K. J. Lee: Essential Otolaryngology and Head and Neck Surgery (IIIrd Ed)

Chapter 9: Congenital Deafness

General Information

1. One person in eight carries a recessive gene for deafness; 1:4000 live births has hereditary deafness.

2. One percent of hereditary hearing loss is sex-linked; 9% are due to an autosomal-dominant inheritance; and 90% are the result of an autosomal-recessive transmission. Hereditary deafness consitutes 15% of all congenital deagness.

3. Dominant hearing loss usually progresses while the recessive type is nonprogressive.

4. Hereditary deafness can be classified as follows:

a. Hereditary (congenital) deafness without associated abnormalities (autosomal-dominant, autosomal-recessive, or sex-linked).

b. Hereditary congenital deafness associated with integumentary system disease (autosomal-dominant, autosomal recessive, or sex-linked).

c. Hereditary congenital deafness associated with skeletal disease (autosomal-dominant, autosomal recessive, or sex-linked).

d. Hereditary congenital deafness associated with other abnormalities (autosomal-dominant, autosomal recessive, or sex-linked).

Each category can be subdivided along three kinds of hearing impairment: sensorineural, conductive, and mixed.

5. Otologists have attempted to classify inner ear developmental anomalies. One of the classifications is:

a. Michel: Complete failure of developmental of the inner ear (bony and membranous aplasia). The middle ear and external auditory canal may be normal.

b. Mondini-Alexander: Incomplete development of the bony and membranous labyrinth. The cochlea may be represented by a single curved tube, and the vestibular labyrinth is not developed.

c. Scheibe: Membranous cochlea-saccular aplasia (pars inferior). The bony labyrinth is normal. The utriculus and semicircular canals (pars superior) are normal.

d. Alexander: Partial aplasia of the cochlear duct giving rise to high-frequency hearing loss.

e. Bing-Siebenmann: The membranous vestibular apparatus is maldeveloped. The membranous cochlea may or may not be normal.

6. The Scheibe's type of inner ear anomaly is the most commonly encountered. It is believed to be transmitted autosomal recessively. The next most common is the Mondini-Alexander type which is believed to be autosomal dominant.

7. The rubella syndrome includes congenital cataract, cardiovascular anomalies, mental retardation, retinitis, and deafness. It has been reported that 5-10% of the mothers with rubella in the first trimester gave birth to children with deafness. Alford reported 90/141 rubella syndrome children presented with deafness. The eye is the most commonly affected organ followed by the ears and then by the heart. Histologically, the middle ear as well as inner ear anomalies have been described. Confirmatory tests for the rubella syndrome include identification of fluorescent antibody, serum hemagglutination, and viral cultures from stool and throat. Deafness of a viral etiology shows: (a) degeneration of the organ of Corti; (b) adhesions between the organ of Corti and Reissner's membrane; (c) rolled up tectorial membrane; (d) partial or complete stria atrophy; and (e) scattered degeneration of neural elements (cochlea-saccule degeneration).

8. Twenty percent of the *kernicterus* babies will have severe deafness secondary to damage to the dorsal and ventral cochlear nuclei as well as the superior and inferior colliculi nuclei. Clinically, bilateral sensorineural loss especially in high frequencies is manifested. The most accepted indication for exchange transfusion is a serum bilirubin of greater than 20 mg/100 mL.

9. Syphilitic deafness: Tamari and Itkin estimated that hearing loss occured in:

17% of early congenital syphilis
18% of late congenital syphilis
25% of late latent syphilis
29% of asymptomatic neurosyphilis
80% of symptomatic neurosyphilis.

Congenital

Contrary to the above, Karmody and Schuknecht reported 25-38% of patients with congenital syphilis had hearing loss. There exist two forms of congenital syphilis: early (infantile) and late (tardive). The infantile form is often severe, bilateral. These children usually have multisystem involvement and hence a fatal outcome.

