvement occurs over weeks or months and exacerbations occur without associated infection occurrence; also known as St. Vitus's dance
Sylvian aqueduct	cerebral aqueduct of the midbrain that connects the third and fourth ventricle
Syme's amputation	amputation at the ankle with removal of the malleoli and placement of the heel pad over the end of the remaining tibia; devised in 1842
Takayasu's disease	aortic arch syndrome, pulseless disease; panartertis of the great vessels that's most common in Asian women
takotsubo syndrome	apical ballooning syndrome, characterized by acute onset of chest pain, EKG changes, cardiac marker release and transient extensive akinesia of the apical portions of left ventricle without significant coronary stenosis.
Tamm-Horsfall protein	uromodulin, major component of renal casts; 30-50 mg secreted per day by cells in thick ascending limb; homologous to GP2, a protein secreted from the acinar cell and a major component of plugs in noncalcific chronic pancreatitis
Tangier disease	rare recessive disease characterized by enlarged orange tonsils, peripheral neuropathy affecting small fibers involved in pain and temperature, and a near-complete absence of HDL-cholesterol; ABC1, the ATP binding-cassette transporter 1 gene, mutated in Tagier disease; gene mediates efflux of cholesterol from cells; named after small island in Chesapeake Bay where disease first identified in five-year-old boy
Tanner stages	stages of sexual development in puberty; described by James Tanner
Tarlov cyst	perineural cyst found in the lower spinal cord
Tarui disease	type VII glyocgen storage disease, deficiency of muscle phophofructokinase, presents as early onset of fatigue and pain with exercise, resulting in myoglobinuria
Tay-Sachs disease	autosomal recessive sphingolipidosis; GM2 gangliosidosis, results from mutations that affect alpha subunit of hexosaminidase A gene and cause a severe deficiency in hexosaminidase A; blindness and cherry-red spot (see also Sandhoff's disease with similar symptoms; involves beta subunit of both hexosaminidase A and B); previously called "amaurotic idiocy"
Terry's nails	mostly whitish with a distal band of reddish brown; may be seen with aging and in people with chronic diseases such as cirrhosis of the liver, congestive heart failure, and non-insulin-dependent diabetes; seen in 10% of uremics
Terson's syndrome	intra-vitreous hemorrhage associated with subarachnoid hemorrhage
third disease	German measles, aka rubella or *rötheln*, caused by rubivirus, described in 1881
Thompson's test	verifies if gastroc-soleus complex intact; squeeze calf belly, foot should plantar flex; see also Simmond's test
Thomsen's disease	myotonia congenita, autosomal dominant
Thomsen-Friedenreich antigen	cryptic antigen in membranes of erythrocytes, platelets, and glomerular capillary endothelial cells exposed by *S. pneumoniae*-derived neuraminidase removal of sialic acid
Thorel's pathway	posterior internodal tract in atrial conduction system
Throckmorton sign	positive sign when penis points towards lesion seen radiographically; see also John Thomas sign
thumb sign	in Marfan's disease, Ehlers-Danlos syndrome, and similar syndromes, thumb protrudes from clenched fist
Tietze syndrome	discomfort localized in swelling of the costochondral and costosternal joints, which are painful on palpation; may be perceived as breast pain
Tinel's sign	a sensation of tingling or pins and needles felt in distal extremity when percussion is made over the site of an injured nerve; it indicates a partial lesion or early regeneration of the nerve; 60% sens, 67% spec
Todd's paralysis	transient hemiparesis in postictal period, resolves over a period of 0.5-36 hours, suggests focal brain lesion as cause
Toldt, white lines of	the peritoneal reflections of the ascending and descending colon
Tolosa-Hunt syndrome	idiopathic inflammation of the cavernous sinus producing painful palsy of third, fourth, or sixth cranial nerve
Tornwaldt's bursa	pharyngeal bursa, located above the pharyngeal tonsil in the midline of the posteror wall of the nasopharynx; remnant of embryonic communication between the anterior tip of the notochord and the roof of the pharynx
Tourette's syndrome	chronic multiple motor and verbal tics, symptoms typically begin before 21 y.o., first signs motor tics in 80% and vocal tics in 20% but eventually both motor and vocal; higher than expected number of left-handedness and ambidexterity; 40-50% involve self-mutilation such as nail-biting, hair-pulling, etc.
