

**Chapter 23: Syndromes and Eponyms**

**Syndromes and Diseases**

**Adult Respiratory Distress Syndrome (ARDS)**

This syndrome is characterized by a delay in onset (12-24 hours) following injury, shock, and/or successful resuscitative effort. Septic shock, extrathoracic trauma, 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 and/or microvascular hemorrhage.

**Adie's Syndrome**

Adie's syndrome is characterized by decreased pupillary reaction and deep tendon reflex. The etiology is unknown.

**Albright's Syndrome**

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

**Aldrich's Syndrome**

Thrombocytopenia, eczema, and recurrent infections in 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's Syndrome**

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

**Ascher's Syndrome**

A combination of blepharochalasis, double lip, and goiter.

**Auriculotemporal Syndrome (Frey's Syndrome)**

This 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 following parotidectomy the parasympathetic fibers of the IX nerve innervate the sweat glands. It has been estimated that 20% of parotidectomy in children results 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. This syndrome is caused by involvement of the nucleus ambiguus or the vagus nerve along with the cranial portion of the XI nerve.

### **Babinski-Nageotte Syndrome**

This 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 will occur. There is also ipsilateral loss of taste on the posterior third of the tongue, loss of pain and temperature sensation around the face, cerebellar asynergia, Horner's syndrome with contralateral spastic hemiplegia, and loss of proprioceptive and tactile sensation.

### **Baelz's Syndrome**

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

### **Barany's Syndrome**

This 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.

### **Barre-Lieou Syndrome**

Occipital headache, vertigo, tinnitus, vasomotor disorders, and facial spasms 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's Syndrome**

Barrett's syndrome is characterized by esophagitis due to change of 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 for this syndrome. It most commonly affects excitable elderly persons.

### **Basal Cell Nevoid Syndrome**

This familial syndrome, non-sex-linked, autosomal dominant with high penetrance and variable expressivity, manifests itself 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's Disease)**

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

### **Beckwith's Syndrome**

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

### **Behcet's 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 will help.

### **Besnier-Beck-Schaumann's Syndrome**

Sarcoidosis.

### **Bogorad's Syndrome**

This also is 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 fibers to the lacrimal gland instead of to the salivary gland.

### **Bonnet's Syndrome**

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

### **Bonnier's Syndrome**

This syndrome is caused by a lesion of Deiter's nucleus and its connection. Its symptoms include ocular disturbances, i.e. paralysis of accommodation, nystagmus, diplopia, deafness, nausea, thirst, 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's disease.

### **Bourneville's Syndrome**

This is a familial disorder. Its symptoms include polyps of the skin, harelip, moles, spina bifida, and microcephaly.

### **Bowen's Disease**

It 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.

### **Briquet's Syndrome**

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

### **Brissaud-Marie Syndrome**

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

### **Brun's 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's Dermatitis**

This dermatitis appears as an eruption of the external ear consisting of red papules and vesicles after exposure to sunlight. The rash usually resolves spontaneously.

### **Caffey's Disease (Infantile Cortical Hyperostosis)**

Of family tendency, its onset is usually in the first year of life. It is characterized by hyperirritability, fever, and hard nonpitting edema that overlies 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 working in high air pressures when they 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, 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 the necrosis of the organ of Corti. There is a question of rupture of the round window membrane; hemotympanum and eustachian tube obstruction may occur.

### **Campomelic Syndrome (Greek = curvature of the extremities)**

The syndrome is characterized by dwarfism, craniofacial anomalies, 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 prominent forehead with flat facies with a broad nasal bridge, and low-set ears, cleft palate, mandibular hypoplasia, tracheobronchial malacia which 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 scala communis. The vestibule and the semicircular canal were deformed by bone invasion.

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

### **Cannon's 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. The 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 upon it causes a marked fall in blood pressure due to vasodilation and cardiac slowing. Symptoms include syncope, convulsions, and heart block.

### **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 (V1) 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 V1. 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 while the internal carotid artery is medial to it.

### **Cestan-Chenais Syndrome**

This 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's syndrome. There is contralateral hemiplegia and diminished proprioception and tactile sensation.

### **Champion-Cregah-Klein Syndrome**

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

### **Chapple's Syndrome**

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

### **Chediak-Higashi Syndrome**

This is the result of an autosomal recessive trait characterized by albinism, photophobia, nystagmus, hepatosplenomegaly, anomalous cellular granules, and development of lymphoma. These patients usually die in childhood of fulminant infections.

### **Cleft Lip-Palate and Congenital Lip Fistulas**

This syndrome is transmitted in an autosomal-dominant manner with 80% penetrance and occurs in 1:100,000 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 found with cleft lip-palate (67% of cases). Associated anomalies of the extremities may include talipes equinovarus, syndactyly and popliteal pterygia - Champion-Cregah-Kelin syndrome. Congenital lip pits have also been seen in association with oral-facial-digital syndrome.

### **Cogan's Syndrome**

Nonsyphilitic interstitial keratitis and vestibulo-auditory symptoms are characteristic of this 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, semicircular canals, and inflammation of the spiral ligament. Treatment with steroids has been advocated.

### **Collet-Sicard Syndrome**

The IX, X, XI, and XII nerves are involved with normal sympathetic nerves. The etiology is usually a meningioma or other lesion involving the nerves in the posterior cranial fossa.

### **Costen's Syndrome**

Costen's syndrome is a temporal-mandibular joint abnormality, usually due to impaired bite and characterized by tinnitus, vertigo, pain in the frontal, parietal, and occipital areas with a blocked feeling and pain in the ear. After a careful work-up 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 diving 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 ginglymo-arthroïdal joint with hinge and transverse movements. The key supporting ligament of the TMJ is the temporomandibular ligament. The boundaries of the glenoid fossa are:

- 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) while it has a hinge motion between the disc and the condyle (lower compartment). It is innervated by the auriculotemporal nerve, masseter nerve, lateral pterygoid nerve and the 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 while the masseter, medial pterygoid, and temporalis muscles act as elevators. All these muscles are innervated by V3 (see Chapter on Facial Trauma for muscles of the mandible). The sphenomandibular and stylomandibular ligaments have no function in the TMJ articulation.