Late congenital syphilis has progressive hearing loss of varying severity and time of onset. Hearing losses that have the onset in early childhood are usually bilateral, sudden, severe, and associated with vestibular symptoms. The symptom complex is similar to Ménière's disease. The late onset form (sometimes as late as the fifth decade of life) has mild hearing loss. Karmody and Schuknecht also pointed out that the vestibular disorders of severe episodic vertigo are more common in the late-onset group than in the infantile group. Histopathologically, osteitis with mononuclear leukocytosis, obliterative endoarteritis, and endolymphatic hydrops is noticed. Serum and CSF serology may or may not be positive.

Treatment with steroids and penicillin seems to be of benefit. Other sites of congenital syphilis are:

a. Nasal cartilaginous and bony framework.

b. Periostitis of the cranial bones (bossing).

c. Periostitis of the tibia (saber shin).

d. Injury to the odontogenous tissues (Hutchinson's teeth).

e. Injury to the epiphyseal cartilages (short stature).

f. Commonly interstitial keratitis (cloudy cornea).

Two signs are associated with congenital syphilis:

a. Hennebert's sign consist of a positive fistula test without clinical evidence of middle ear or mastoid disease or a fistula. Nadol postulated that the vestibular stimulation is mediated by fibrous bands between the footplate and the vestibular membranous labyrinth. He observed that Hennebert's sign also may be present in Ménière's disease. The other explanatin was that the vestibular response is due to an excessive mobile footplate. The nystagmus in Hennebert's sign usually is more marked upon application of a negative pressure (see under Hennevert's Sign, Chap. 23: Syndromes and Eponyms).

b. Tullio's phenomenon consists of vertigo and nystagmus on stimulation with high-intensity sound such as the Barany noise box. This phenomenon occurs not only in congenital syphilis patients with a semicircular canal fistula but also in postfenestration patients if the footplate is mobile and the fenestrum patent. It also can be demonstrated in chronic otitis media should the patient have an intact tympanic membrane, ossicular chain, and a fistula - a rare combination.

For Tullio's phenomenon to take place, a fistula of the semicircular canal and intact sound transmission mechanism to the inner ear (i.e. intact tympanic membrane, intact ossicular chain, mobile footplate) must be present. The pathophysiology is that the high-intensity noise energy transmitted through the footoplate finds the course of least resistance and displaces toward the fistula instead of toward the round window membrane.

Acquired

Hearing loss may occur in the secondary or tertiary forms of acquired syphilis. Histopathologically, osteitis with round cell infiltration is noticed. In tertiary syphilis, gummatous lesions may involve the auricle, mastoid, middle ear, and petrous pyramid. These lesions can cause a mixed hearing loss. Since penicillin and other antibiotic therapy is quite effective in treating acquired syphilis, this form of deafness is rare nowadays.

10. Cretinism: Retarded growth, mental retardation and mixed hearing loss, are seen in this condition.

I. Hereditary Deafness Without Associated Abnormalities

A. Stria Atrophy (hereditary, not congenital).

1. Autosomal dominant.

2. The sensorineural hearing loss begins at middle-age and is progressive.

3. Good discrimination is maintained.

4. Flat audiometric curve.

5. Positive SISI test.

6. Bilaterally symmetrically hearing loss.

7. The patient never becomes profoundly deaf.

B. Otosclerosis (hereditary, not congenital).

1. See Chap. 8.

II. Hereditary Congenital Deafness Associated With Integumentary System Disease

A. Albinism with Blue Irides.

1. Autosomal dominant or recessive.

2. Sensorineural hearing loss.

B. Ectodermal Dysplasia, Hidrotic (Anhidrotic ectodermal dysplasia is sex-linked recessive. Mixed or conductive hearing loss).