Towne's view	AP view with the X ray tube angled caudad to show the occipital bone
TRALI syndrome	Transfusion Related Acute Lung Injury; transfusion reaction occurring within 6 hours after transfusion of blood product, characterized by pulmonary edema; due to anti-granulocyte antibodies in donor's plasma causing pulmonary sequestration of recipient

	leukocytes in susceptible patient
Traube's sign	in aortic regurgitation "pistol shot sounds" referring to booming systolic and diastolic sounds over the femoral artery
Traube's space	a crescentic space about 12 cm wide, bounded medially by the left sternal border, above by an oblique line from 6th costal cartilage to the lower border of the 8th or 9th rib in the mid-axillary line and below by costal margin; usually tympanitic because of stomach but can be affected by emphysema, pleural effusion, or splenomegaly
Treacher Collins syndrome	first arch syndrome, mandibulofacial dysplasia, caused by autosomal dominant gene, resulting in malar hypoplasia with down-slanting palpebral fissures, defects in the lower eyelids, deformed external ears, and sometimes abnormalities of the middle and internal ears
Trendelenburg position	position where angle of the head of bed or table is inclined at 45 degrees down; used in surgery to push the abdominal organs towards the chest
Trendelenburg's gait	waddling gait in people with weakness or paresis of gluteal muscles; seen in progressive muscular dystrophy
Trendelenburg's sign	sign of weakness of gluteus medius muscle seen when standing on one leg, failure to elevate contralateral side of pelvis; may be due to congenital dislocation of hip, trochanteric fracture, polio, spinal nerve root lesions with muscular atrophy
Trietz, ligament of	the suspensory muscle of the duodenum which supports the duodenojejunal flexure
Trotter's syndrome	triad of unilateral deafness due to Eustachian tube involvement, neuralgia affecting branches of the trigeminal nerve, and defective mobility of the soft palate due to nasopharyngeal cancer
Trousseau's sign	in hypocalcemia and latent tetany, carpal spasm induced by occluding the brachial artery for 3 min with an inflated BP cuff
Trousseau's syndrome	migratory thrombophlebitis, may be encountered with deep-seated cancers, most often with carcinomas of the pancreas or lung
Tullio's phenomenon	induction of vertigo by loud noises, i.e. sound-induced vestibular activation; associated with and previously considered pathognominc of syphilis
Turcot's syndrome	rare variant of familial adenomatous polyposis, with combination of adenomatous colonic polyposis and tumors of the CNS, mostly glioblastoma multiforme
Turner's syndrome	females with XO, short, low-set ears, shield chest, congenital heart defect (usually coarctation), café-au-lait spots, freckles, webbed neck, lymphedema
Tzanck smear	cytologic technique most often used in the diagnosis of herpesvirus infections (simplex or varicella-zoster); multinucleated giant cells suggest the presence of herpes; named after Arnault Tzanck
Uhl's anomaly	paper thin parietal myocardium, usually but not always limited to right ventricle, presents as heart failure in infancy or early childhood
Uhthoff's phenomenon	in multiple sclerosis, sensitivity of symptoms to changes in body temperature or exercise (e.g., visual loss with exercise); initial description in 1890 amblyopia following exercise
unhappy triad	lateral knee injury resulting in ACL tear, MCL tear, and medial meniscal injury
Unna boot	compression dressing impregnated with medication for treating venous stasis ulcers; named after Paul Gerson Unna, German dermatologist 1850-1929
Unterberger's stepping test	for assessing vestibular function, having patient step in one spot with the eye closed; in peripheral lesions, body rotates to side of lesion whereas in central disorders, deviation is irregular
Unverricht-Lundborg disease	progressive myoclonus epilepsy (EPM1), onset at age 6-15, stimulus-sensitive myoclonus, tonic-clonic seizures, marked sensitivity to photic stimulation, leads to cerebellar ataxia and mental deterioration; from unstable expansion of a dodecamer minisatellite repeat unit in the promoter region of cystatin B, a widely expressed cysteine protease inhibitor; more common in Finland and western Mediterranean; also known as Baltic myoclonus
Upshaw-Schulman syndrome	congenital thrombotic thrombocytopenic purpura (TTP); caused by an inherited deficiency in von Willebrand factor cleaving metalloprotease (ADAMTS13), characterized by the constellation of findings seen in TTP, namely microangiopathic hemolytic anemia, thrombocytopenia, neurologic symptoms, renal dysfunction, and fever.
