Part 1
General Otolaryngology

Chapter 1
Syndromes and Eponyms

Syndromes and Diseases

Adult Respiratory Distress Syndrome

Adult respiratory distress syndrome (ARDS) is characterized by a delay in onset (12-24 hours) following injury, shock, and/or successful resuscitative effort. Septic shock, extrathoracic trauma, central nervous system (CNS) pathology, fat embolism, oxygen toxicity, head and facial injuries, and massive blood transfusions can lead to ARDS. It is characterized by hypoxia and pulmonary infiltrates secondary to increased pulmonary vascular permeability, microvascular hemorrhage, or both.

Aide Syndrome

Aide syndrome is characterized by decreased pupillary reaction and deep tendon reflex. The etiology is unknown.

Alagille Syndrome

Alagille syndrome is marked by cardiovascular abnormalities, characteristic facial appearance, chronic cholestasis, growth retardation, hypogonadism, mental retardation, vertebral arch defect, temporal bone anomalies in the cochlear aqueduct, ossicles, semicircular canals (SCCs), and subarcuate fossa. Liver transplantation is a possible treatment.

Albers-Schönberg Disease

A genetic disorder also known as osteopetrosis, Albers-Schönberg disease results in progressive increase in the density (but also increase in weakness) of the bones in the skeletal system. Vascular nutrition to affected bones is also decreased by this disease. Broken down into three categories, there is osteopetrosis with precocious manifestations, osteopetrosis with delayed manifestations, and pyknodysostosis. In the mandible long-term antibiotic therapy, multiple debridements, sequestrectomies, or even resection are possible treatments.

Albright Syndrome

Polyostotic fibrous dysplasia usually manifests early in life as multicentric lesions involving the long bones and bones of the face and skull with scattered skin lesions similar to melanotic café au lait spots and precocious puberty in female patients. Frequently, there is an elevation of serum alkaline phosphatase as well as endocrine abnormalities.

Aldrich Syndrome

Thrombocytopenia, eczema, and recurrent infections occur during the first year of life. It is inherited through a sex-linked recessive gene. The bleeding time is prolonged, the platelet count is decreased, and the bone marrow megakaryocytes are normal in number.

Amalric Syndrome

Granular macular pigment epitheliopathy (foveal dystrophy) is associated with sensorineural hearing loss. Visual acuity is usually normal. Amalric syndrome may be a genetic disorder, or it may be the result of an intrauterine rubella infection.

Aortic Arch Syndrome

See Takayasu Disease.

Apert Syndrome

Apert syndrome is not to be confused with Pfeiffer syndrome, which has different types of hand malformations.

Ascher Syndrome

Ascher syndrome is a combination of blepharochalasis, double lip, and goiter.

Auriculotemporal Syndrome (Frey Syndrome)

Auriculotemporal syndrome is characterized by localized flushing and sweating of the ear and cheek region in response to eating. It usually occurs after parotidectomy. It is assumed that the parasympathetic fibers of the ninth nerve innervate the sweat glands after parotidectomy. It has been estimated that 20% of the parotidectomies in children result in this disorder.

Avellis Syndrome

Unilateral paralysis of the larynx and velum palati, with contralateral loss of pain and temperature sensitivity in the parts below the larynx characterize Avellis syndrome. The syndrome is caused by involvement of the nucleus ambiguus or the vagus nerve along with the cranial portion of the ninth nerve.

Babinski-Nageotte Syndrome

Babinski-Nageotte syndrome is caused by multiple or scattered lesions, chiefly in the distribution of the vertebral artery. Ipsilateral paralysis of the soft palate, larynx, pharynx, and sometimes

tongue occurs. There is also ipsilateral loss of taste on the posterior third of the tongue, loss of pain and temperature sensation around the face, and cerebellar asynergia. Horner syndrome with contralateral spastic hemiplegia and loss of proprioceptive and tactile sensation may also be present.

Baelz Syndrome

Painless papules at the openings of the ducts of the mucous glands of the lips with free exudation of mucus are characteristic. Congenital and familial forms are precancerous. Acquired forms are benign and caused by irritating substances.

Bannwarth Syndrome (Facial Palsy in Lymphocytic Meningoradiculitis)

A relatively benign form of acute unilateral or bilateral facial palsy that is associated with lymphocytic reactions and an increased protein level in the cerebrospinal fluid (CSF) with minimal, if any, meningeal symptoms is known as Bannwarth syndrome. Neuralgic or radicular pain without facial palsy and unilateral or bilateral facial palsy of acute onset are symptoms of this syndrome. A virus has been suggested as a possible etiology. Males are more often affected than females, with the greatest number of cases occurring in the months of August and September.

Barany Syndrome

Barany syndrome is a combination of unilateral headache in the back of the head, periodic ipsilateral deafness (alternating with periods of unaffected hearing), vertigo, and tinnitus. The syndrome complex may be corrected by induced nystagmus.

Barclay-Baron Disease

Vallecular dysphagia is present.

Barre-Lieou Syndrome

Occipital headache, vertigo, tinnitus, vasomotor disorders, and facial spasm due to irritation of the sympathetic plexus around the vertebral artery in rheumatic disorders of the cervical spine are characteristic. It is also known as cervical migraine.

Barrett Syndrome

Barrett syndrome is characterized by esophagitis due to change in the epithelium of the esophagus.

Barsony-Polgar Syndrome

A diffuse esophageal spasm, caused by disruption of the peristaltic waves by an irregular contraction resulting in dysphagia and regurgitation, is evidence of this syndrome. It most commonly affects excitable elderly persons.

Basal Cell Nevoid Syndrome

This familial syndrome, non–sex-linked and autosomal dominant with high penetrance and variable expressivity, manifests early in life. It appears as multiple nevoid basal cell epitheliomas of the skin, cysts of the jaw, abnormal ribs and metacarpal bones, frontal bossing, and dorsal scoliosis. Endocrine abnormalities have been reported and it has been associated with medulloblastoma. The cysts in the jaw, present only in the maxilla and mandible, are destructive to the bone. The basal cell epitheliomas are excised as necessary, and the cysts in the jaw rarely recur after complete enucleation.

Bayford-Autenrieth Dysphagia (Arkin Disease)

Dysphagia lusoria is said to be secondary to esophageal compression from an aberrant right subclavian artery.

Beckwith Syndrome

This is a congenital disorder characterized by macroglossia, omphalocele, hypoglycemia, pancreatic hyperplasia, noncystic renal hyperplasia, and cytomegaly of the fetal adrenal cortex.

Behçet Syndrome

Of unknown etiology, this disease runs a protracted course with periods of relapse and remission. It manifests as indolent ulcers of the mucous membrane and skin, stomatitis, as well as anogenital ulceration, iritis, and conjunctivitis. No definitive cure is known, though steroids help.

Besnier-Boeck-Schaumann Syndrome

Sarcoidosis is present.

Bloom Syndrome

An autosomal recessive growth disorder, Bloom syndrome is associated with chromosomal breaks and rearrangements. It is also associated with an unusually high rate of cancer at an early age. Associated with facial erythema, growth retardation, immunodeficiency, infertility, and sun sensitivity, diagnosis is confirmed by chromosome analysis. Anomalous numbers of digits or teeth, asymmetric legs, heart malformation, hypopigmented spots in blacks, protruding ears, sacral dimple, simian line, and urethral or meatal narrowing are less common characteristics. For head and neck tumor patients, there is an increased chance of secondary and primary tumors.

Bogorad Syndrome

Bogorad syndrome is also known as the syndrome of crocodile tears, characterized by residual facial paralysis with profuse lacrimation during eating. It is caused by a misdirection of regenerating autonomic fibers to the lacrimal gland instead of to the salivary gland.