### **Cowden's Syndrome**

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, liver and CNS abnormalities.

### **Cri du Chat Syndrome**

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's Disease** (see Chap. 9)

### **Curtius' Syndrome**

This is a form of hypertrophy that may involve a single small part of the body, or an entire system (i.e. muscular, nervous, or skeletal systems). It is also known as congenital facial hypertrophy.

### **Dandy's 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's 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's 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.

### **Dejerine's 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-Richet Syndrome**

This 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's Syndrome**

This is an autosomal-recessive disorder associating (DI) diabetes insipidus, (DM) diabetes mellitus, (OA) optic atrophy, and (D) deafness. Diabetes mellitus is usually juvenile in onset and is 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 supraoptico-hypophyseal tract. The hearing loss is sensorineural, 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's Syndrome**

Lischaneri reported three categories of this syndrome:

1. Third and fourth pharyngeal pouch syndrome which is characterized by cardiovascular and craniofacial anomalies as well as abdominal visceral abnormalities.
2. DiGeorge syndrome (thymus agenesis).
3. 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 Mundini type as well as absence of hair cells in the hook region, hypertelorism with nasal cleft, shortened philtrum, and micrognathia. Other middle ear anomalies include absence of stapedial muscle, hypoplastic facial nerve, and absent oval window. Most of the findings are symmetrical.

### **Down's Syndrome** (see Trisomy in Chap. 9)

### **Dysphagia Lusoria**

This dysphagia 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's 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.

### **Ectodermal Dysplasia, Hydrotic** (see Chap. 9)

### **Ectodermal Dysplasia, Hypohidrotic**

This syndrome consists of hypodontia, hypotrichosis, and hypohidrosis. Principally, the structures involved are of ectodermal derivatives. Eyelashes and especially the 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 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's Syndrome**

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

### **Elschnig's 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 primary empty sella syndrome is due to a congenital absence of the diaphragm sella, with gradual enlargement of the sella secondary to pulsations of the brain.

The secondary empty sella syndrome may be due to necrosis of an existing pituitary tumor, after surgery, postradiation directed at the pituitary, or with pseudotumor cerebri.

### **Face-Hand Syndrome**

This 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's 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. It rarely occurs in adults. A variant of this 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.

### **Felty's Syndrome**

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

### **First and Second Branchial Arch Syndromes**

#### **(Hemifacial Microsomia, Lateral Facial Dysplasia)**

This 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.

### **Fordyce's Disease**

The 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 scotoma and contralateral papilledema occurring with tumors or other lesions of the frontal lobe or sphenoidal meningioma. Anosmia may be seen.

### **Fothergill's Disease**

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

### **Foville's 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's 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's syndrome postparotidectomy in children has been estimated to be about 20%.

Inferior salivatory nucleus (parasympathetic of the medulla) -->IX--> Jacobson's nerve --> otic ganglion --> auriculotemporal nerve --> parotid VII --> lesser superficial petrosal --> otic ganglion.

### **Friedreich's 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's Syndrome**

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

### **Gard-Gignoux Syndrome**

This syndrome involves paralysis of the XI nerve and the X nerve below the nodose ganglion. The cricothyroid function and sensation are normal. The symptoms include vocal cord paralysis and weakness of the trapezius and sternocleidomastoid muscle.

### **Gardner's Syndrome**

An autosomal-dominant disease, its 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's Syndrome)**

(See Chap. 9)

### **Gerlier's Disease**

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

### **Gilles de la Tourette's Syndrome**

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

### **Goldenhar's Syndrome**

This is a variant of the hemifacial microsomia which is one of the more common congenital syndromes of the first and second arches. Goldenhar's syndrome manifests in varying degrees of underdevelopment of the craniofacial structures. Major deformities involve the mandible, the ear, the orbit, and the vertebral column.

### **Goodwin's Tumor (Benign Lymphoepithelial Lesion)**

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

### **Gradenigo's Syndrome**

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

### **Grisel's Disease**

This is a nasopharyngeal disease causing nontraumatic subluxation of the atlas. This disorder usually occurs after tonsillectomy or nasal cavity inflammation. It also is known as nasopharyngeal torticollis.

### **Guillain-Barre Syndrome**

An infectious polyneuritis of unknown etiology (? viral) causing marked paresthesias of the limbs, muscular weakness or a flaccid paralysis. CSF protein is increased without an increase in cell count.

### **Hallerman-Streiff Syndrome**

This syndrome consists of dyscephaly, parrot nose, mandibular hypoplasia, proportionate nanism, hypotrichosis of scalp, brows, and cilia, and bilateral congenital cataracts. The majority of patients exhibit nystagmus or strabismus. There is no demonstrable genetic basis.

### **Heerfordt's Syndrome or Disease**

It gives uveoparotid fever and is a form of sarcoidosis (see Chap. 30).

### **Hick's Syndrome**

A rare condition characterized by a sensory disorder of the lower extremities resulting in perforating foot ulcers associated with progressive deafness due to atrophy of the cochlear and vestibular ganglia.

### **Hippel-Lindau Disease**

Angioma of the cerebellum, usually cystic, associated with angioma of the retina and polycystic kidneys.

### **Hollander's Syndrome**

With this syndrome there is appearance of a goiter in 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.

### **Homocystinuria**

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

### **Horner's Syndrome**

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

### **Horton's Neuralgia**

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

### **Hunt's Syndrome**

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

### **Immotile Cilia Syndrome**

This appears to be a congenital defect in the ultrastructure of cilia that renders them incapable of movement. Both the respiratory tract cilia and the 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 V nerve, touching the cornea may produce a brisk movement of the mandible to the opposite side.