1. Autosomal dominant.

2. Small dystrophic nails.

3. Coniform teeth.

4. Elevated sweat electrolytes.

5. Sensorineural hearing loss.

C. Forney's Syndrome.

1. Autosomal dominant.

2. Lentigines.

3. Mitral insufficiency.

4. Skeletal malformations.

5. Conductive hearing loss.

D. Lentigines.

1. Autosomal dominant.

2. Brown's spots appear on the skin. These begin to appear at age 2.

3. Ocular hypertelorism.

4. Pulmonary stenosis.

5. Abnormalities of the genitalia.

6. Retarded growth.

7. Sensorineural hearing loss.

E. Leopard Syndrome.

1. Autosomal dominant with variable penetrance.

2. Variable sensorineural hearing loss.

3. Ocular hypertelorism.

4. Pulmonary stenosis.

5. Hypogonadism.

6. ECG changes with widened QRS or bundle branch block.

7. Retardation of growth.

8. Normal vestibular apparatus.

9. Lenitigines.

10. Skin changes progressive over the first and second decades.

F. Piebaldness.

1. Sex-linked or autosomal recessive.

2. Blue irides.

3. Fine retinal pigmentation.

4. Depigmentation of scalp, hair, and face.

5. Areas of depigmentation on limbs and trunk.

6. Sensorineural hearing loss.

G. Tietz's Syndrome.

1. Autosomal dominant.

2. Profound deafness.

3. Albinism.

4. Eyebrows absent.

5. Blue irides.

6. No photophobia or nystagmus.

H. Waardenburg's Disease.

1. Autosomal dominant with variable penetrance.

2. Contributes 1-7% of all hereditary deafness.

3. Widely spaced medial canthi. This is present in all cases.

4. Flat nasal root in 75% of the cases.

5. Confluent eyebrow.

6. Sensorineural hearing loss (unilateral or bilateral). Hearing loss is present in 20% of cases.

7. Colored irides.

8. White forelock.

9. 10% of these patients have areas of depigmentation.

10. Abnormal tyrosine metabolism.

11. 75% of these patients have diminished vestibular function.

12. 10% of these patients are associated with cleft lip and palate.