Bonnet Syndrome

Sudden trigeminal neuralgia accompanied by Horner syndrome and vasomotor disorders in the area supplied by the trigeminal nerve are manifestations of this syndrome.

Bonnier Syndrome

Bonnier syndrome is caused by a lesion of Deiters nucleus and its connection. Its symptoms include ocular disturbances (eg, paralysis of accommodation, nystagmus, diplopia), deafness, nausea, thirst, and anorexia, as well as other symptoms referable to involvement of the vagal centers, cranial nerves VIII, IX, X, and XI, and the lateral vestibular nucleus. It can simulate Ménière disease.

Bourneville Syndrome

Bourneville syndrome is a familial disorder whose symptoms include polyps of the skin, harelip, moles, spina bifida, and microcephaly.

Bowen Disease

Bowen disease is a precancerous dermatosis characterized by the development of pinkish or brownish papules covered with a thickened horny layer. Histologically, it shows hyperchromatic acanthotic cells with multinucleated giant cells. Mitoses are frequently observed.

Branchio-Oto-Renal Syndrome

Branchio-oto-renal syndrome is an autosomal dominant disorder characterized by anomalies of the external, middle, and inner ear in association with preauricular tissues, branchial cleft anomalies, and varying degrees of renal dysplasia, including aplasia. Many of the following symptoms (but not necessarily all) are present.

A. Conductive or mixed hearing loss
B. Cup-shaped, anteverted pinnae with bilateral preauricular sinuses
C. Bilateral branchial cleft fistulas or sinuses
D. Renal dysplasia

This syndrome is among a group of syndromes characterized by deformities associated with the first and second branchial complexes. The precise incidence of the disorder is unknown.

Briquet Syndrome

Briquet syndrome is characterized by a shortness of breath and aphonia due to hysteric paralysis of the diaphragm.

Brissaud-Marie Syndrome

Unilateral spasm of the tongue and lips of a hysteric nature are characteristic.

Brown Syndrome

Brown syndrome is a congenital or acquired abnormality of the superior oblique muscle tendon characterized by vertical diplopia and the inability to elevate the eye above midline or medial gaze. This syndrome is of two types: true and simulated. True Brown syndrome is always congenital. Simulated Brown syndrome is either congenital or acquired. The congenital simulated type may be caused by thickening of an area in the posterior tendon or by the firm attachment of the posterior sheath to the superior oblique tendon. The acquired simulated type may be caused by inflammation extending from the adjacent ethmoid cells to the posterior sheath and tendon, an orbital floor fracture, frontal ethmoidal fracture, crush fracture of nasal bones, sinusitis, frontal sinus surgery, or surgical tucking of the superior oblique tendon.

Brun Syndrome

Vertigo, headache, vomiting, and visual disturbances due to an obstruction of CSF flow during positional changes of the head are seen. The main causes of this syndrome include cysts and cysticercosis of the fourth ventricle as well as tumors of the midline cerebellum and third ventricle.

Burckhardt Dermatitis

Burckhardt dermatitis appears as an eruption of the external ear. It consists of red papules and vesicles that appear after exposure to sunlight. The rash usually resolves spontaneously.

Caffey Disease (Infantile Cortical Hyperostosis)

Of familial tendency, its onset is usually during the first year of life. It is characterized by hyperirritability, fever, and hard nonpitting edema that overlie the cortical hyperostosis. Pathologically, it involves the loss of periosteum with acute inflammatory involvement of the intratrabecular bone and the overlying soft tissue. Treatment is supportive, consisting of steroids and antibiotics. The prognosis is good. The mandible is the most frequently involved site.

Caisson Disease

This symptom complex occurs in men and women who work in high air pressures and are returned too suddenly to normal atmospheric pressure. Similar symptoms may occur in fliers when they suddenly ascend to high altitudes unprotected by counterpressure. It results from the escape from solution in the body fluids of bubbles (mainly nitrogen) originally absorbed at higher pressure. Symptoms include headache; pain in the epigastrium, sinuses, and tooth sockets; itchy skin; vertigo; dyspnea; coughing; nausea; vomiting; and sometimes paralysis. Peripheral circulatory collapse may be present. Nitrogen bubbles have been found in the white matter of the spinal cord. It also can injure the inner ear through necrosis of the organ of Corti. There is a question of rupture of the round window membrane; hemotympanum and eustachian tube obstruction may occur.

Camptomelic Syndrome

The name is derived from a Greek word meaning *curvature of extremities*. The syndrome is characterized by dwarfism, craniofacial anomalies, and bowing of the tibia and femur, with malformation of other bones. The patient has cutaneous dimpling overlying the tibial bend. Respiratory distress is common, and the patient has an early demise in the first few months of life. In the otolaryngologic area, the patient exhibits a prominent forehead, flat facies with a broad nasal bridge and low-set ears, cleft palate, mandibular hypoplasia, and tracheobronchial malacia that contributes to the respiratory distress and neonatal death. Histologically, two temporal bone observations showed defective endochondral ossification with no cartilage cells in the endochondral layer of the otic capsule. The cochlea was shortened and flattened, presenting a scalar communis. The vestibule and the SCC were deformed by bone invasion.

This syndrome is often of unknown etiology, although some believe it is autosomal recessive. Others believe it may be due to an exogenous cause.

This syndrome is not to be confused with Pierre Robin syndrome, which presents with very similar clinical features.

Cannon Nevus

This is an autosomal dominant disorder characterized by spongy white lesions of the oral and nasal mucosa. The lesions are asymptomatic and may be found from the newborn period with increasing severity until adolescence. The histologic picture is that of keratosis, acanthosis, and parakeratosis.

Carcinoid Syndrome

The symptoms include episodic flushing, diarrhea, and ascites. The tumor secretes serotonin. Treatment is wide excision. The tumor may give a positive dopa reaction.

Carotid Sinus Syndrome (Charcot-Weiss-Barber Syndrome)

When the carotid sinus is abnormally sensitive, slight pressure on it causes a marked fall in blood pressure due to vasodilation and cardiac slowing. Symptoms include syncope, convulsions, and heart block.

Castleman Disease

Castleman disease was first described by Castelman et al in 1954. It is a benign lymphoepithelial disease that is most often mistaken for lymphoma. It is also known as localized nodal hyperplasia, angiomatous lymph node hyperplasia, lymphoid hamartoma, and giant lymph nodal hyperplasia. Symptoms include tracheobronchial compression, such as cough, dyspnea, hemoptysis, or dysphagia. Masses in the neck are also not uncommon. There are two histologic types: the

hyaline vascular type and the plasma cell type. Follicles in the hyaline vascular type are traversed by radially oriented capillaries with plump endothelial cells and collagenous hyalinization surrounding the vessels. The follicles in the plasma cell type are normal in size without capillary proliferation or hyalinization. Intermediate forms exist but are rare. Treatment entails complete excision of the mass. Etiology is unknown.

Cavernous Sinus Syndrome

The cavernous sinus receives drainage from the upper lip, nose, sinuses, nasopharynx, pharynx, and orbits. It drains into the inferior petrosal sinus, which in turn drains into the internal jugular vein. The cavernous sinus syndrome is caused by thrombosis of the cavernous intracranial sinus, 80% of which is fatal. The symptoms include orbital pain (V_1) with venous congestion of the retina, lids, and conjunctiva. The eyes are proptosed with exophthalmos. The patient has photophobia and involvement of nerves II, III, IV, and V_1. The treatment of choice is anticoagulation and antibiotics. The most common cause of cavernous sinus thrombosis is ethmoiditis. The ophthalmic vein and artery are involved as well. (The nerves and veins are lateral to the cavernous sinus, and the internal carotid artery is medial to it.)

Cestan-Chenais Syndrome

Cestan-Chenais syndrome is caused by occlusion of the vertebral artery below the point of origin of the posteroinferior cerebellar artery. There is paralysis of the soft palate, pharynx, and larynx. Ipsilateral cerebellar asynergia and Horner syndrome are also present. There is contralateral hemiplegia and diminished proprioception and tactile sensation.