### **Jackson's Syndrome**

Cranial nerves X, XI, and XII are affected by a 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's Syndrome**

This 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's Syndrome**

This is one of the group of hyper-IgE syndromes, which 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's being smitten with boils by Satan. It is of interest to the otolaryngologist because of head and neck infections.

### **Vernet's Syndrome**

Cranial nerves IX, X, and XI are paralyzed while XII is spared because of its separate hypoglossal canal. Horner's syndrome is not present since 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 the jugular bulb, and basal skull fracture can cause the syndrome. The glomus jugulare usually gives a hazy margin of involvement while 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.

### **Kallman's Syndrome**

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

### **Kaposi's Sarcoma**

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

### **Kartagener's Syndrome**

The symptoms are complete situs inversus associated with chronic sinusitis and bronchiectasis. This also is called Kartagener's triad.

### **Keratosis Palmaris or Solaris**

This is an unusual inherited malformation. If these people live to 65 years old 50-75% of them would have developed carcinoma of the esophagus.

### **Kleinschmidt's Syndrome**

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

### **Klinefelter's Syndrome**

This 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's 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. The left side involvement is more common than right side involvement. This can be a part of Pancoast's syndrome.

### **Larsen's Syndrome**

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



### **Lermoyez's Syndrome**

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

### **Loeffler's Syndrome**

This consists of pneumonitis characterized by eosinophiles in the tissues. This is possibly of parasitic etiology.

### **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 20 years of age.

### **Maffucci's Syndrome**

This 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 early in life 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 giving rise to frequent fractures. Five to ten percent of Maffucci's 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 twenty percent of these patients later undergo sarcomatous degeneration in one or more of the enchondromas. The malignant changes are greater in the older patients. The percentage of malignant degeneration approaches 44% in patients over 40 years old.

### **Markus-Gunn Syndrome (Jaw Winking Syndrome)**

There is increase in 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-Strumpell Disease**

This is rheumatoid arthritis of the spine.

### **Melkersson-Rosenthal Syndrome**

A congenital disease of unknown etiology, it manifests as recurring attacks of unilateral or bilateral facial paralysis (see Chap. 10), swelling of the lips, and furrowing of the tongue.

### **Middle Lobe Syndrome**

This 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 by surgical resection.

### **Mikulicz's Disease**

The symptoms characteristic of Mikulicz's disease (swelling of lacrimal and salivary gland) occurs as a complication of some other disease such as lymphocytosis, leukemia, or uveoparotid fever (see Chap. 17).

### **Millard-Gubler Syndrome**

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

### **Möbius Syndrome**

This is a congenital facial diplegia (usually bilateral), with unilateral or bilateral loss of the abductors of the eye, anomalies of the extremities, aplasia of the brachial and thoracic muscles, and frequently involves other cranial nerves. The etiology could be a primary muscle defect or neurogenic in nature. Saito showed evidence that the site of nerve lesions is in the peripheral nerve.

### **Morgagni-Stewart-Morel Syndrome**

It occurs in menopausal women and is characterized by obesity, dizziness, psychological disturbances, inverted sleep rhythm, and hyperostosis frontalis interna. Treatment is supportive.

### **Multiple Endocrine Adenomatosis**

#### MEA type IIA Sipple's Syndrome

A familial syndrome consisting of medullary carcinoma of the thyroid, hyperparathyroidism, and pheochromocytoma.

#### MEA Type IIB

This 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's Syndrome**

This syndrome was named after Baron Hieronymus Carl Friedrich von Munchausen (1720-1791) by Asher in 1951. The integral features of this syndrome are:

1. A real organic lesion from the past which has left some genuine signs but is causing no organic symptoms.
2. Exorbitant lying with dramatic presentation of nonexistent symptoms.
3. Travelling widely with multiple hospitalizations.
4. These patients have criminal tendencies.

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

### **Myenburg's Syndrome (Familial Myositis Fibrosa Progressiva)**

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

### **Nager's Syndrome**

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

### **Nager-de Reynier Syndrome**

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

### **Neurofibromatosis (von Recklinghausen's Disease)**

Salient features:

1. Autosomal dominant.
2. Mental retardation common in families with neurofibromatosis.
3. Arises from neurilemmal cells or sheath of Schwann and fibroblasts of peripheral nerves.
4. Cafe-au-lait spots - giant melanosomes. The presence of six or more spots greater than 1.5 cm is diagnostic of neurofibromatosis even if the family history is negative.
5. Of all neurofibromatosis 4-5% undergo malignant degeneration with sudden increase of growth of formerly static nodules. These may become neurofibrosarcomas. They may metastasize widely.

External features:

1. Cafe-au-lait spots.
2. Fibromas.

Internal features:

1. Pheochromocytoma.
2. Meningioma.
3. Acoustic neurinoma: often bilateral.
4. GI bleeding.
5. Intussusception bowel.
6. Hypoglycemia (intraperitoneal fibromas).
7. Fibrous dysplasia.
8. Subperiosteal bone cysts.
9. The optic nerve may be involved causing blindness and proptosis.
10. May present with macroglossia.
11. May involve the parotid or submaxillary gland.
12. The nodules may be painful.
13. Nodules may enlarge suddenly if bleeding of the tumor occurs or if there is malignant degeneration.

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

### **Nothnagel's Syndrome**

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

### **Oculopharyngeal Syndrome**

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

### **Odiene's Curse**

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

### **Ollier's Disease**

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

### **Oral-Facial-Digital Syndrome I** (see Ch. 9 for oral-facial-digital syndrome II)

A lethal trait in males, it is inherited as an X-linked dominant trait limited to females. Symptoms include multiple hyperplastic frenula, cleft tongue, dystopia canthorum, hypoplasia of the nasal alar cartilages, median cleft of the upper lip, asymmetric cleft palate, digital malformation and mild mental retardation. About 50% of the patients have hamartoma between the lobes of the divided tongue. This mass consists of fibrous connective tissue, salivary gland tissue, few striated muscle fibers, and rarely cartilage. One-third of the patients present with ankyloglossia.