III. Hereditary Congenital Deafness Associated With Skeletal Disease

A. Achondroplasia.

1. Autosomal dominant.

2. Large head, short extremities.

3. Dwarfism.

4. Mixed hearing loss (fused ossicles).

5. Saddle nose, frontal and mandibular prominence.

B. Apert's Disease (Acrocephalosyndactyly).

1. Autosomal dominant.

2. Syndactylia.

3. Flat conductive hearing loss secondary to stapes fixation.

4. Patent cochlear aqueduct has been noted histologically.

5. Frontal prominence, exophatlmos.

6. Craniofacial dysostosis, hypoplastic maxilla.

7. Proptosis, saddle nose, high arched palate, and occasionally, with spina bifida.

8. Occurs in about 1:150.000 live births.

C. Atresia Auris Congenita.

1. Autosomal dominant.

2. Unilateral or bilateral involvement are possible.

3. Middle ear abnormalities with VII nerve anomaly.

4. Internal hydrocephalus.

5. Mental retardation.

6. Epilepsy.

6. Choanal atresia and cleft palate.

D. Cleidocranial Dysostosis.

1. Autosomal dominant.

2. Absent or hypoplastic clavicle.

3. Failure of fontanelles to close.

4. Sensorineural hearing loss.

E. Crouzon's Disease (Craniofacial Dysostosis).

1. Autosomal dominant.

2. One-third of the cases are associated with hearing loss.

3. Mixed hearing loss in some cases.

4. Cranial synostosis.

5. Exophthalmos and divergent squint.

6. Parrot-beaked nose.

7. Short upper lip.

8. Mandibular prognathism and small maxilla.

9. Hypertelorism.

10. The external auditory canal may be atretic.

11. Congenital enlargement of the sphenoid bone.

12. Premature closure of the cranial suture lines can lead to mental retardation.

F. Engelmann's Syndrome (Diaphyseal Dysplasia).

1. Autosomal dominant; ? recessive.

2. Progressive mixed hearing loss.

3. Progressive cortical thickening of diaphyseal regions of long bones and skull.

G. Hand-Hearing Syndrome.

1. Autosomal dominant.

2. Congenital flexion contractures of fingers and toes.

3. Sensorineural hearing loss.

H. Klippel-Feil (Brevicollis; Wilderevanck's) Syndrome.

1. Autosomal recessive or dominant.

2. The incidence in females if greater than in males.

3. Sensorineural hearing loss along with middle ear anomalies.

4. Short neck due to fused cervical vertebrae.

5. Spina bifida.

6. External auditory canal atresia.

I. Madelung's Deformity (Related to Dyschondrosteosis of Leri-Weill).

1. Autosomal dominant.

2. Short stature.

3. Ulna and elbow dislocation.

4. Conductive hearing loss secondary to ossicular malformation with normal tympanic membrane and external auditory canal.

5. Spina bifida occulta.

6. The ratio of female to male is 4:1.

J. Marfan's Syndrome (Arachnodactyly, Ectopia Lentis, Deafness).

1. Autosomal dominant.

2. Thin elongated individuals with long spidery fingers.

3. Scoliosis.

4. Hammer toes.

5. Mixed hearing loss.

K. Mohr's Syndrome (Oral-Facial-Digital Syndrome II).

1. Autosomal recessive.

2. Conductive hearing loss.

3. Cleft lip, high-arched palate.

4. Lobulated nodular tongue.

5. Broad nasal root, bifid tip of nose.

6. Hypoplasia of the body of the mandible.

7. Polydactyly and syndactyly.

L. Osteopetrosis (Albers-Schönberg Disease; Marble Bone Disease).

1. Autosomal recessive (A rare dominant transmission has been reported).

2. Conductive or mixed hearing loss.

3. Fluctuating facial nerve paralysis.

4. Sclerotic, brittle bone due to failure of resorption of calcified cartilage.

5. Cranial nerves II, V, VII also may be involved.

6. Optic atrophy.

7. Atresia of paranasal sinuses.

8. Choanal atresia.

9. Increased incidence of osteomyelitis.

10. The widespread form of this disease may lead to obliteration of the bone marrow, severe anemia, and rapid demise.

11. May have hepatosplenomegaly.

M. Oto-Facial-Cervical Syndrome.

1. Ausomal dominant.

2. Depressed nasal root.

3. Protruding narrow nose.

4. Narrow elongated face.

5. Flattened maxilla and zygoma.

6. Prominent ears.

7. Preauricular fistulas.

8. Poorly developed neck muscles.

9. Conductive hearing loss.

N. Oto-Palatal-Digital Syndrome.

1. Autosomal recessive.

2. Conductive hearing loss.

3. Mild dwarfism.

4. Cleft palate.

5. Mental retardation.

6. Broad nasal root, hypertelorism.

7. Frontal and occipital bossing.

8. Small mandible.

9. Stubby, clubbed digits.

10. Low-set small ears.

11. Winged scapulae.

12. Malar flattening.

13. Downward obliquity of eye.

14. Down-turned mouth.

O. Paget's Disease (Osteitis Deformans).

1. Autosomal dominant with variable penetrance.

2. Mainly sensorineural hearing loss but mixed hearing loss is seen as well.

3. Occasionally may develop cranial nerve involvement.

4. Onset usually at middle-age, involving skull and long bones of the legs.

5. The endochondral bone is somewhat resistant to this disease.

P. Pierre Robin Syndrome (Cleft Palate, Micrognathia, and Glossoptosis).

1. Autosomal dominant with variable penetrance; possibly not hereditary but due to intrauterine insult.

2. It occurs in 1:30.000 to 1:50.000 live births.

3. Glossoptosis.

4. Micrognathia.

5. Cleft palate (in 50% of the cases).

6. Mixed hearing loss.

7. Malformed auricles.

8. Mental retardation.

9. Hypoplastic mandible.

10. Möbius' syndrome.

11. Subglottic stenosis not uncommon.

12. Aspiration a common cause of death.

Q. Pyle's Disease (Craniometaphyseal Dysplasia).

1. Autosomal dominant (less often autosomal recessive).

2. Conductive hearing loss can begin at any age. It is progressive, and it is secondary to fixation of the stapes or other ossicular abnormalities. Mixed hearing loss is also possible.