Champion-Cregah-Klein Syndrome

This is a familial syndrome consisting of popliteal webbing, cleft lip, cleft palate, lower lip fistula, syndactyly, onychodysplasia, and pes equinovarus.

Chapple Syndrome

This disorder is seen in the newborn with unilateral facial weakness or paralysis in conjunction with comparable weakness or paralysis of the contralateral vocal cord, the muscles of deglutition, or both. The disorder is secondary to lateral flexion of the head in utero, which compresses the thyroid cartilage against the hyoid or cricoid cartilages or both, thereby injuring the recurrent or superior laryngeal nerve, or both.

Charcot-Marie-Tooth Disease

This is a hereditary and degenerative disease that includes the olivopontocerebellar, cerebelloparenchymal, and spinocerebellar disorders and the neuropathies. This disease is characterized by chronic degeneration of the peripheral nerves and roots; and distal muscle atrophy in feet, legs, and hands. Deep tendon reflexes are usually nil. It is also associated with hereditary cerebellar ataxia features, optic atrophy, and other cranial involvement. Some suggest that this disease is linked to auditory dysfunction and that it is also linked to other CNS dysfunctions. This disease can be progressive, and it can also spontaneously arrest.

CHARGE Syndrome

CHARGE syndrome (**c**oloboma of the eye, **h**eart defects, **a**tresia of the choanae, **r**etardation of growth and development, **g**enital and/or urinary abnormalities, and **e**ar abnormalities and deafness) is a genetic pattern of birth defects which occur one in 10,000 births worldwide, without any family history. It involves heart defects, breathing and swallowing difficulties, hearing loss, vision loss, and balance problems.

Major Features of CHARGE Syndrome (Very Common in CHARGE and Relatively Rare in Other Conditions)

Feature	*Includes*	*Frequency (%)*
Coloboma of the eye	Coloboma (sort of like a cleft) of the iris, retina, choroid, macula, or disc (not the eyelid); microphthalmos (small eye) or anophthalmos (missing eye): *Causes vision loss*	80-90
Choanal atresia or stenosis	They can be stenosed or atretic. It can be unilateral or bilateral, bony, or membranous.	50-60
Cranial nerve abnormality	I—Missing or decreased sense of smell IX/X—Swallowing difficulties, aspiration VII—Facial palsy (one side or both)	90-100 70-90 40
CHARGE outer ear	Short, wide ear with little or no lobe, "snipped off" helix, prominent antihelix which is discontinuous with tragus, triangular concha, decreased cartilage (floppy), often stick out, usually asymmetric	> 50
CHARGE middle ear	Malformed bones of the ossicles: *Conductive hearing loss*	Common
CHARGE inner ear	Mondini defect; small or absent semicircular canals: *Balance problems and sensorineural loss*	90

Minor Characteristics of CHARGE: Significant, But More Difficult to Diagnose or Less Specific to CHARGE

Feature	*Includes*	*Frequency (%)*
Heart defects	Can be any type, but many are complex, such as tetralogy of Fallot	75
Cleft lip ± cleft palate	Cleft lip with or without cleft palate, cleft palate, submucous cleft palate	20
TE fistula	Esophageal atresia, tracheoesophageal fistula (TEF), H-shaped TEF	15-20
Kidney abnormalities	Small kidney, missing kidney, misplaced kidney, reflux	40
Genital abnormalities	Males: small penis, undescended testes Females: small labia, small or missing uterus Both: lack of puberty without hormone intervention	50 25 90
Growth deficiency	Growth hormone deficiency Other short stature	15 70
Typical CHARGE face	Square face with broad prominent forehead, arched eyebrows, large eyes, occasional ptosis, prominent nasal bridge with square root, thick nostrils, prominent nasal columella, flat midface, small mouth, occasional small chin, larger chin with age. Facial asymmetry even without facial palsy	
Palm crease	Hockey-stick palmar crease	50
CHARGE behavior	Perseverative behavior in younger individuals, obsessive compulsive behavior in older individuals	> 50

Chédiak-Higashi Syndrome

Chédiak-Higashi syndrome is the result of an autosomal recessive trait. It is characterized by albinism, photophobia, nystagmus, hepatosplenomegaly, anomalous cellular granules, and development of lymphoma. These patients usually die during childhood of fulminant infections.

Cleft Lip Palate and Congenital Lip Fistulas

This syndrome is transmitted in an autosomal dominant manner with 80% penetrance; it occurs in 1 per 100,000 live births. Usually bilateral, symmetrically located depressions are noted on the vermilion portion of the lower lip and communicate with the underlying minor salivary glands. The lip pits may be an isolated finding (33%) or be found with cleft lip palate (67% of cases). Associated anomalies of the extremities may include talipes equinovarus, syndactyly, and popliteal pterygia. Congenital lip pits have also been seen in association with the oral-facial-digital syndrome.

Cockayne Syndrome

Cockayne syndrome is autosomal recessive, progressive bilateral sensorineural hearing loss, associated with dwarfism, facial disharmony, microcephaly, mental deficiency, retinitis pigmentosa, optic atrophy, intracranial calcification, and multiple dental caries. Patients succumb to respiratory or genitourinary infection in the teens or twenties.

Cogan Syndrome

Nonsyphilitic interstitial keratitis and vestibuloauditory symptoms are characteristics of Cogan syndrome. Interstitial keratitis gives rise to rapid visual loss. Symptoms include episodic severe vertigo accompanied by tinnitus, spontaneous nystagmus, ataxia, and progressive sensorineural hearing loss. There are remissions and exacerbations. It is believed to be related to periarteritis nodosa. Eosinophilia has been reported in this entity. Pathologically, it is a degeneration of the vestibular and spiral ganglia with edema of the membranous cochlea, SCCs, and inflammation of the spiral ligament. Treatment with steroids has been advocated.

Cyclophosphamide and azathioprine have been used in addition to prednisone (40 mg daily). This syndrome is not to be confused with Ménière disease despite vertiginous symptoms and fluctuating hearing loss. Vogt-Koyanagi-Harada syndrome is also similar but involves alopecia, poliosis, and exudative uveitis. Syphilis is also confused with this syndrome, but in syphilis, the interstitial keratitis is old and usually does not demonstrate active inflammatory changes. Syphilitic involvement of the cornea is often centrally located. Follow-up treatment of patients must be thorough in order to detect more extensive involvement, such as systemic vasculitis or aortitis.

Collet-Sicard Syndrome

The 9th, 10th, and 11th nerves are involved with normal sympathetic nerves. The etiology is usually a meningioma or other lesion involving the nerves in the posterior cranial fossa.

Conradi-Hünermann Syndrome

The most common variant of chondrodysplasia punctata; this syndrome is characterized by punctate epiphyseal calcifications. Clinical features include saddle nose deformity, micromelia, rhizomelia, short stature, flexion contractures, and dermatoses. This syndrome is also known as chondrodystrophia epiphysialis punctata, stippled epiphysis disease, dysplasia epiphysialis punctata, chondroangiopathia calcarea punctata, and Conradi disease. Some cases point to sporadic mutations and others to autosomal dominant patterns of inheritance. The clinical features

of this syndrome are so varied from case to case that only a complete workup can exclude other versions of this syndrome.