### **Orbital Apex Syndrome**

This involves the nerves and vessels pathing through the superior sphenoid fissure and the optic foramen with paresis of cranial nerves III, IV, and VI. External ophthalmoplegia is associated with internal ophthalmoplegia with a dilated pupil which does not react to either light or convergence. Ptosis as well as periorbital edema are due to the IV nerve paresis. Sensory changes are secondary to the lacrimal frontal nasal ciliary nerves as well as the three branches of the ophthalmic nerve. The optic nerve usually is involved.

### **Ortner's Syndrome**

Cardiomegaly associated with laryngeal paralysis secondary to compression of the recurrent laryngeal nerve is observed.

### **Osler-Weber-Rendu Disease**

#### **Hereditary Hemorrhagic Telangiectasia**

It is an autosomal-dominant disease in which the heterozygote lives to adult life while the homozygous state is lethal at an early age. The patient has punctate hemangioma (elevated, dilated, capillaries and venules) in the mucous membrane of the lips, tongue, mouth, gastrointestinal tract, etc. Pathologically, these are vascular sinuses of irregular size and shape lined by a thin layer of endothelium. The muscular and elastic coats are absent. Because of their thin walls these vascular sinuses bleed easily and because of the lack of muscular coats, this bleeding is difficult to control. The patient has normal blood elements and no coagulation defect. The other blood vessels are normal as well. If a person with this disease marries a normal person, what are the chances that the offspring will have this condition? Since the patient with this disease is an adult, we can assume that he is heterozygous since the homozygote dies early in life. Therefore, the child will have a 50% chance of having this hereditary disease.

### **Paget's Disease (Osteitis Deformans)** (See Chap. 9)

This term also is used to characterize a disease of elderly women with an infiltrated, eczematous lesion surrounding the nipple and areola associated with subjacent intraductal carcinoma of the breast.

### **Paget's Osteitis**

This is related to sarcomas.

### **Pancoast's Syndrome** (See Chap. 28)

### **Peutz-Jeghers' Syndrome**

The patient has pigmentation of the lips, oral mucosa, and benign polyps of the gastrointestinal tract. Granulosa theca cell tumors have been reported in females with this syndrome.

### **Pheochromocytoma**

Pheochromocytoma is associated with neurofibromatosis, cerebellar hemangioblastoma, ependymoma, astrocytoma, meningioma, spongioblastoma, multiple endocrine adenoma, or with medullary carcinoma of the thyroid. Pheochromocytoma with or without the above tumors may be inherited as an autosomal-dominant trait. Some patients have megacolon, other suffer neurofibromatosis of Auerbach's and Meissner's plexuses.

### **Pierre-Robin Syndrome**

This consists of glossoptosis, micrognathia, and cleft palate. There is no sex predilection. The etiology is believed to be intrauterine insult at the fourth month of gestation or perhaps hereditary. Two-thirds of the cases are associated with ophthalmologic difficulties (i.e. detached retina or glaucoma), one-third are associated with otologic problems (i.e. chronic otitis media and low-set ears). Mental retardation is present occasionally. If the patient lives past 5 years old, he can lead a fairly normal life (see Chap. 9). The symptoms are choking and aspiration as a result of negative pressure created by excessive inspiratory effort. Passing of a NA tube may alleviate the negative pressure. Aerophagia has to be treated to prevent vomiting, airway compromise, and aspiration. Tracheotomy may not be the answer.

### **Plummer-Vinson Syndrome (Patterson-Kelly Syndrome)**

Symptoms include dysphagia due to degeneration of the esophageal muscle, atrophy of the papillae of the tongue, as well as microcytic hypochromic anemia. Achlorhydria, glossitis, pharyngitis, esophagitis, and fissures at the corner of the mouth also are observed. The prevalence of this disease is higher in females than in males, and usually presents in patients who are in their fourth decade. Treatment consists of iron administration, with esophagoscopy for dilatation and to rule out carcinoma of the esophagus, particularly at the postcricoid region. Pharyngoesophageal webs or stenosing may be noted.

This disease is to be contrasted with pernicious anemia which is a megaloblastic anemia with diarrhea, nausea and vomiting, neurologic symptoms, enlarged spleen, and achlorhydria. Pernicious anemia is secondary to failure of the gastric fundus to secrete intrinsic factors necessary for vitamin B12 absorption. Treatment consists of IM B12 (riboflavin).

Folic acid deficiency also gives rise to megaloblastic anemia, cheilosis, glossitis, ulcerative stomatitis, pharyngitis, esophagitis, dysphagia, and diarrhea. No neurologic symptoms and no achlorhydria are present. Treatment is through administration of folic acid.

### **Pyknodysostosis**

This is a syndrome consisting of dwarfism, osteopetrosis, partial agenesis of the terminal phalanges of the hands and feet, cranial anomalies (persistent fontanelles), frontal and occipital bossing and hypoplasia of the angle of the mandible. The facial bones are usually underdeveloped with pseudoprognathism. The frontal sinuses are consistently absent and the other paranasal sinuses are hypoplastic. The mastoid air cells often are not pneumatized. Toulouse-Lautrec probably had this disease.

### **Reichert's Syndrome**

Neuralgia of the glossopharyngeal nerve, usually precipitated by movements of the tongue or throat.

### **Reiter's Syndrome**

Arthritis, urethritis, conjunctivitis.