3. Cranial nerve palsy secondary to narrowing of the foramen.

4. Splayed appearance of long bones.

5. Choanal atresia.

6. Prognathism.

7. Optic atrophy.

8. Obstruction of sinuses and nasolacrimal duct.

R. Roaf's Syndrome.

1. Not hereditary.

2. Retinal detachment, cataracts, myopia, coxavara, kyphoscoliosis, and retardation.

3. Progressive sensorineural hearing loss.

S. Dominant Proximal Symphalangia and Hearing Loss.

1. Autosomal dominant.

2. Ankylosis of proximal interphalangeal joint.

3. Conductive hearing loss early in life.

T. Treacher Collins Disease (Mandibulofacial Dysostosis; Franceschetti-Zwahlen-Klein Syndrome).

1. Autosomal dominant or intrauterine abuse.

2. Antimongoloid palpebral fissures with notched lower lids.

3. Malformation of ossicles (stapes is usually normal).

4. Auricular deformity, atresia of external auditory canal.

5. Conductive hearing loss.

6. Preauricular fistulas.

7. Mandibular hypoplasia and malar hypoplasia.

8. "Fish-mouth".

9. Normal IQ.

10. Usually bilateral involvement.

11. May have cleft palate and cleft lip.

12. Arrest in embryonic development occurs at 6-8 weeks to give the above findings.

U. Van Buchem's Disease (Hyperostosis Corticalis Generalisata).

1. Autosomal recessive.

2. Generalized osteosclerotic overgrowth of skeleton including skull, mandible, ribs, long and short bones.

3. Cranial nerve palsies due to obstruction of the foramina.

4. Increased serum alkaline phosphatase.

5. Progressive sensorineural hearing loss.

V. Van der Hoeve's Syndrome (Osteogenesis Imperfecta).

1. Autosomal dominant with variable expressivity.

2. Fragile bones, loose ligaments.

3. Blue or clear sclera, triangular facies, dentinogenesis imperfecta.

4. 60% of osteogenesis imperfecta patients have blue sclera and hearing loss which are most frequently noticed after age 20. The hearing loss is conductive and is due to stapes fixation by otosclerosis. Hearing loss also can be due to ossicular fracture. Some use the term Van der Hoeve's syndrome to describe osteogenesis imperfecta with otosclerosis. Others use the term interchangeably with osteogenesis imperfecta regardless of whether otosclerosis is present or not.

5. The basic pathologic defect is "abnormal osteoblastic activity".

6. When operating on such a patient, it is important to avoid fracture of the tympanic ring or the long process of the incus. It is also important to realize that the stapes footplate may be "floating".

7. The sclera may have increased mucopolysaccharide content.

8. These patients have normal calcium, phosphorus, and alkaline phosphatase in the serum.

9. Occasionally capillary fragility is noticed in these patients.

IV. Hereditary Congenital Deafness Associated With Other Abnormalities

A. Acoustic Neurinomas (Inherited).

1. Autosomal dominant.

2. Progressive sensorineural hearing loss in the second or third decades of life.

3. Ataxia, visual loss.

4. No cafe au lait spots.

B. Alport's Disease.

1. Autosomal dominant.

2. Progressive nephritis and sensorineural hearing loss.

3. Hematuria, proteinuria beginning the first or second decade of life.

4. Males with this disease usually die of uremia by the age of 30. Females are less severely affected.

5. Kidneys are affected by chronic glomerulonephritis with interstitial lymphocytic infiltrate and foam cells.

6. Progressive sensorineural hearing loss begins at age 10. Although it is considered not sex-linked, hearing loss affects almost all males but not all females. Histologically, degeneration of the organ of Corti and stria vascularis is observed.