Urbach-Wiethe disease	lipoid proteinosis with cutaneous and mucosal infiltrations of eyelids, tongue, larynx; associated with early hoarseness with an unusual skin eruption; autosomal recessiave; associated with mutation in *ECM1* gene
Usher's syndrome	type I, profound bilateral sensorineural congenital deafness with onset of loss of vision due to retinitis pigmentosa by age 10 (type 1B due to mutation in myosin-VIIa); type 2, moderate to severe congenital deafness with onset of retinitis pigmentosa by age 10; type III, retinitis pigmentosa first noted at puberty with progressive hearing loss; type IV, possible X-linked form
VACTERL association	vertebral, anal, cardiovascular, tracheoesophageal, renal, and limb defects
Valentino's syndrome	duodenal ulcer with retroperitoneal perforation presenting with pain in the right lower quadrant
Valsalva maneuver	first described in 1704 as a method for expelling pus from the middle ear: deep inspiration followed by forced exhalation against a closed glottis for 10-12 seconds; 4 phases, phase 1 transient rise in BP with straining; phase 2 decrease in systemic venous return, blood pressure, and reflex tachycardia; phase 3 begins with cessation of straining, associated with abrupt transient decrease in blood pressure and in systemic venous return (generally not perceivable); phase 4 an overshoot of systemic arterial pressure and relatively obvious reflex bradcycardia associated
Van den Bergh reaction	used to distinguish between unconjugated and conjugated bilirubin; bilirubin pigments are exposed to sulfanilic acid to generate diazo conjugates, forming chromogenic products

Van der Woude syndrome	an AD condition in which lip pits are seen in all gene carriers but only some individuals have cleft lips with or without cleft palate owing to variable expressivity
Van Wyk-Grumbach syndrome	primary hypothyroidism associated with precocious puberty and galactorrhea
Van't Hoff's law	for calculating osmotic pressure
Vanek tumor	benign inflammatory fibroid polyp seen in gastrointestinal tract
Vater, ampulla	location where common bile duct enters the duodenum; described by German anatomist Vater (1684-1751)
Vaughn-Jackson lesion	seen in rheumatoid arthritis, rupture of tendons in distal ulnar head, associated with loss of extension in the little and ring, finger; associated with caput ulna syndrome
Verner-Morrison syndrome	secretory diarrhea associated with VIPoma; characterized by watery diarrhea, hypokalemia, achlorhydria
Vespignani sign	edema of the ureteral vesical junction in renal colic
Vietnamese time bomb	*Burkholderia pseudomallei*, causative agent of melioidosis, described as "time bomb" because of its occurrence in war veterans more than two decades after their return from Vietnam
Villaret's syndrome	involvement of four lower cranial nerves (IX-XII) clinical manifestations manifesting as Horner's syndrome and paralysis of the soft palate, pharynx, and vocal cords; associated with tumor in posterior retroparotid space
Vincent's infection	acute necrotizing ulcerative gingivitis, occurring in patients with decreased resistance to infection, from concurrent infection with the symbiotic bacteria *Fusobacterium fusiforme* and *Borrelia vincentii*
Virchow's node	supraclavicular adenopathy associated with a malignancy, often on left side, associated with stomach cancer among other neoplasms but also GI and pelvic malignancies in general. First described by Virchow in 1848, more cases added by Troisier in 1886; referred to as Troisier's node in France
Virchow's triad	predisposing factors in thrombus formation 1. endothelial injury, 2. hypercoagulability 3. stasis or turbulence of blood flow; first described in 1860
Virchow-Robin spaces	perivascular spaces in brain; become unusually widened in edema of the brain
Vogt's triad	in tuberous sclerosis, triad of seizures, mental retardation, and facial angiofibromas. Occurs in fewer than 50% of patients with tuberous sclerosis
Vogt-Koyanagi-Harada syndrome	bilateral, diffuse granulomatous uveitis associated with poliosis (premature graying of some or all of the hair), vitiligo, alopecia, and central nervous system and auditory signs (including dysacusia).