### **Riedel's Struma**

This is a form of thyroiditis seen most frequently in middle-aged females manifested by compression of surrounding structures (i.e. trachea). There is loss of the normal thyroid lobular architecture and replacement with collagen and lymphocyte infiltration.

### **Rivalta's Disease**

This is an actinomycotic infection characterized by multiple indurated abscesses of the face, neck, chest, and abdomen that discharge through numerous sinus tracts.

### **Rollet's Syndrome (Orbital Apex - Sphenoidal Syndrome)**

It is caused by lesions of the orbital apex which cause paralysis of cranial nerves III, IV, and VI. This syndrome is characterized by ptosis, diplopia, ophthalmoplegia, optic atrophy, hyperesthesia or anesthesia of the forehead, upper eyelid, and cornea, and retrobulbar neuralgia. Exophthalmos and papilledema may occur.

### **Romberg's Syndrome**

Progressive atrophy of tissues on one side of the face occasionally extending to other parts of the body, that may involve the tongue, gums, soft palate, cartilages of the ear, nose, and larynx. Pigmentation disorders, trigeminal neuralgia, and ocular complications may be seen.

### **Rutherford's Syndrome**

A familial oculodental syndrome characterized by corneal dystrophy, gingival hyperplasia, and failure of tooth eruption.

### **Scalenus Anticus Syndrome**

The symptoms are identical to those of cervical rib syndrome. In scalenus anticus syndrome, symptoms are caused by compression of the brachial plexus and subclavian artery against the first thoracic rib, probably as the result of spasms of the scalenus anticus muscle bringing pressure on the brachial plexus and the subclavian artery. Any pressure on the sympathetic nerves may cause vascular spasm resembling Raynaud's disease.

### **Schafer's Syndrome**

Hereditary mental retardation, sensorineural hearing loss, prolinemia, hematuria, and photogenic epilepsy are characteristic. This syndrome is due to a deficiency of proline oxidase with resultant build up of the amino acid proline.

### **Schaumann's Syndrome**

This is generalized sarcoidosis (see Chap. 30).

### **Schmidt's Syndrome**

Unilateral paralysis of a vocal cord, of the velum palati, of the trapezius, and of the sternocleidomastoid muscles is found. The lesion is located in the caudal portion of the medulla and is usually of vascular origin.

### **Seckel's Syndrome**

A disorder which consists of dwarfism associated with a bird-like facies, beaked nose, micrognathia, palate abnormalities, low-set lobeless ears, antimongoloid slant of the palpebral fissures, clinodactyly, mental retardation, and bone disorders.

### **Sheehan's Syndrome**

Ischemic necrosis of the anterior pituitary associated with postpartum hypotension characterizes this syndrome.



### **Sjögren's Syndrome (Sicca Syndrome)**

Manifesting as keratoconjunctivitis sicca, dryness of the mucous membranes, telangiectasias or purpuric spots on the face, and bilateral parotid enlargement, this syndrome is often seen in menopausal women associated with rheumatoid arthritis, Raynaud's phenomenon, and dental caries. Changes in the lacrimal and salivary glands resemble those of Mikulicz's disease. Some attribute this syndrome to vitamin A deficiency. A positive LE preparation, rheumatoid factor, and an abnormal protein can be identified in this disorder.

### **Sleep Apnea Syndrome**

The definition of apnea is a "cessation of airflow greater than 10 seconds in duration". Sleep apnea syndrome is said to occur when at least 30 episodes of apnea occur within a 7-hour period. Another definition of sleep apnea syndrome is when 1% of the sleeping time is spent in apnea. The cause of sleep apnea is unclear. Some people believe it is of central origin; others think that it may be aggravated by hypertrophied and occluding tonsils and adenoids. Some investigators classify sleep apnea into central apnea, upper airway apnea, and mixed apnea. Monitoring of the EEG and other brain stem evoked response measurements may help to identify central apnea.

### **Sluder's Neuralgia**

The symptoms are neuralgia of the lower half of the face, nasal congestion, and rhinorrhea associated with lesions of the sphenopalatine ganglion. Ocular hyperemia and increased lacrimation may be seen.

### **Stevens-Johnson Syndrome**

This is a skin disease (erythema multiforme) with involvement of the oral cavity (stomatitis) and the eye (conjunctivitis). Stomatitis may appear as the first symptom. It is most common in the third decade of life. Treatment is largely with steroid and supportive therapy. This is a self-limiting disease, but has a 25% recurrence rate. The differential diagnosis would include (1) herpes simplex, (2) pemphigus, (3) acute fusospirochetal stomatitis, (4) chickenpox, (5) monilial infection, and (6) secondary syphilis.

### **Still's Disease**

Rheumatoid arthritis in children is sometimes called Still's disease (see pediatric textbook for more details).

### **Sturge-Weber-Syndrome**

This is a congenital disorder which affects both sexes equally, and is of unknown etiology. It is characterized by venous angioma of the leptomeninges over the cerebral cortex, ipsilateral port-wine nevi, and frequent angiomatous involvement of the globe, mouth, and nasal mucosa. The patient may have convulsions, hemiparesis, glaucoma, and intracranial calcifications. There is no specific treatment.

### **Subclavian Steal Syndrome**

Stenosis or occlusion of the subclavian or innominate artery proximal to the origin of the vertebral artery causes the pressure in the vertebral artery to be less than that of the basilar artery, particularly when the upper extremity is in action. Hence, the brain receives less blood and may be ischemic. The symptoms consist of intermittent vertigo, occipital headache, blurred vision, diplopia, dysarthria, and pain in the upper extremity. The diagnosis, made through the patient's medical history, can be confirmed by the difference in blood pressure in the two upper extremities, by a bruit over the supraclavicular fossa, and by angiography.