7. Spherophalera cataract.

8. Hypofunction of the vestibular organ.

9. Contributes 1% of hereditary deafness.

C. Alstrom's Disease.

1. Autosomal recessive.

2. Retinal degeneration giving rise to visual loss.

3. Diabetes, obesity.

4. Progressive sensorineural hearing loss.

D. Cockayne's Syndrome.

1. Autosomal recessive.

2. Dwarfism.

3. Mental retardation.

4. Retinal atrophy.

5. Motor disturbances.

6. Progressive hearing loss bilaterally.

E. Congenital Cretinism (To be distinguished from Pendred's Syndrome).

1. 35% of the patients with congenital cretinism present with congenital hearing loss of the mixed type (irreversible).

2. Goiter, hypothyroid.

3. Mental and physical retardation.

4. Abnormal development of the petrous pyramid.

5. This disease is not inherited in a specific Mendelian manner. It is restricted to a certain geographical locale where a dietary deficiency exists.

F. Duane's Syndrome.

1. Autosomal dominant (some sex-linked recessive).

2. Inability to abduct eyes, retract globe.

3. Narrowing of palpebral fissure.

4. Torticolis.

5. Cervical rib.

6. Conductive hearing loss.

G. Fanconi's Anemia Syndrome.

1. Autosomal recessive.

2. Absent or deformed thumb.

3. Other skeletal, heart, and kidney malformations.

4. Increased skin pigmentation.

5. Mental retardation.

6. Pancytopenia.

7. Conductive hearing loss.

H. Fehr's Corneal Dystrophy.

1. Autosomal recessive.

2. Progressive visual and sensorineural hearing loss.

I. Flynn-Aird Syndrome.

1. Autosomal dominant.

2. Progressive myopia, cataracts, retinitis pigmentosa.

3. Progressive sensorineural hearing loss.

4. Ataxia.

5. Shooting pains in the joints.

J. Friedreich's Ataxia.

1. Autosomal recessive.

2. Childhood onset of nystagmus, ataxia, optic atrophy, hyperreflexia, and sensorineural hearing loss.

K. Goldenhar's Syndrome.

1. Autosomal recessive.

2. Epibulbar dermoids.

3. Preauricular appendages.

4. Fusion or absence of cervical vertebrae.

5. Colobomas of the eye.

6. Conductive hearing loss.

L. Hallgren's Syndrome.

1. Autosomal recessive.

2. Retinitis pigmentosa.

3. Progressive ataxia.

4. Mental retardation occurs in 25% of these patients.

5. Sensorineural hearing loss.

6. Constitutes about 5% of hereditary deafness.

M. Hermann's Syndrome.

1. Autosomal dominant.

2. Onset of photomyoclonus and sensorineural hearing loss in late of childhood or adolescence.

3. Diabetes mellitus.

4. Progressive dementia.

5. Pyelonephritis and glomerulonephritis.

N. Hurler's Syndrome (Gargoylism).

1. Autosomal recessive.

2. Abnormal mucopolysaccharide are deposited in tissues (when mucopolysaccharide is deposited in the neutrophiles they are called Adler bodies); middle ear mucosa with large foamy gargoyle cells staining PAS positive.

3. Chondroitin sulfate B and heparitin are found in urine.

4. Forehead prominence with coarsening of the facial features and low-set ears.

5. Mental retardation.

6. Progressive corneal opacities.

7. Hepatosplenomegaly.

8. Mixed hearing loss.

9. Dwarfism.

10. Cerebral storage of three gangliosides, GM3, 2, 1.

11. Beta-galactosides deficient.

O. Hunter's Syndrome.

1. Same as above except that it is sex-linked.

P. Jervell-Lange-Nielson Syndrome.

1. Autosomal recessive.

2. Profound bilateral sensorineural hearing loss; high frequencies more severely

impaired.

3. Associated with heart disease (prolonged QT interval on ECG); has been associated with Stokes-Adams disease.

4. Recurrent syncope.

5. Usually terminates fatally; death is sudden.

6. Histopathologically, PAS-positive nodules can be seen in the cochlea.

Q. Laurence-Moon-Biedl-Bardet Syndrome.

1. Autosomal recessive.

2. Dwarfism.

3. Obesity.

4. Hypogonadism.

5. Retinitis pigmentosa.

6. Mental retardation.

7. Sensorineural hearing loss.

R. (Recessive) Malformed Low-Set Ears and Conductive Hearing Loss.

1. Autosomal recessive.

2. 50% show mental retardation.