Voight, lines of	boundaries which delimit distribution area of main cutaneous nerves; seen in black and Asian skin and rarely in white subjects
Volkmann contracture	sequelae of compartment syndrome where there is contraction of forearm flexors
von Braun-Fernwald's sign	see Piskacek's sign
Von Economo's encephalitis	encephalitis lethargica, also associated with influenza A epidemic of 1918, associated with postencephalitic parkinsonism, first described in 1917
von Frey hairs	hairs originally derived from horses tails and graded according to their stiffness and caliber, used as a method of grading sensation in the neurologic examination
Von Gierke's disease	glycogen storage disease type I, deficiency in glucose-6-phosphatase, results in hepatomegaly and hypoglycemia
Von Graefe sign	in Graves's disease, lag of the upper eyelid as it follows the rotation of the eyeball downward
Von Hippel-Lindau disease	hemangioblastoma or cavernous hemangioma of the cerebellum, brain stem, or retina, adenomas, and cysts of the liver, kidney, pancreas, and other organs, pheochromacytomas; 35% develop renal cell carcinoma; associated with mutation in *VHL* gene
Von Myenburg complexes	close to or within portal tracts, small clusters of modestly dilated bile ducts embedded in a fibrous, sometimes hyalinized stroma; contain inspissated bile concrements and may communicate with the biliary tree; common and usually without clinical significance
Von Recklinghausen's disease	neurofibromatosis type 1
Von Recklinghausen's disease of bone	generalized osteitis fibrosa cystica; hallmark of severe hyperparathyroidism, including increased bone cell activity, peritrabecular fibrosis, and cystic brown tumors
Von Willebrand factor	vWF bridges collagen and platelets and favors platelet aggregation, ensuring platelet and vessel wall interactions; glycoprotein Ib-IX major receptor for vWF; vWF also serves as carrier for factor VIII; made in endothelial cells and megakaryocytes
Von Willebrand's disease	deficiency in von Willebrand factor; frequency of 1%, most common inherited disorder of bleeding; type 1 and 3 reduced quantity of vWF; type 2 qualitative defects in vWF
Von Zumbusch psoriasis	generalized acute pustular psoriasis

Waardenburg's syndrome	TYPE I, hearing loss; dystopia canthorum, high nasal root, heterochromia irides, white forelock, early graying; from mutation in *PAX3* gene, transcription factor involved in neural crest development; TYPE II, same as type I except for absence of dystopia; associated with mutation in MITF; TYPE III (Klein-Waardenburg), camptodactyly and other upper limb defects in addition to type I; TYPE IV (Shah-Waardenburg), deafness, pigmentary defects, and aganglionic megacolon
Wada test	test for hemispheric dominance for language by injecting amobarbital into carotid artery
Waddell's signs	in low back pain, psychological responses that predict worse outcome, including tenderness unrelated to anatomic structures, inconsistent performance of seated versus supine straight leg raise, pain on axial loading, and neurological deficits without physiologic explanation
WAGR	Wilms's tumor, anirida, genital anomalies, and mental retardation; 33% chance of developing Wilms's tumor; associated with mutation in WT1 gene
Waldenström's macroglobulinemia	marked by diffuse, leukemia-like infiltration of the bone marrow by lymphocytes, plasma cells, and hybrid forms that synthesize a monoclonal IgM, leading to macroglobulinemia; disease of old age, macroglubilinemia giving rise to visual impairment, neurologic problems, bleeding, cryoglobulinemia; hyperviscosity
Waldeyer's throat ring	the broken ring of lymphoid tissue, formed of the lingual, facial, and pharyngeal tonsils, commonly involved in non-Hodgkin's lymphoma and rarely in Hodgkin's disease