### **Superior Orbital Fissure Syndrome**

#### **Orbital Apex Syndrome; Optic Foramen Syndrome, Sphenoid Fissure Syndrome**

There is involvement of cranial nerves III, IV, VI, V1, the ophthalmic veins, and sympathetics of the cavernous sinus. This syndrome can be caused by sphenoid sinusitis or by any neoplasia in that region. Symptoms include paralysis of the upper lid, orbital pain, photophobia, and paralysis of the above nerves. The optic nerve may be damaged as well.

### **Superior Vena Cava Syndrome**

This is obstruction of the superior vena cava or its main tributaries by bronchogenic carcinoma, mediastinal neoplasm, or lymphoma. Rarely, the presence of a substernal goiter causes edema and engorgement of the vessels of the face, neck, and arms, as well as a nonproductive cough and dyspnea.

### **Tapia's Syndrome**

Unilateral paralysis of the larynx and tongue is coupled with atrophy of the tongue while the soft palate and cricothyroid muscle are intact. This syndrome is usually caused by a lesion at the point where XII and X nerves together with the internal carotid artery cross one another.

### **Tay-Sachs Disease**

An infantile form of amaurotic familial idiocy with strong familial tendencies, it is of questionably recessive inheritance. It is more commonly found among those of Semitic extraction. Histologically, the nerve cells are distorted and filled with a lipid material. The juvenile form of this is called Spielmeier-Vogt's disease in which the patient is normal until after 5-7 years of age. This juvenile form is seen in children of non-Semitic extraction as well.

### **Tietze's Syndrome**

A costal chondritis, chondropathia tuberosa, of unknown etiology, its symptoms include pain, tenderness, and swelling of one or more of the upper costal cartilages (usually of the second rib). Treatment is symptomatic.

### **Tolosa-Hunt Syndrome**

A cranial polyneuropathy usually presenting as recurrent unilateral painful ophthalmoplegia. Cranial nerves II, III, IV, V1 and VI may be involved. The etiology is unknown and there is a tendency for spontaneous resolution and for recurrence. An orbital venogram may show occlusion of the superior ophthalmic vein and at least partial obliteration of the cavernous sinus. The clinical course often responds well to systemic steroids.

### **Treacher-Collins Syndrome** (See Chap. 9)

### **Trotter's Syndrome (Sinus of Morgagni Syndrome)**

Neuralgia of the inferior maxillary nerve, conductive hearing loss secondary to eustachian tube blockage, preauricular edema caused by neoplastic invasion of the sinus of Morgagni, ipsilateral akinesia of the soft palate, and trismus are observed in this syndrome.

### **Tube-Feeding Syndrome** (See Chap. 37, 65)

### **Turner's Syndrome** (See Chap. 9)

### **Turpin's Syndrome**

Patients have congenital bronchiectasis, megaesophagus, tracheoesophageal fistula, vertebral deformities, rib malformations and heterotopic thoracic duct.

### **Vail's Syndrome**

This syndrome consists of unilateral, usually nocturnal, vidian neuralgia which may be associated with sinusitis.

### **Vernet's Syndrome**

See Jugular Foramen Syndrome.

### **Villaret's Syndrome**

This is the same as the jugular foramen syndrome except that Horner's syndrome is present here, suggesting more extensive involvement in the region of the jugular foramen, the retroparotid area, and the lateral pharyngeal space.

### **Vogt-Koyanagi-Harada Syndrome**

Spastic diplegia with athetosis and pseudobulbar paralysis associated with a lesion of the caudate nucleus and putamen, bilateral uveitis, vitiligo, deafness, alopecia, increased CSF pressure, and retinal detachment are evidenced.

### Wallenberg's Syndrome

Also called syndrome of the posterior inferior cerebellar artery thrombosis or lateral medullary syndrome, this syndrome is due to thrombosis of the posteroinferior cerebellar artery giving rise to ischemia of the brain stem (lateral medullary region). Symptoms include vertigo, nystagmus, nausea, vomiting, Horner's syndrome, dysphagia, dysphonia, hypotonia, asthenia, ataxia, falling to the side of the lesion and loss of pain and temperature sense on the ipsilateral face and contralateral side below the neck.

### Weber's Syndrome

This is characterized by paralysis of the oculomotor nerve on the side of the lesion and paralysis of the extremities, face, and tongue on the contralateral side. It indicates a lesion in the ventral and internal part of the cerebral peduncle.

### Wilson's Disease (Hepatolenticular Degeneration)

There are two chief types: one rapidly progressive which occurs in late childhood, the other slowly progressive occurring in the third or fourth decades. Familial, its symptoms are cirrhosis with progressive damage to the nervous system, brown pigmentation of the outer margins of the cornea called Kayser-Fleischer ring, and it can present with hearing loss as well.

### Winkler's Disease (Chondrodermatitis Nodularis Chronica Helicis)

Arteriovenous anastomosis and nerve endings accumulation at the helical portion of the ear are evident. It presents with pain and is characterized by hard, round nodules involving the skin and cartilage of the helix. Ninety percent occurs in males. The treatment is to excise it or treat it with steroids.

### Xeroderma Pigmentosum (Autosomal Recessive)

This presents with photosensitive skin with multiple basal cell epitheliomas. Squamous cell carcinoma or malignant melanoma can result from it. This condition occurs mainly in children. These children should be kept away from the sun.

### **Eponyms**

Adenoid Facies: Crowded teeth, high-arched palate, under-developed nostrils.

Adler Bodies: Deposits of mucopolysaccharide found in neutrophils of the patients with Hurler's syndrome.

Antoni's Type A and Type B: See Chap. 37.

Arnold-Chiari Malformation:

Type I: Downward protrusion of the long, thin, cerebellar tonsils through the foramen magnum.

Type II: Protrusion of the inferior cerebellar vermis through the foramen.

Type III: Bony occipital defect with descent of the entire cerebellum.

Type IV: Cerebellar hypoplasia.