S. (Dominant) Mitral Insufficiency, Joint Fusion and Hearing Loss.

1. Autosomal dominant with variable penetrance.

2. Conductive hearing loss, usually due to fixation of the stapes.

3. Narrow external auditory canal.

4. Fusion of the cervical vertebrae, carpal, and tarsal bones.

T. Möbius' Syndrome (Congenital Facial Diplegia).

1. Autosomal dominant, ? recessive.

2. Facial diplegia.

3. External ear deformities.

4. Ophthalmoplegia.

5. Hands or feet may be missing.

6. Mental retardation.

7. Paralysis of the tongue.

8. Mixed hearing loss.

U. (Dominant) Saddle Nose, Myopia, Cataract, and Hearing Loss.

1. Autosomal dominant.

2. Saddle nose.

3. Severe myopia.

4. Juvenile cataract.

5. Sensorineural hearing loss which is progressive, moderately severe, and of an early onset.

V. Norrie's Syndrome.

1. Autosomal recessive.

2. Congenital blindness due to pseudotumor retinae.

3. Progressive sensorineural hearing loss in 30% of patients.

W. Pendred's Disease.

1. Autosomal recessive.

2. Variable amount of bilateral hearing loss secondary to atrophy of the organ of Corti. A U-shaped audiogram is often seen.

3. These patients are euthyroid. They develop diffuse goiter at the time of puberty. It is said that the metabolic defect is faulty iodination of tyrosine.

4. Positive perchlorate test.

5. The goiter is treated with exogenous hormone to suppress TSH secretion.

6. Normal IQ.

7. Unlike congenital cretinism, the bony petrous pyramid is well developed.

8. Constitutes 10% of hereditary deafness.

X. Refsum's disease (Heredopathia Atactica Polyneuritiformis).

1. Autosomal recessive.

2. Retinitis pigmentosa.

3. Polyneuropathy,.

4. Ataxia.

5. Sensorineural hearing loss.

6. Visual impairment usually begins in the second decade.

7. Ichthyosis is often present.

8. Elevated plasma phytanic acid levels.

9. Etiology: Neuronal lipid storage disease and hypertrophic polyneuropathy.

Y. (Recessive) Renal, Genital, Middle Ear Anomalies.

1. Autosomal recessive.

2. Renal hypoplasia.

3. Internal genital malformation.

4. Middle ear malformation.

5. Moderate to severe conductive hearing loss.

Z. Richards-Rundel Disease.

1. Autosomal recessive.

2. Mental deficiency.

3. Hypogonadism (decreased urinary estrogen, pregnanediol, and total 17-ketosteroids).

4. Ataxia.

5. Horizontal nystagmus to bilateral gazes.

6. Sensorineural hearing loss begins at infancy.

7. Muscle wasting in early childhood and absent deep tendon reflexes.

AA. Taylor's Syndrome.

1. Autosomal recessive.

2. Unilateral microtia or anotia.

3. Unilateral facial bone hypoplasia.

4. Conductive hearing loss.

BB. Trisomy 13-15 (Group D) (Patau's Syndrome).

1. Low-set pinnae.

2. Atresia of external auditory canals.

3. Cleft lip and cleft palate.

4. Colobomas of the eyelids.

5. Micrognathia.

6. Tracheoesophageal fistula.

7. Hemangiomas.

8. Congenital heart disease.

9. Mental retardation.

10. Mixed hearing loss.

11. Hypertelorism.

12. Incidence 0.45:1.000 live births.

13. Patients usually die early in childhood.

CC. Trisomy 16, 17, 18 (Group E).

1. Low-set pinnae.

2. External canal atresia.

3. Micrognathia, high-arched palate.

4. Peculiar finger position.

5. Prominent occiput.

6. Cardiac anomalies.

7. Hernias.

8. Pigeon breast.

9. Mixed hearing loss.

10. Incidence 0.25-2:1.000 live births.

11. Ptosis.

12. Patients usually die early in life.

DD. Trisomy 21 or 22 (Down's Syndrome) (G Trisomy).