Wallenberg syndrome	infarction in posterior inferior cerebellar artery (PICA), lateral medulla 1. lesion in nucleus ambiguus, difficulty in swallowing and hoarseness, loss of gag reflex 2. vestibular nucleus, dizziness and nystagmus 3. trigeminal, loss of pain and temperature on ipsilateral 4. inferior cerebellar peduncle, ipsilateral limb ataxia 5. anterolateral system, reduced pain and temperature on contralateral limb 6. ipsilateral Horner's syndrome hiccup, for reasons not known solitary nucleus may also be destroyed, leading to loss of taste on ipsilateral half of tongue
Wallerian degeneration	pattern of degeneration of distal portion of nerve following axonal injury with break down of axon and formation of myelin ovoids from catabolized axon fragments
Warburg effect	in malignant transformation, increased anaerobic glycolysis leads to increased lactic acid production; described by Otto Warburg in 1930
Warren shunt	for treating portal hypertension, distal splenorenal shunt
Wartenberg migratory sensory neuritis	benign relapsing and remitting condition with pain and loss of sensation in distribution of individual cutaneous nerves; induced by stretch; intact deep-tendon reflexes
Warthin's tumor	papillary cystadenoma lymphomatosum, parotid gland involved, benign, more in males than females, 50s-70s
Warthin-Finkeldey cells	in measles pneumonia, cells with multiple nuclei and eosinophilic intranuclear inclusions
Warthin-Starry stain	a silver stain, will stain *H. pylori*, *Bartonella henselae*
Waterhouse-Friderichsen syndrome	with *N. meningitidis* or gonococci, pneumococci, or *Staph*.: a form of septicemia characterized by hypotension leading to shock; DIC with widespread purpura, adrenocortical insufficiency associated with massive bilateral adrenal hemorrhage
Waterston-Cooley shunt	for treating tetralogy of Fallot, anastomosis of direct ascending aorta to right pulmonary artery; rarely performed currently
Watson's water hammer pulse	also known as Corrigan's pulse in aortic regurgitation
Watson-Schwartz reaction	screening test for porphobilinogen (elevated for example in acute intermittent porphyria); mixing 4-dimethylaminobenzaldehyde with urine and then extract with chloroform; porphobilinogen extracts into upper aqueous layer giving it a pink color and urobilinogen extracts into chloroform layer; 50% positive at urinary concentration 5x upper limit of normal and consistently positive 10-20x upper limit of normal
Weber fracture	used to determine the severity of tibiofibular ligament injury by the level of fibular fracture; Weber A, fibular fractures distal to the level of the tibiotalar joint; Weber B, fracture of the fibula near the joint and a transverse fracture of the medial malleolus (or disruption of the deltoid ligament); Weber C consists of a proximal fracture of the fibula and a transverse fracture of the medial malleolus (or an intact malleolus and a ruptured deltoid ligament)
Weber syndrome	medial midbrain syndrome with ipsilateral third nerve palsy combined with contralateral hemiplegia due to peduncular lesion
Weber test	512 Hz tuning fork placed on patients's forehead; sound localizes towards size with conductive hearing loss; localizes away from the side with soensorineural loss
Weber-Christian disease	relapsing febrile nodular nonsuppurative nonvasculitic panniculitis (an inflammatory reaction in the subcutaneous fat); usually occurs in young white females; characterized by tender skin nodules and constitutional symptoms (fever, arthralgias, myalgias)
Wegener's granulomatosis	systemic vasculitis of medium and small arteries, as well as venules and arterioles defined by a clinical triad of manifestations that includes involvement of the upper airways, lungs, and kidneys and by a pathological triad consisting of necrotizing granuloma in the upper respiratory tract and lungs, vasculitis involving both arteries and veins, and focal glomerulonephritis

Weibel-Palade bodies	found only in endothelial cells of vessels larger than capillaries; granules contain von Willebrand's factor (VIII) and P-selectin
Weigert stain	iron hematoxylin, preceded by a dichromate mordant, stains myelin
Weil's disease	severe form of leptospirosis with hepatic dysfunction, renal dysfunction, and hemorrhage