Abrikossoff's Tumor (granular cell myoblastoma): Causes pseudoepithelial hyperplasia in the larynx, the site most favored in the larynx being the posterior half of the vocal cord. Three percent of granular cell myoblastoma progresses to malignancy. In order of decreasing frequency of involvement, the granular cell myoblastoma occurs in tongue, skin, breast, subcutaneous tissue, and respiratory tract.

Arnold's Ganglion: Otic ganglion.

Aschoff Body: Rheumatic nodule found in rheumatic disease.

Ballet's Sign: Paralysis of voluntary movement of the eyeball with preservation of the automatic movements. Sometimes this sign is present in exophthalmic goiter and hysteria.

Bechterew's Symptom: Paralysis of facial muscles limited to automatic movements. The power of voluntary movement is retained.

Bednar's Aphthae: Symmetrical excoriations of the hard palate in the region of the pterygoid plates, due to sucking of the thumb or foreign object or to scalding.

Bezold's Abscess: Abscess in the sternocleidomastoid muscle secondary to perforation of the tip of the mastoid by infection.

Blandin, Gland of: A minor salivary gland situated in the anterior portion of the tongue.

Brooke's Tumor (epithelioma adenoid cystica): This originates from the hair follicles in the external auditory canal and auricle. It is of basal cell origin. Treatment is through local resection.

Broyle's Ligament: Anterior commissure ligament of the larynx.

Brudzinski's Sign: In meningitis, a passive flexion of the leg on one side causes a similar movement to occur in the opposite leg. Passive flexion of the neck brings about a flexion of the legs as well.

"Brunner's" Abscess: Abscess of the posterior floor of the mouth.

Brun's Sign: Intermittent headache, vertigo, and vomiting, especially with sudden movements of the head. This occurs in cases of tumor of the fourth ventricle of the brain.

Bryce's Sign: A gurgling is heard in a neck mass. It suggests a laryngocele.

Carhart's Notch: Maximum dip at 2000 kHz (bone conduction) seen in patients with otosclerosis.

Charcot-Leyden Crystals: Crystals in the shape of elongated double pyramids, composed of spermine phosphates and present in the sputum of asthmatic patients. Synonyms are: Charcot-Newman crystals, Charcot-Robin crystals.

Charcot's Triad: The nystagmus, scanning speech, and intention tremor seen in multiple sclerosis.

Cherubism: Familial, with the age of predilection between 2-5 years old. It is characterized by giant cell reparative granuloma causing cystic lesions in the posterior rami of the mandible. The lesions are usually symmetric. It is a self-limiting disease with remissions after puberty. The maxilla also may be involved.

Chvostek's Sign: It is the facial twitch obtained by tapping the distribution of the facial nerve. It is indicative of hypocalcemia and is the most reliable test for hypocalcemia.

Curschmann's Spirale: Spirally twisted masses of mucus present in the sputum of bronchial asthmatic patients.

Demarquay's Sign: Absence of elevation of the larynx during deglutition. This is said to indicate syphilitic induration of the trachea.

di Sant' Agnese Test: It measures the elevated sodium and chloride in the sweat of cystic fibrotic children.

Dupre's Sign: Meningism.

Ebner, Gustatory Glands of: These are the minor salivary glands near the circumvallate papillae.

Escherich's Sign: In hypoparathyroidism, tapping of the skin at the angle of the mouth causes protrusion of the lips.

Galen's Anastomosis: An anastomosis between the superior laryngeal nerve and the recurrent laryngeal nerve.

Goodwin's Tumor: Benign lymphoepithelioma. (See Chap. 17)

Griesinger's Sign: Edema of the tip of the mastoid in thrombosis of the sigmoid sinus.

Guttman's Test: In the normal subject, frontal pressure on the thyroid cartilage lowers the tone of voice produced while lateral pressure produces a higher tone of voice. The opposite is true in paralysis of the cricothyroid muscle.

Gyon's Sign: The twelfth nerve lies directly upon the external carotid artery, whereby this vessel may be distinguished from the internal carotid artery. The safer way prior to ligation of the external carotid artery is to identify the first few branches of the external carotid artery.

Henle, Gland of: These are the small glands situated in the areolar tissue between the buccopharyngeal fascia anteriorly and the prevertebral fascia posteriorly. Infection of these glands can lead to retropharyngeal abscess. Since these glands atrophy after the age of 5, retropharyngeal abscess is less likely to occur after that age.

Hennebert's Sign: (See Chap. 9, Congenital Deafness) The presence of a positive fistula test in the absence of an obvious fistula is called Hennebert's sign. The patient has a normal appearing tympanic membrane and external auditory canal. The nystagmus is more marked upon application of a negative pressure. This sign is present in congenital syphilis and is believed to be due to an excessively mobile footplate or caused by motion of the sacculle mediated by fibrosis between the footplate and the sacculle.

Hering-Breuer Reflex: Respiratory reflexes from pulmonary stretch receptors. Inflation of the lungs sends an inhibitory impulse to the central nervous system via the vagus nerve to stop inspiration. Similarly, a deflation of the lungs sends an impulse to stop expiration.

Kernig's Sign: When the subject lies on his back, with the thigh at a right angle to the trunk, straightening of the leg (extending the leg) will elicit pain, supposedly due to the pull on the inflamed lumbosacral nerve roots. This sign is present in meningitis.

Kiesselbach's Plexus: This is an area in the anterior septum where the capillaries merge. It is often the site of anterior epistaxis. It also has been referred to as the Little's region.

Koplik's Spots: Pale round spots on the oral mucosa, conjunctiva, and lacrimal caruncle, seen in the beginning stages of measles.

Krause's Nodes: These are the nodes in the jugular foramen.

Lhermitte's Sign: Patients develop sensory deficits and complain of "electric shocks" radiating down the back and into the extremities after neck flexion. This occurs 2-4 months after completion of radiation therapy to the CNS or spinal canal and is classified as an early transient myelopathy.