Costen Syndrome

Costen syndrome is a temporomandibular joint (TMJ) abnormality, usually due to impaired bite and characterized by tinnitus, vertigo, and pain in the frontal, parietal, and occipital areas with a blocked feeling and pain in the ear. After a careful workup to rule out other abnormalities, the patient is treated with aspirin, heat, and slow exercise of the joint. An orthodontist may help the patient. The TMJ differs from other joints by the presence of avascular fibrous tissue covering the articulating surfaces with an interposed meniscus dividing the joint into upper and lower compartments. The right and left TMJs act as one functional unit. The condyle is made up of spongy bone with marrow and a growth center. The condyle articulates with the glenoid fossa of the temporal bone (squamosa). The squamotympanic fissure separates the fossa from the tympanic bone. The joint is a ginglymoarthrodial joint with hinge and transverse movements. The key supporting ligament of the TMJ is the temporomandibular ligament. The boundaries of the glenoid fossa are as follows:

Anterior:	Margins of the articular eminence
Posterior:	Squamosotympanic fissure
Lateral:	Zygomatic process of the temporal bone
Medial:	Temporal spine

The TMJ derives its nourishment from the synovial membrane, which is richly vascularized and produces a mucinous-like substance. The joint has a gliding motion between the meniscus and the temporal bone (upper compartment). It has a hinge motion between the disk and the condyle (lower compartment). It is innervated by the auriculotemporal nerve, masseter nerve, lateral pterygoid nerve, and temporal nerve. It is supplied by the superficial temporal artery and the anterior tympanic branch of the internal maxillary artery. The lateral pterygoid muscle protracts the jaw, and the masseter, medial pterygoid, and temporalis muscles act as elevators. All these muscles are innervated by V_3. The sphenomandibular and stylomandibular ligaments have no function in TMJ articulation.

Cowden Syndrome

This is a familial syndrome characterized by adenoid facies, hypoplasia of the mandible and maxilla, high-arched palate, hypoplasia of the soft palate and uvula, microstomia, papillomatosis of the lips and pharynx, scrotal tongue, multiple thyroid adenomas, bilateral breast hypertrophy, pectus excavatum, and liver and CNS abnormalities.

Creutzfeldt-Jakob Disease

Creutzfeldt-Jakob disease is a rare spongiform encephalopathy. Constitutional symptoms lead to mental retardation and movement disorder.

Cri du Chat Syndrome

Cri du Chat syndrome is a condition caused by a B group chromosome with a short arm; its symptoms are mental retardation, respiratory stridor, microcephaly, hypertelorism, midline oral clefts, and laryngomalacia with poor approximation of the posterior vocal cords.

Crouzon Disease

See Chapter 17.

Curtius Syndrome

Curtius syndrome is a form of hypertrophy that may involve a single small part of the body or an entire system (ie, muscular, nervous, or skeletal systems). It is also known as congenital hemifacial hypertrophy.

Dandy Syndrome

Oscillopsia or jumbling of the panorama common in patients after bilateral labyrinthectomy is characteristic of this syndrome. These patients are unable to focus while walking or moving.

Darier Disease (Keratosis Follicularis)

Autosomal dominant, this skin disorder of the external auditory canal is characterized by keratotic debris in the canal. Some investigators have advocated the use of vitamin A or steroids.

De'Jean Syndrome

Exophthalmos, diplopia, superior maxillary pain, and numbness along the route of the trigeminal nerve are found with lesions of the orbital floor in this syndrome.

Déjérine Anterior Bulbar Syndrome

This syndrome is evidenced by thrombosis of the anterior spinal artery, resulting in either an alternating hypoglossal hemiplegia or an alternating hypoglossal hemianesthetic hemiplegia.

Demarquay-Richter Syndrome

Demarquay-Richter syndrome is a congenital orofacial disorder characterized by cleft lip, cleft palate, lower lip fistulas, and progeria facies. Defective dentition, heart defects, dwarfism, and finger abnormalities may be seen.

DIDMOAD Syndrome

DIDMOAD syndrome is an autosomal recessive disorder associating **d**iabetes **i**nsipidus, **d**iabetes **m**ellitus, **o**ptic **a**trophy, and **d**eafness. Diabetes mellitus is usually juvenile in onset and insulin dependent. The diabetes insipidus has a varied time of onset and is vasopressin sensitive, indicative of degeneration of the hypothalamic cells or of the supraopticohypophyseal tract. The hearing loss is sensorineural and progressive, and primarily affects the higher tones. Urinary tract abnormalities ranging from atonic bladder to hydronephrosis and hydroureter have been reported with this disorder.

DiGeorge Syndrome

Lischaneri reported three categories of this syndrome:

A. Third and fourth pharyngeal pouch syndrome, characterized by cardiovascular and craniofacial anomalies as well as abdominal visceral abnormalities
B. DiGeorge syndrome (thymus agenesis)
C. Partial DiGeorge syndrome (thymic hypoplasia in which the thymus gland weighs less than 2 g)

The patients have small malformed pinnae with narrow external auditory canals and abnormal ossicles. The patients also have shortened cochlea of the Mondini type as well as an absence of hair cells in the hook region, hypertelorism with nasal cleft, shortened philtrum, and

micrognathia. Other middle ear anomalies include an absence of stapedial muscle, hypoplastic facial nerve, and absent oval window. Most of the findings are symmetrical.

Down Syndrome

See section on trisomy in Chapter 17.

Dysphagia Lusoria

Dysphagia lusoria is secondary to an abnormal right subclavian artery. The right subclavian arises abnormally from the thoracic aorta by passing behind or in front of the esophagus, thus compressing it.

Eagle Syndrome

The patient has elongation of the styloid process or ossification of the stylohyoid ligament causing irritation of the trigeminal, facial, glossopharyngeal, and vagus nerves. Symptoms include recurrent nonspecific throat discomfort, foreign body sensation, dysphagia, facial pain, and increased salivation. Carotidynia may result from impingement of the styloid process on the carotid artery, producing regional tenderness or headaches. The only effective treatment for Eagle syndrome is surgical shortening of the styloid process.

Ectodermal Dysplasia, Hidrotic

See Chapter 17.

Ectodermal Dysplasia, Hypohidrotic

This syndrome consists of hypodontia, hypotrichosis, and hypohidrosis. Principally, the structures involved are of ectodermal origin. Eyelashes and especially eyebrows are entirely missing. Eczema and asthma are common. Aplasia of the eccrine sweat glands may lead to severe hyperpyrexia. The inheritance is X-linked recessive.

18q Syndrome

This syndrome consists of psychomotor retardation, hypotonia, short stature, microcephaly, hypoplastic midface, epicanthus, ophthalmologic abnormalities, cleft palate, congenital heart disease, abnormalities of the genitalia, tapered fingers, aural atresia, and conductive hearing loss.

Eisenlohr Syndrome

Numbness and weakness in the extremities; paralysis of the lips, tongue, and palate; and dysarthria are evidenced.

Elschnig Syndrome

Extension of the palpebral fissure laterally, displacement of the lateral canthus, ectropion of the lower lid, and lateral canthus are observed. Hypertelorism, cleft palate, and cleft lip are frequently seen.

Empty Sella Syndrome

The patient has an enlarged sella, giving the appearance of a pituitary tumor. An air encephalogram shows an empty sella. The syndrome consists of the abnormal extension into the sella turcica of an arachnoid diverticulum filled with CSF, displacing and compressing the pituitary gland. Four causal theories of this syndrome exist: (1) rupture of an intrasellar or parasellar cyst;

(2) infarction of a pituitary adenoma; (3) pituitary hypertrophy and subsequent involution; and (4) the most common theory, the syndrome is due to CSF pressure through a congenitally deficient sella diaphragm leading to the formation of an intrasellar arachnoidocele. A trans-septal or trans-sphenoidal route to the sella is a treatment to consider.

The primary empty sella syndrome is due to congenital absence of the diaphragm sella, with gradual enlargement of the sella secondary to pulsations of the brain. Secondary empty sella syndrome may be due to necrosis of an existing pituitary tumor after surgery, postirradiation directed at the pituitary, or pseudotumor cerebri.