Weil-Felix reaction	*Proteus* cell wall O antigens, such as OX-2, OX-19, and OX-K, cross-reacting with antigens of several species of rickettsiae
Weill-Marchesani syndrome	rare syndrome of ectopia lentis, short stature, brachydactyly, and glaucoma
Welander distal myopathy	distal myopathy seen in Sweden and Finland
Well's syndrome	eosinophilic cellulitis, characterized by recurrent cutaneous swellings which resemble acute bacterial cellulitis, and by distinctive histopathological changes. Skin lesions show dermal eosinophilic infiltration and the characteristic "flame figures" are composed of eosinophil major protein deposited on collagen bundles
Wellens's sign	in critical stenosis high in LAD, a pattern of ST-T segment in V2 and V3: isoelectric or minimally elevated (1 mm) takeoff of the ST segment, a concave or straight ST segment passing into a negative T wave at an angle of 60 to 90 degrees, and a symmetrically inverted T wave (Am Heart J 103:730, 1982)
Wenckebach block	second-degree AV blcok, Mobitz type I
Wenckebach's bundle	middle internodal tract in atrial conduction system
Werdnig-Hoffman syndrome	spinal muscular atrophy type I, autosomal recessive lower motor neuron disease, survival motor neuron protein affected (linked to 5q11.2-13.3), presents between birth and 6 months of age, death before 2 y.o.
Werlhof's disease	immune thrombocytopenic purpura; described by Werlhof in 1735
Wermer's syndrome	MEN type I, hyperplasias or tumors of the thyroid, parathyroid, adrenal cortex, pancreatic islets, or pituitary
Werner's syndrome	a form of progeria characterized by scleroderma-like skin changes (especially in extremities), bilateral juvenile cataracts, subcutaneous calcifications, wizened and prematurely-aged facies, hypogonadism, and diabetes mellitus; autosomal recessive inheritance; from mutation in *WRN* helicase locus.
Wernicke's area	important cortical center for recognizing speech, found in the superior temporal gyrus; communicates with Broca's area with arcuate fasciulus
Wernicke's encephalopathy	chronic thiamine deficiency (seen in alcoholics since alcohol impairs thiamine absorption) resulting in ataxia (primarily of gait), global confusion, ophthalmoplegia, and often nystagmus; may lead to a particular focal necrotizing encephalopathy affecting the hypothalamus, medial thalamus, and oculomotor nuclear groups in the periventricular brainstem; can be precipitated by administration of glucose to patient depleted of thiamine; after treatment with thiamine, a minority of patients have profound memory deficit (Korsakoff's syndrome)
West's syndrome	triad of infantile spasms, psychomotor developmental arrest, and characteristic EEG pattern of hypsarrhythmia (multifocal, chaotic, high-amplitude spike-and-slow wave pattern)
Westermark's sign	in chest film, an abrupt tapering of a vessel caused by pulmonary embolism, focal oligemia
Wharton's duct	submandibular duct
Whipple procedure	pancreaticoduodenectomy with cholecystectomy, truncal vagotomy, choledochojejunostomy, pancreaticojejunostomy, gastrojejunostomy
Whipple's disease	systemic bacterial infection characterized by fever (50%), weight loss (most common presenting symptom), diarrhea, lymphadenopathy, and polyarthritis (in 80%, first symptoms experienced) and, occasionally, by cardiac manifestations such as myocarditis, pericarditis, and endocarditis or by central nervous system involvement (10%); most commonly affects men in 40-60s; see small intestinal mucosa laden with distended macrophages in the lamina propria, PAS positive granules, with no inflammation; gram positive actinomycete *Tropheryma whippelii*
Whipple's triad	in insulinoma, 1. attacks precipitated by fasting or exertion 2. fasting blood glucose <50 mg/dL 3. symptoms relieved by glucose administration
Whitaker's syndrome	autoimmune polyglandular syndrome type 1 (mucocutaneous candidiasis, hypoparathyroidism, autoimmune adrenal insufficiency)
Wickham's striae	in lichen planus, papules are highlighted by a shiny surface with a lacy white pattern
Widal test	typhoid agglutination test