Lillie-Crowe Test: Used in the diagnosis of unilateral sinus thrombophlebitis. Digital compression of the opposite internal jugular vein causes the retinal vessels to dilate.

Little's Area: See Kiesselbach's Plexus.

Ludwig's Angina: See Chap. 16.

Luschka's Pouch: See Thornwaldt's Disease.

Marcus Gunn's Phenomenon: Unilateral ptosis of the eyelid with exaggerated opening of the eye during movements of the mandible.

Marjolin's Ulcer: This is a carcinoma that arises at the site of an old burn scar. It is a well differentiated squamous cell carcinoma, aggressive, and metastasizes rapidly.

Meckel's Ganglion: Sphenopalatine ganglion.

Mikulicz's Cells: These are macrophages in rhinoscleroma. Russel bodies which are eosinophilic, round structures associated with plasma cells also are found in rhinoscleroma.

Mollaret-Debre Test: This is a test performed for cat-scratch fever.

Morgagni, Sinus of: A dehiscence of the superior constrictor muscle and the buccopharyngeal fascia where the eustachian tube opens.

Morgagni, Ventricle of: This separates the quadrangular membrane from the conus elasticus in the larynx.

Nikolsky's Sign: Detachment of the sheets of the superficial epithelial layers when any traction is applied over the surface of the epithelial involvement in pemphigus is characteristic of Nikolsky's sign. Pemphigus involves the intraepithelial layer while pemphigoid involves the subepithelial layer. The former is a lethal disease in many instances.

Oliver-Cardarelli Sign: Recession of the larynx and trachea is synchronous with cardiac systole in cases of aneurysm of the arch of the aorta, or in cases of a tumor in that region.

Parinaud's Sign: Extraocular muscle impairment with decreased upward gaze and ptosis seen in association with pinealomas and other lesions of the tectum.

Paul-Bunnell Test: It measures the elevated heterophile titer in infectious mononucleosis.

Psammoma Bodies: These are found in papillary carcinoma of the thyroid.

Rathke's Pouch: See Thorwaldt's Disease.

Reinke's Tumor: This is a "soft" tumor variant of lymphoepithelioma in which the lymphocytes predominate. In the hard tumor the epithelial cells predominate and this is called Schmincke's tumor.

Rhomberg's Sign: If a patient standing with feet together, "falls" when he closes his eyes, the Romberg's test is positive. It is indicative of either abnormal proprioception or abnormal vestibular function. It does not necessarily distinguish central from peripheral lesion. The cerebellar function is not tested in this test.



Rosenbach's Sign: Fine tremor of the closed eyelids seen in hyperthyroidism and hysteria.

Rouvier's Node: Lateral retropharyngeal node. It is a common target of metastases in nasopharyngeal carcinoma.

Russell's Bodies: Eosinophilic, round structures, associated with plasma cells found in rhinoscleroma.

Santorini's Cartilage: Corniculate cartilage of the larynx, composed of fibroelastic cartilage.

Santorini's Fissures: Fissures in the anterior bony external auditory canal leading to the parotid region.

Schaumann's Bodies: Together with asteroid, they are found in sarcoid granuloma.

Schmincke's Tumor: The "hard" variant of lymphoepithelioma in which the epithelial cells predominate (see Reinke's Tumor).

Schneiderian Mucosa: Pseudostratified ciliated columnar mucosa of the nose.

Seeligmüller's Sign: Contraction of pupil on the affected side in facial neuralgia.

Semon's Law: A law stating that injury to the recurrent laryngeal nerve results in paralysis of the abductor muscle of the larynx (cricothyroid muscle) before paralysis of the adductor muscles. In recovery, the adductor recovers before the abductor.

Straus' Sign: In facial paralysis, the lesion is peripheral if injection of pilocarpine is followed by sweating on the affected side later than on the normal side.

Sulkowitch's Test: It determines an increase in calciuria.

Tobey-Ayer-Queckenstedt Test: Used in the diagnosis of unilateral and bilateral sinus thrombophlebitis. In cases where the lateral sinus is obstructed on one side, compression of the jugular vein on the intact side will cause a rise in CSF pressure, whereas compression of the obstructed side does not raise the CSF pressure.

Thornwaldt's Cyst: A depression exists in the nasopharyngeal vault which is a remnant of the pouch of Luschka. When this depression becomes infected a Thornwaldt's cyst results. In the early embryo, this area has a connection between the notochord and ectoderm. The Thornwaldt's cyst is lined with respiratory epithelium with some squamous metaplasia. Anterior to this pit, the path taken by Rathke's pouch sometimes persists as the craniopharyngeal canal, running from the sella turcica through the body of the sphenoid to an opening on the undersurface of the skull.

Toynbee's Law: When CNS complications arise in chronic otitis media, the lateral sinus and cerebellum are involved in mastoiditis while the cerebrum alone is involved in the instances of cholesteatoma of the attic.

Trousseau's Sign: In hypocalcemia, a tourniquet placed around the arm will cause tetany.

Tullio's Phenomenon: (See Chap. 9, Congenital Deafness) This is said to be present when a loud noise precipitates vertigo. It can be present in congenital syphilis with a semicircular canal fistula or in a postfenestration patient if the footplate is mobile. The tympanic membrane and ossicular chain have to be intact with mobile footplate.

Wartenberg's Sign: Intense pruritus of the tip of the nose and nostril indicates cerebral tumor.

Warthin-Finkelday Giant Cells: These are found in the lymphoid tissue in measles.

Weber's Gland: These are minor salivary glands in the superior pole of the tonsil.

Wrisberg's Cartilage: This is the cuneiform cartilage of the larynx, made of fibroelastic cartilage.

Xeroderma Pigmentosa: Hereditary precancerous condition which begins in early childhood. These patients die at puberty.

Zaufal's Sign: Saddle nose.