Face-Hand Syndrome

Face-hand syndrome is a reflex sympathetic dystrophy that is seen after a stroke or myocardial infarction. There may be edema and erythema of the involved parts along with persistent burning.

Fanconi Anemia Syndrome

Patients have aplastic anemia with skin pigmentation, skeletal deformities, renal anomalies, and mental retardation. Death due to leukemia usually ensues within 2 years. The disorder rarely occurs in adults. (A variant is congenital hypoplastic thrombocytopenia, which is inherited as an autosomal recessive trait.) It is characterized by spontaneous bleeding and other congenital anomalies. The bleeding time is prolonged, the platelet count is decreased, and the bone marrow megakaryocytes vary from decreased to absent.

It is associated with unrepaired chromosome breakage. Congenital anomalies of the inner, middle, and external ear could be causes of the deafness that accompanies this syndrome.

Felty Syndrome

Felty syndrome is a combination of leukopenia, arthritis, and enlarged lymph nodes and spleen.

First Bite Syndrome

First bite syndrome is characterized by facial pain, severe cramping in the parotid region with the first bite of each meal. The pain and spasm diminishes with subsequent bites. The etiology is unknown but may be related to deep lobe parotid or infratemporal fossa lesion or surgery. Treatment included tegretal (carbamazepine), gabapentinoids (calcium channel blockers), amitriptyline (tricyclic antidepressants and anticholinergic). But there is no conclusive evidence as to the effects of these treatments.

First and Second Branchial Arch Syndromes (Hemifacial Microsomia, Lateral Facial Dysplasia)

This disorder consists of a spectrum of craniofacial malformations characterized by asymmetric facies with unilateral abnormalities. The mandible is small with hypoplastic or absent ramus and condyle. Aural atresia, hearing impairment, tissue tags from the tragus to the oral commissure, coloboma of the upper eyelid, malar hypoplasia, and cleft palate also may be present. Cardiovascular, renal, and nervous system abnormalities have been noted in association with this disorder.

Fish Odor Syndrome

Clinical symptoms of this peculiar syndrome consist of a fish odor emanating from the mucus, particularly in the morning. A challenge test with either choline bitartrate or trimethylamine is diagnostic of this disease. Eating non–choline-containing foods usually helps. No long-term effects are known.

Fordyce Disease

Fordyce disease is characterized by pseudocolloid of the lips, a condition marked by the presence of numerous, small yellowish-white granules on the inner surface and vermilion border of the lips. Histologically, the lesions appear as ectopic sebaceous glands.

Foster Kennedy Syndrome

Patients with this disorder show ipsilateral optic atrophy and scotomas and contralateral papilledema occurring with tumors or other lesions of the frontal lobe or sphenoidal meningioma. Anosmia may be seen.

Fothergill Disease

The combination of tic douloureux and anginose scarlatina is characteristic of this disease.

Foville Syndrome

Facial paralysis with ipsilateral paralysis of conjugate gaze and contralateral pyramidal hemiplegia are diagnostic. Tinnitus, deafness, and vertigo may occur with infranuclear involvement. Loss of taste of the anterior two-thirds of the tongue with decreased salivary and lacrimal secretions is seen with involvement of the nervus intermedius.

Frey Syndrome

In the normal person, the sweat glands are innervated by sympathetic nerve fibers. After parotidectomy, the auriculotemporal nerve sends its parasympathetic fibers to innervate the sweat glands instead. The incidence of Frey syndrome after parotidectomy in children has been estimated to be about 20%.

Also called preauricular gustatory sweating, parotidectomy is considered the most common etiology.

Friedreich Disease

The disease consists of facial hemihypertrophy involving the eyelids, cheeks, lips, facial bones, tongue, ears, and tonsils. It may be seen alone or in association with generalized hemihypertrophy.

Garcin Syndrome

Paralysis of cranial nerves III through X, usually unilateral or occasionally bilateral, is observed. It may be the result of invasion by neoplasm, granulomas, or infections in the retropharyngeal space.

Gard-Gignoux Syndrome

Gard-Gignoux syndrome involves paralysis of the 11th and 10th nerves below the nodose ganglion. The cricothyroid function and sensation are normal. The symptoms include vocal cord paralysis and weakness of the trapezius and sternocleidomastoid muscles.

Gardner Syndrome

Gardner syndrome is an autosomal dominant disease whose symptoms include fibroma, osteoma of the skull, mandible, maxilla, and long bones, with epidermoid inclusion cysts in the skin and polyps in the colon. These colonic polyps have a marked tendency toward malignant degeneration.

Gargoylism (Hurler Syndrome)

See Chapter 17.

Gaucher Disease

As an autosomally recessive inherited disorder of lipid metabolism, this syndrome results in a decrease in activity of the glucocerebrosidase. This leads to an increased accumulation of glucocerebrosides, particularly in the retroendothelial system. There are three classifications of the disease: (1) the chronic non-neuronopathic form, characterized by joint pain, aseptic necrosis, pathologic fractures, hepatosplenomegaly, thrombocytopenia, anemia, and leukopenia; (2) the acute neuronopathic Gaucher disease (infantile form), causing increased neurologic complications that often end in death before the first 2 years of life; and (3) the juvenile and less severe forms than the infantile form.

Gerlier Disease

With the presence of vertigo and kubisagari, it is observed among cowherds. It is marked by pain in the head and neck with visual disturbances, ptosis, and generalized weakness of the muscles.

Giant Apical Air Cell Syndrome

Giant apical air cell syndrome, first described in 1982, consists of giant apical air cells, spontaneous CSF rhinorrhea, and recurrent meningitis. It is caused by the constant pounding of the brain against the dura overlying the giant apical air cell, which leads to dural rupture and CSF leak.

Gilles de la Tourette Syndrome

Characterized by chorea, coprolalia, and tics of the face and extremities, it affects children (usually boys 5-10 years old). Repetitive facial grimacing, blepharospasms, and arm and leg contractions may be present. Compulsive grunting noises or hiccupping subsequently become expressions of frank obscenities.

Goldenhar Syndrome

A rare, nonhereditary congenital variant of hemifacial microsomia, Goldenhar syndrome is a congenital syndrome of the first and second arch. It is characterized by underdevelopment of craniofacial structures, vertebral malformations, and cardiac dysfunction. Clinical features of this syndrome are malar and maxillary hypoplasia, poor formation of external auditory canal, supernumerary ear tags and antetragal pits, orbit, enlarged mouths, renal anomalies, and missing growth centers in the condyle, causing delayed eruption of teeth and teeth crowding. Intelligence is usually normal or mildly retarded. Maxillofacial reconstruction in young patients demands consideration of future growth and development. It is also recommended for psychologic reasons as well as reasons involving the proper expansion of the skin that will later aid in further reconstruction. This syndrome is not to be confused with Treacher Collins, Berry, or Franceschetti-Zwahlen-Klein syndromes. These tend to show well-defined genetic patterns (irregular but dominant), whereas Goldenhar syndrome does not.

Goodwin Tumor (Benign Lymphoepithelial Lesion)

This syndrome is characterized by inflammatory cells, lymphocytes, plasma cells, and reticular cells.

Gradenigo Syndrome

Gradenigo syndrome is due to an extradural abscess involving the petrous bone. The symptoms are suppurative otitis, pain in the eye and temporal area, abducens paralysis, and diplopia.