Wilkie's disease	partial obstruction of third part of duodenum by superior mesenteric artery resulting in abdominal pain, nausea, vomiting, weight loss
Will Rogers phenomenon	stage migration with improvements in diagnosis, causing apparent improvements in survival, i.e., stage I cases becoming stage II (*N Engl J Med* 1985;312:1604)
Williams's syndrome	supravalvular aortic stenosis, mental retardation, elfin facies, association with hypercalcemia due to abnormal sensitivity to vitamin D, idiopathic hypercalcemia of pregnancy, loquacious personality, abnormally sensitive hearing; from deletion in elastin gene and probably several adjacent genes
Williams-Campbell syndrome	congenital cartilage deficiency, associated with bronchiectasis

Williams-Fitzgerald-Flaujeac factor	high molecular weight kininogen; see description under Fitzgerald factor
Willis, circle of	cerebral arterial circle, an anastomosis between the two vertebral and two internal carotid arteries
Wilms's tumor	childhood primary renal tumor , 5% of patients with sporadic Wilms tumor have mutations in WT-1, cancer suppressor gene on 11p13
Wilson's disease	hepatolenticular degeneration due to mutation in gene involved in incorporation of copper into ceruloplasmin and excretion of copper into bile; Kayser-Fleischer ring; gene on chr 13, cation transporting P-type ATPase; incidence 1:200,000, diagnosis based on decrease in serum ceruloplasmin, increased urinary excretion of copper, increase in hepatic copper content; 40% have neurologic findings (Parkinson's, psychosis) and subclinical liver
Winslow, foramen of	anterior portal triad; posterior IVC and right crus of diaphragm; superior caudate lobe; inferior superior part of duodenum, portal triad
Winter's formula	gives expected pCO2 (respiratory compensation) in uncomplicated metabolic acidosis; expected CO2=[HCO3]*1.54 + 8.36
Winterbottom's sign	in West African trypanosomiasis (sleeping sickness, caused by *Trypanosoma brucei gambiense*, humans primary reservoir), classic finding of posterior cervical triangle lymphadenopathy; described by Thomas Winterbottom who noted that slave traders in late 18th century used neck swelling as indicator of sleeping sickness in slaves
Wintrobe indices	mean cell volume; mean cell hemoglobin; mean cell hemoglobin concentration
Wirsung, duct of	embryologically confined to the ventral pancreas, becomes functionally the main pancreatic duct after duct fusion occurs; drains the bulk of pancreatic secretion through the major papilla
Wiskott-Aldrich syndrome	X-linked characterized by triad of eczema, thrombocytopenia (from autoantibodies), and repeated infections; small platelets (3-5 fL); failure to express sialic acid-rich glycoprotein, sialophorin or CD15, ending in early death
Wohlfart-Kugelberg-Welander disease	spinal muscular atrophy type III, presents in late childhood, runs a slow, indolent course, weakness greatest in proximal muscles, autosomal recessive or autosomal dominant, survival motor neuron protein affected
Wolff-Chaikoff effect	when increasing doses of iodide inhibit organification and hormonogenesis of thyroid hormone
Wolff-Parkinson-White syndrome	paroxysmal supraventricular tachycardia caused by conduction through abnormal accessory bypass tract (bundle of Kent) between atria and ventricles characterized by triad of 1. wide QRS complex, 2. relatively short PR interval, 3. slurring of initial part of QRS delta wave; in familial WPW, associated with mutation in gamma2 regulatory subunit of AMP-activated protein kinase (*PRKAG2*)
Wolf-Hirschhorn syndrome	deletion 4p syndrome, from de novo deletion of 4p16.3 to 4pter; associated with growth and mental deficiency, seizure disorder, prominent glabella, among other features
Wolfram syndrome	diabetes insipidus and mellitus with optic atrophy and deafness, also referred to as DIDMOAD; diabetes is non autoimmune; caused by mutation in gene encoding wolframin
Wollfian duct	in male embryonic development, duct that gives rise to epididymis, vasa deferentia, seminal vesicles, and ejaculatory ducts