1. Extra chromosome on no. 21 or no. 22.

2. Mental retardation.

3. Short stature.

4. Brachycephaly.

5. Flat occiput.

6. Slanted eyes.

7. Epicanthus.

8. Strabismus, nystagmus.

9. Seen in association with leukemia.

10. Subglottic stenosis not uncommon.

11. Decreased pneumatized or absent frontal and sphenoid sinuses.

12. 1:600 live births.

EE. Turner's Syndrome.

1. Not inherited; ? due to intrauterine insult.

2. Low hairline.

3. Webbing of neck and digits.

3. Widely spaced nipples.

4. XO; 80% are sex chromatin negative.

6. Gonadal aplasia.

7. Incidence 1:5.000 live births (Klinefelter's syndrome is XXY).

8. Ossicular deformities.

9. Low-set ears.

10. Mixed hearing loss.

11. Large ear lobes.

12. Short stature.

13. Abnormalities found in the heart and kidney.

14. Some with hyposomia.

FF. (Dominant) Urticaria, Amyloidosis, Nephritis, and Hearing Loss.

1. Autosomal dominant.

2. Recurrent urticaria.

3. Amyloidosis.

4. Progressive sensorineural hearing loss due to degeneration of the organ of Corti; ossification of the basilar membrane, and cochlear nerve degeneration.

5. Patient usually dies of uremia.

GG. Usher's Syndrome (Recessive Retinitis Pigmentosa with Congenital Severe Deafness).

1. Autosomal recessive.

2. Retinitis pigmentosa giving rise to progressive visual loss. The patient is usually completely blind by the second or third decade.

3. These patients usually are born deaf secondary to atrophy of the organ of Corti. Hearing for low frequencies may be present in some patients.

4. Ataxia and vestibular dysfunction are very common. Usher's syndrome, among all congenital deafness, is the one most likely to include vestibular symptoms.

5. It constitutes 10% of hereditary deafness.

6. Gorlin et al classified Usher's syndrome into three types.

a. Type I: Profound congenital deafness with the onset of retinitis pigmentosa by 10 years of age; has no vestibular responses; constitutes 90% of all cases of Usher's syndrome.

b. Type II: Moderate to severe congenital deafness with the onset of retinitis pigmentosa in late teens or early 20s; normal or decreased vestibular response; constitutes 10% of all cases.

c. Type III: Progressive hearing loss; retinitis pigmentosa begins at puberty; constitutes less than 1% of all cases.

Types I, II, III are autosomal recessive.

c. Type IV: X-linked inheritance; phenotype similar to type II.

HH. Weil's Syndrome.

1. Nephritis.

2. Hearing loss.

3. Autosomal dominant.

V. Middle and External Ear Congenital Deformities

A. These have been classified into class I, II, III. However, the classification is less commonly used than that for inner ear developmental anomaly.

1. Class I.

a. Normal auricle in shape and size.

b. Well pneumatized mastoid and middle ear.

c. Ossicular problem.

d. This type is the most common.

2. Class II.

a. Microtia.

b. Atretic canal and abnormal ossicles.

c. Normal aeration of mastoid and middle ear.

3. Class III.

a. Microtia.

b. Atretic canal and abnormal ossicles.

c. Middle ear and mastoid poorly aerated.

B. External deformity does not correlate necessarily with the middle ear abnormality.

C. Patients with congenitally fixed footplate have the following points to differentiate them from those patients with otosclerosis.

1. Onset in childhood.

2. Nonprogressive.

3. Negative family history.

4. Flat 50-60 dB conductive hearing loss.

5. Carhart's notch is not present.

6. Schwartze's sign is not present.