Grisel Syndrome

Grisel syndrome, also known as nasopharyngeal torticollis, is the subluxation of the atlantoaxial joint and is usually associated with children. It is associated with pharyngitis, nasopharyngitis,

adenotonsillitis, tonsillar abscess, parotitis, cervical abscess, and otitis media. This syndrome has been known to occur after nasal cavity inflammation, tonsillectomy, adenoidectomy, mastoidectomy, choanal atresia repair, and excisions of a parapharyngeal rhabdomyosarcoma. Proposals for etiology include overdistention of the atlantoaxial joint ligaments by effusion, rupture of the transverse ligament, excessive passive rotation during general anesthesia, uncoordinated reflex action of the deep cervical muscles, spasm of the prevertebral muscles, ligamentous relaxation from decalcification of the vertebrae, and weak lateral ligaments. Clinical features include spontaneous torticollis in a child, a flexed and rotated head with limited range of motion, flat face, and Sudeck sign (displacement of the spine of the axis to the same side as the head is turned). Treatment includes skeletal skull traction under fluoroscopic control to realign the odontoid process within the transverse ligament sling, followed by 6 to 12 weeks of immobilization. Timely treatment is usually successful.

Guillain-Barré Syndrome

Guillain-Barré syndrome is infectious polyneuritis of unknown etiology ("perhaps" viral) causing marked paresthesias of the limbs, muscular weakness, or a flaccid paralysis. CSF protein is increased without an increase in cell count.

Hallermann-Streiff Syndrome

Hallermann-Streiff syndrome consists of dyscephaly, parrot nose, mandibular hypoplasia, proportionate nanism; hypotrichosis of scalp, brows, and cilia; and bilateral congenital cataracts. Most patients exhibit nystagmus or strabismus. There is no demonstrable genetic basis.

Hanhart Syndrome

A form of facial dysmorphia, Hanhart syndrome is characterized by (1) bird-like profile of face caused by micrognathia, (2) opisthodontia, (3) peromelia, (4) small growth, (5) normal intelligence, (6) branchial arch deformity resulting in conductive hearing loss, (7) tongue deformities and often a small jaw, and (8) possibly some limb defects as well. Ear surgery should be carefully considered because of the abnormal course of the facial nerve due to this syndrome.

Heerfordt Syndrome or Disease

In Heerfordt syndrome, the patient develops uveoparotid fever. Heerfordt syndrome is a form of sarcoidosis (see Chapter 12).

Hick Syndrome

Hick syndrome is a rare condition characterized by a sensory disorder of the lower extremities, resulting in perforating feet and by ulcers that are associated with progressive deafness due to atrophy of the cochlear and vestibular ganglia.

Hippel-Lindau Disease

Hippel-Lindau disease consists of angioma of the cerebellum, usually cystic, associated with angioma of the retina and polycystic kidneys.

Hollander Syndrome

With Hollander syndrome, there is appearance of a goiter during the third decade of life related to a partial defect in the coupling mechanism in thyroxine biosynthesis. Deafness due to cochlear abnormalities is usually related to this.

Homocystinuria

Homocystinuria is a recessive hereditary syndrome secondary to a defect in methionine metabolism with resultant homocystinemia, mental retardation, and sensorineural hearing loss.

Horner Syndrome

The presenting symptoms of Horner syndrome are ptosis, miosis, anhidrosis, and enophthalmos due to paralysis of the cervical sympathetic nerves.

Horton Neuralgia

Patients have unilateral headaches centered behind or close to the eye accompanied or preceded by ipsilateral nasal congestion, suffusion of the eye, increased lacrimation and facial redness, and swelling.

Hunt Syndrome

A. Cerebellar tumor, an intention tremor that begins in one extremity gradually increasing in intensity and subsequently involving other parts of the body
B. Facial paralysis, otalgia, and aural herpes due to disease of both motor and sensory fibers of the seventh nerve
C. A form of juvenile paralysis agitans associated with primary atrophy of the pallidal system

Hunter Syndrome

A hereditary and sex-linked disorder, this incurable syndrome involves multiple organ systems through mucopolysaccharide infiltration. Death, usually by the second decade of life, is often caused by an infiltrative cardiomyopathy and valvular disease leading to heart failure. Physical characteristics include prominent supraorbital ridges, large flattened nose with flared nares, low-set ears, progressive corneal opacities, generous jowls, patulous lips and prognathism, short neck, abdominal protuberance, hirsutism, short stature, extensive osteoarthritis (especially in the hips, shoulders, elbows, and hands), TMJ arthritis, pseudopapilledema, and low-pressure hydrocephalus. Chondroitin sulfate B and heparitin in urine, mental retardation, beta-galactoside deficiency, and hepatosplenomegaly are also features of this syndrome. There is cerebral storage of three gangliosides: GM_1, GM_2, and GM_3. Compressive myelopathy may result from vertebral dislocation. High spinal cord injury is a great complication in surgery. Neurologic development is often slowed or never acquired. Abdominal abnormalities, respiratory infections, and cardiovascular troubles plague the patient.

Immotile Cilia Syndrome

This syndrome appears to be a congenital defect in the ultrastructure of cilia that renders them incapable of movement. Both respiratory tract cilia and sperm are involved. The clinical picture includes bronchiectasis, sinusitis, male sterility, situs inversus, and otitis media. Histologically, there is a complete or partial absence of dynein arms, which are believed to be essential for cilia movement and sperm tail movement. Also no cilia movements were observed in the mucosa of the middle ear and the nasopharynx.

Inversed Jaw-Winking Syndrome

When there are supranuclear lesions of the fifth nerve, touching the cornea may produce a brisk movement of the mandible to the opposite side.

Jackson Syndrome

Cranial nerves X, XI, and XII are affected by nuclear or radicular lesion. There is ipsilateral flaccid paralysis of the soft palate, pharynx, and larynx with weakness and atrophy of the sternocleidomastoid and trapezius muscles and muscles of the tongue.

Jacod Syndrome

Jacod syndrome consists of total ophthalmoplegia, optic tract lesions with unilateral amaurosis, and trigeminal neuralgia. It is caused by a middle cranial fossa tumor involving the second through sixth cranial nerves.

Job Syndrome

Job syndrome is one of the groups of hyperimmunoglobulin E (hyper-IgE) syndromes that are associated with defective chemotaxis. The clinical picture includes fair skin, red hair, recurrent staphylococcal skin abscesses with concurrent other bacterial infections and skin lesions, as well as chronic purulent pulmonary infections and infected eczematoid skin lesions. This syndrome obtained its name from the Biblical passage referring to Job being smitten with boils. It is of interest to the otolaryngologist because of head and neck infections.

Jugular Foramen Syndrome (Vernet Syndrome)

Cranial nerves IX, X, and XI are paralyzed, whereas XII is spared because of its separate hypoglossal canal. Horner syndrome is not present because the sympathetic chain is below the foramen. This syndrome is most often caused by lymphadenopathy of the nodes of Krause in the foramen. Thrombophlebitis, tumors of the jugular bulb, and basal skull fracture can cause the syndrome. The glomus jugulare usually gives a hazy margin of involvement, whereas neurinoma gives a smooth, sclerotic margin of enlargement. The jugular foramen is bound medially by the occipital bone and laterally by the temporal bone. The foramen is divided into anteromedial (pars nervosa) and posterolateral (pars vascularis) areas by a fibrous or bony septum. The medial area transmits nerves IX, X, and XI as well as the inferior petrosal sinus. The posterior compartment transmits the internal jugular vein and the posterior meningeal artery. The right foramen is usually slightly larger than the left foramen.

Kallmann Syndrome

Kallmann syndrome consists of congenital hypogonadotropic eunuchoidism with anosmia. It is transmitted via a dominant gene with variable penetrance.

Kaposi Sarcoma

Patients have multiple idiopathic, hemorrhagic sarcomatosis particularly of the skin and viscera. Radiotherapy is the treatment of choice.

Kartagener Syndrome

The symptoms are complete situs inversus associated with chronic sinusitis and bronchiectasis. It is also called the Kartagener triad.

Cilia and flagella of a patient lack normal dynein side arms of ciliary A-tubes. Deficient mucociliary transport causes sterility in both sexes.

Keratosis Palmaris et Solaris

This disorder is an unusual inherited malformation. If these people live to 65 years of age, 50% to 75% of them develop carcinoma of the esophagus.