Wolman disease	lysosomal acid lipase deficiency hydrolysis of cholesteryl esters and triglycerides in the lysosome leading to mild mental retardation, hepatomegaly, adrenal calcification, fatal in infancy with inanition, malabsorption, and intractable diarrhea
Wood's lamp	lamp that emits UV light
Wood's maneuver	in management of shoulder dystocia, rotating posterior shoulder 180 degrees
Woodruff's plexus	site for posterior epistaxis; made up of anastamoses between branches of the internal maxillary artery; located where the sphenopalatine artery enters the nasal cavity through the sphenopalatine foramen at the posterior limit of the middle turbinate
Woolf's syndrome	Autosomal dominant syndrome with albinismus circumscriptus and deaf-mutism without other features of Waardenburg syndrome
Woronets trait	dominantly-inherited trait characterized by small population of markedly-distorted RBCs resembling keratocytes; normal RBC life span
Wright's stain	stain used for blood and bone marrow films
Wright's maneuver	in thoracic outlet obstruction: evaluating the radial pulse at the wrist with the shoulder in external rotation and abduction, positive sign if it reproduces shoulder and arm symptoms and obliterates radial pulse
Wunderlich's syndrome	spontaneous renal bleeding of non-traumatic origin, confined to the subcapsular and perirenal space
Yergason's sign	in biceps tendinitis or shoulder impingement, worsening of pain with resisted supination while the elbow is flexed to 90 degrees, arm adducted
Young's syndrome	clinical features similar to cystic fibrosis, including bronchiectasis, sinusitis, and obstructive azoospermia, but don't have increased sweat chloride values, nor pancreatic insufficiency, nor abnormal nasal potential differences, nor the CF delta F508 mutation; affected individuals are often middle-aged males identified during evaluation for infertility
Yuzpe regimen	ethinyl estradiol 100 mcg and levonorgestrel 0.5 mg taken twice,12 hours apart, reduces number of anticipated pregnancies by 75-80%
Zahn, infarct of	in occlusion of an intrahepatic branch of portal vein, sharply demarcated area of red-blue discoloration, not infarct, not necrosis, only marked stasis in distended sinusoids, with secondary hepatocellular atrophy
Zahn, lines of	thrombi formed within a cardiac chamber or the aorta, may have apparent laminations, produced by alternating layers of paler

	platelets admixed with some fibrin, separated by darker layers containing more red cells
Zavanelli maneuver	for management of shoulder dystocia, cephalic replacement of infant followed by caesarian delivery
Zellweger syndrome	disorder of peroxisomes characterized by the congenital absence of functioning peroxisomes resulting in a cerebrohepatorenal syndrome; associated with prenatal development problems, hepatomegaly, increased serum iron and copper, visual disturbances; generally fatal by 1 year; caused by mutations in genes involved in peroxisome biogenesis (PEX genes)
Zenker's diverticulum	a pharyngeal diverticulum from premature contraction of the cricopharyngeus muscle on swallowing, leads to progressive UES narrowing, leading to a posteriorly directed hypopharynx; causes progressive food stasis and dysphagia
Zieve's syndrome	jaundice, hyperlipemia, and hemolytic anemia in alcoholic liver disease; described in 1958
Zollinger-Ellison syndrome	hallmark, circulating hypergastrinemia; gastric acid hypersecretion and severe peptic ulcer diathesis secondary to unbridled release of gastrin from a gastrinoma; associated with peptic ulcers and diarrhea; 60% malignant, only 20% resectable; 25% of gastrinoma patients have MEN I; >80% of gastrinomas found in gastrinoma triangle
Zoon's balanitis	plasma cell balanitis (also known as balanitis circumscripta plasmacellularis), idiopathic, rare, benign, penile dermatosis with plasma cell infiltration; treated with circumcision; described in 1952
Zuckerkandl, organ of	collection of para-aortic, paraganglion cells around the origin of the inferior mesenteric artery; a site where extra-adrenal pheochromocytomas may arise