Kimura Disease

Kimura disease was first described by Kimura et al in 1949 as a chronic inflammatory condition occurring in subcutaneous tissues, salivary glands, and lymph nodes. Etiology is unknown. Histologically, there is dense fibrosis, lymphoid infiltration, vascular proliferation, and eosinophils. This is different from angiolymphoid hyperplasia with eosinophilia (ALHE). It is much more prevalent in people of Oriental descent. Laboratory studies show eosinophilia and elevated IgE. Differential diagnosis includes ALHE, eosinophilic granuloma, benign lymphoepithelial lesion, lymphocytoma, pyogenic granuloma, Kaposi sarcoma, hamartoma, and lymphoma. Treatment includes corticosteroids, cryotherapy, radiation, and surgery.

The differences between Kimura disease and ALHE are as follows:

Kimura	Age:	30-60 (ALHE age 20-50)
	Sex:	Male (ALHE female)
	Larger lesions	(ALHE < 1 cm)
	Deep	(ALHE superficial)
	More lymphoid follicles than ALHE	
	Fewer mast cells than ALHE	
	Less vascular hyperplasia than ALHE	
	More fibrosis than ALHE	
	More eosinophilia than ALHE	
	More IgE than ALHE	

Kleinschmidt Syndrome

Symptoms include influenzal infections, resulting in laryngeal stenosis, suppurative pericarditis, pleuropneumonia, and occasionally meningitis.

Klinefelter Syndrome

Klinefelter syndrome is a sex chromosome defect characterized by eunuchoidism, azoospermia, gynecomastia, mental deficiency, small testes with atrophy, and hyalinization of seminiferous tubules. The karyotype is usually XXY.

Klinkert Syndrome

Paralysis of the recurrent and phrenic nerves due to a neoplastic process in the root of the neck or upper mediastinum is evidenced. The sympathetics may be involved. (Left-side involvement is more common than right-side involvement.) It can be a part of Pancoast syndrome.

Lacrimoauriculodentodigital Syndrome

Autosomal dominant, occasional middle ear ossicular anomaly with cup-shaped ears, abnormal or absent thumbs, skeletal forearm deformities, sensorineural hearing loss, and nasolacrimal duct obstruction are the characteristics of lacrimoauriculodentodigital syndrome.

Large Vestibular Aqueduct Syndrome

The large vestibular aqueduct as an isolated anomaly of the temporal bone is associated with sensorineural hearing loss. It is more common in childhood than in adulthood. In this syndrome, the rugose portion of the endolymphatic sac is also enlarged. Endolymphatic sac procedures

to improve hearing are not often successful. A vestibular aqueduct is considered enlarged if its anteroposterior diameter on computed tomography (CT) scan is greater than 1.5 mm.

Larsen Syndrome

Larsen syndrome is characterized by widely spaced eyes, prominent forehead, flat nasal bridge, midline cleft of the secondary palate, bilateral dislocation of the knees and elbows, deformities of the hands and feet, and spatula-type thumbs; sometimes tracheomalacia, stridor, laryngomalacia, and respiratory difficulty are present. Therapy includes maintaining adequate ventilation.

Lemierre Syndrome

First discussed by André LeMierre in 1936, usually caused by anaerobic, nonmotile gram-negative rod, *Fusobacterium necrophorum*, Lemierre syndrome can be found in normal flora of oropharynx, gastrointestinal (GI) tract, female genital tract; sensitive to clindamycin and metronidazole, penicillin, and chloramphenicol. Usually in young adults first presenting with oropharynx infection, progress to neck and parapharyngeal abscess, leading to internal jugular and sigmoid sinus thrombosis leading to septic embolism causing septic arthritis, liver and splenic abscess, sigmoid sinus thrombosis findings include headache, otalgia, vertigo, vomiting, otorrhea and rigors, proptosis retrobulbar pain, papilledema, and ophthalmoplegia.

Lermoyez Syndrome

Lermoyez syndrome is a variant of Ménière disease. It was first described by Lermoyez in 1921 as deafness and tinnitus followed by a vertiginous attack that relieved the tinnitus and improved the hearing.

Lethal Midline Granuloma Syndrome

Destroying cartilage, soft tissue, and bone, this disease manifests itself by a number of entities, including idiopathic midline destructive disease, Wegener granulomatosis, polymorphic reticulosis, nasal lymphoma, and non-Hodgkin lymphoma (NHL). High-dose local radiation totaling 5000 rad is the treatment of choice for localized cases. Chemotherapy involving an alkylating agent (cyclophosphamide) is recommended for disseminated cases.

Löffler Syndrome

Löffler syndrome consists in pneumonitis characterized by eosinophils in the tissues. It is possibly of parasitic etiology.

Loose Wire Syndrome

Loose wire syndrome occurs in patients with stapedectomy and insertion of a prosthesis that attaches to the long process of the incus by means of a crimped wire. It is a late complication, occurring on an average 15 years after surgery. A triad of symptoms is present that improves temporarily with middle ear inflation: auditory acuity, distortion of sound, and speech discrimination. Treatment in revision surgery involves finding the loose wire attachment at the incus and tightening that wire to allow the incus and prosthesis to move as one.

Louis-Bar Syndrome

This autosomal recessive disease presents as ataxia, oculocutaneous telangiectasia, and sinopulmonary infection. It involves progressive truncal ataxia, slurred speech, fixation nystagmus, mental deficiency, cerebellar atrophy, deficient immunoglobulin, and marked frequency of lymphoreticular malignancies. The patient rarely lives past age 20.

Maffucci Syndrome

Maffucci syndrome is characterized by multiple cutaneous hemangiomas with dyschondroplasia and often enchondroma. The origin is unknown, and it is not hereditary. Signs and symptoms of this syndrome usually appear during infancy. It equally affects both sexes and has no racial preference. The dyschondroplasia may cause sharp bowing or an uneven growth of the extremities as well as give rise to frequent fractures. Five to 10% of Maffucci syndrome patients have head and neck involvement giving rise to cranial nerve dysfunction and hemangiomas in the head and neck area. The hemangiomas in the nasopharynx and larynx could cause airway compromise as well as deglutition problems. Fifteen to 20% of these patients later undergo sarcomatous degeneration in one or more of the enchondromas. The percentage of malignant changes is greater in older patients, with the percentage of malignant degeneration approaching 44% in patients older than 40.

This syndrome is not to be confused with Klippel-Trenaunay syndrome, which causes no underdeveloped extremities, Sturge-Weber syndrome, or von Hippel-Lindau syndrome. No treatment is known for this syndrome, although surgical procedures to treat the actual deformities are sometimes necessary.

Mal de Debarquement Syndrome

Mal de Debarquement syndrome (or MDDS) is an imbalance or rocking sensation that occurs after prolonged exposure to motion (most commonly after a sea cruise or a long airplane flight). Travelers often experience this sensation temporarily after disembarking, but in the case of MDDS sufferers, it can persist for 6 to 12 months or even many years in some cases.

The imbalance is generally not associated with any nausea, nor is it alleviated by typical motion sickness drugs such as scopolamine or meclizine. Symptoms are usually most pronounced when the patient is sitting still; in fact, the sensations are usually minimized by actual motion such as walking or driving.

The functional cause of MDDS is unknown. (Vestibular and CNS tests for MDDS patients invariably turn out normal results.)

Speculation about the cause of MDDS includes the following:

- Psychiatric condition (particularly linked to depression)
- A hormonal-related condition (may occur more often in females)
- Otolith organ or CNS abnormalities
- Some link to a variant of migraine

Diagnosis of MDDS is generally a process of exclusion.

The medical literature describes MDDS as *self-limiting* condition.

Valium and other derivatives (particularly *Klonopin*) have been known to help alleviate some of the severe symptoms in MDDS patients, but there is always a worry that these are habit forming and may prolong the eventual disappearance of the condition.

In general, physical activity is recommended for vestibular rehabilitation.

Marcus Gunn Syndrome (Jaw-Winking Syndrome)

Marcus Gunn syndrome results in an increase in the width of the eyelids during chewing. Sometimes the patient experiences rhythmic elevation of the upper eyelid when the mouth is open and ptosis when the mouth is closed.

Marie-Strümpell Disease

Marie-Strümpell disease is rheumatoid arthritis of the spine.

Masson Tumor

Masson tumor is intravascular papillary endothelial hyperplasia caused by excessive proliferation of endothelial cells. It is a benign condition. Differential diagnosis includes angiosarcoma, Kaposi sarcoma, and pyogenic granuloma.

Melkersson-Rosenthal Syndrome

(Triad: Recurrent orofacial swelling, one or more episodes of facial paralysis, and lingua plicata.) Melkersson-Rosenthal syndrome is a congenital disease of unknown etiology, and it manifests as recurring attacks of unilateral or bilateral facial paralysis (see Chapter 21), swelling of the lips, and furrowing of the tongue. It is associated with high serum levels of angiotensin-converting enzyme during affliction, and also known as orofacial granulomatosis, cheilitis granulomatosis, Scheuermann glossitis granulomatosis, and Miescher cheilitis.

Treatment should focus on facial paralysis and edema. Steroids and facial nerve decompression have had limited success.

Meyenburg Syndrome (Familial Myositis Fibrosa Progressiva)

Meyenburg syndrome is a disease in which the striated muscles are replaced by fibrosis. Fibrosarcoma rarely originates from this disease.

Middle Lobe Syndrome

Middle lobe syndrome is a chronic atelectatic process with fibrosis in one or both segments of the middle lobe. It is usually secondary to obstruction of the middle lobe bronchus by hilar adenopathy. The hilar adenopathy may be transient, but the bronchiectasis that resulted persists. Treatment is surgical resection.

Mikulicz Disease

The symptoms characteristic of Mikulicz disease (swelling of the lacrimal and salivary glands) occur as complications of some other disease, such as lymphocytosis, leukemia, or uveoparotid fever.

Millard-Gubler Syndrome

Patients present with ipsilateral paralysis of the abducens and facial nerves with contralateral hemiplegia of the extremities due to obstruction of the vascular supply to the pons.

Möbius Syndrome

Möbius syndrome is a nonprogressive congenital facial diplegia (usually bilateral) with unilateral or bilateral loss of the abductors of the eye, anomalies of the extremities, and aplasia of the brachial and thoracic muscles. It frequently involves other cranial nerves. Saito showed evidence that the site of nerve lesions is in the peripheral nerve. The etiology could be CNS hypoplasia, primary peripheral muscle defect with secondary nerve degeneration, or lower motor neuron involvement.

Morgagni-Stewart-Morel Syndrome

Morgagni-Stewart-Morel syndrome occurs in menopausal women and is characterized by obesity, dizziness, psychologic disturbances, inverted sleep rhythm, and hyperostosis frontalis interna. Treatment is supportive.

Multiple Endocrine Adenomatosis

Multiple Endocrine Adenomatosis Type IIA (Sipple Syndrome) Sipple syndrome is a familial syndrome consisting of medullary carcinoma of the thyroid, hyperparathyroidism, and pheochromocytoma.

Multiple Endocrine Adenomatosis Type IIB This multiple endocrine adenomatosis (MEA) variant consists of multiple mucosal neuromas, pheochromocytoma, medullary carcinoma of the thyroid, and hyperparathyroidism. This syndrome is inherited in an autosomal dominant pattern. Mucosal neuromas principally involve the lips and anterior tongue. Numerous white medullated nerve fibers traverse the cornea to anastomose in the pupillary area.

Munchausen Syndrome

Munchausen syndrome was named after Baron Hieronymus Karl Freidrich von Münchausen (1720-1791) by Asher in 1951. The integral features of this syndrome include the following:

A. A real organic lesion from the past that has left some genuine signs but is causing no organic symptoms.
B. Exorbitant lying with dramatic presentation of nonexistent symptoms.
C. Traveling widely with multiple hospitalizations.
D. Criminal tendencies.
E. Willingness to undergo painful and dangerous treatment.
F. Presenting challenging illnesses for treatment.
G. Unruly behavior during hospital stays and early self-discharge without prior approval.
H. Patients often inflict pain on their own children and forcibly create symptoms to indirectly receive hospital treatment.

The patients usually go from one medical center to another to be admitted with dramatic presentations of nonorganic symptoms related to a real organic lesion on the past medical history.

Nager Syndrome (Acrofacial Dysostosis)

Acrofacial dysostosis patients have facies similar to those seen with Treacher Collins syndrome. They also present with preaxial upper limb defects, microtia, atresia of the external auditory canals, and malformation of the ossicles. Conductive and mixed hearing losses may occur.

Nager de Reynier Syndrome

Hypoplasia of the mandible with abnormal implantation of teeth associated with aural atresia characterizes this syndrome.

Neurofibromatosis (von Recklinghausen Disease)

Salient Features

A. Autosomal dominant.
B. Mental retardation common in families with neurofibromatosis.
C. Arises from neurilemmal cells or sheath of Schwann and fibroblasts of peripheral nerves.
D. Café au lait spots—giant melanosomes (presence of six or more spots > 1.5 cm is diagnostic of neurofibromatosis even if the family history is negative).
E. Of all neurofibromatoses, 4% to 5% undergo malignant degeneration with a sudden increase in growth of formerly static nodules. These nodules may become neurofibrosarcomas, and they may metastasize widely.

External Features

A. Café au lait spots.
B. Fibromas.

Internal Features

A. Pheochromocytoma.
B. Meningioma.
C. Acoustic neuroma: bilateral in type 2 neurofibromatosis.
D. GI bleeding.
E. Intussusception bowel.
F. Hypoglycemia (intraperitoneal fibromas).
G. Fibrous dysplasia.
H. Subperiosteal bone cysts.
I. Optic nerve may be involved, causing blindness and proptosis.
J. May present with macroglossia.
K. May involve the parotid or submaxillary gland.
L. The nodules may be painful.
M. Nodules may enlarge suddenly if bleeding of the tumor occurs or if there is malignant degeneration.

The treatment is only to relieve pressure from expanding masses. It usually does not recur if the tumor is completely removed locally.

Nothnagel Syndrome

The symptoms include dizziness, a staggering and rolling gait with irregular forms of oculomotor paralysis, and nystagmus often is present. This syndrome is seen with tumors of the midbrain.

Oculopharyngeal Syndrome

Oculopharyngeal syndrome is characterized by hereditary ptosis and dysphagia, and is an autosomal dominant disease having equal incidence in both sexes. It is related to a high incidence of esophageal carcinoma. Age of onset is between 40 and 50 years, and it is particularly common among French Canadians. Marked weakness of the upper esophagus is observed together with an increase in serum creatinine phosphokinase. It is a myopathy and not a neuropathy. Treatment includes dilatation and cricopharyngomyotomy.

Ollier Disease

Ollier disease consists of multiple chondromatosis, 10% of which is associated with chondrosarcoma.

Ondine Curse

Failure of respiratory center automaticity with apnea, especially evident during sleep, is symptomatic. Also known as the alveolar hypoventilation syndrome, it may be associated with increased appetite and transient central diabetes insipidus. Hypothalamic lesions are thought to be the cause of this disorder.

Oral-Facial-Digital Syndrome I

See Chapter 17 for oral-facial-digital syndrome II.

A lethal trait in men, it is inherited as an X-linked dominant trait limited to women. Symptoms include multiple hyperplastic frenula, cleft tongue, dystopia canthorum, hypoplasia of the