

Syndromes commonly associated with hearing loss in children

It is estimated that at least 50% of congenital hearing loss is due to hereditary factors. Approximately 70% of hereditary hearing loss is non-syndromal.

Patterns of inheritance of non-syndromal hearing loss can be autosomal recessive, autosomal dominant, x-linked, and mitochondrial.

Here is a list of syndromes commonly associated with hearing loss:

Alport syndrome: collagen synthesis disease characterized by renal disease

Alström syndrome: pigmentary retinopathy, diabetes mellitus, and obesity

Apert Syndrome: craniosynostosis, syndactyly of hands and feet, mental retardation

Branchio-Oto-Renal syndrome: kidney, ears, and neck abnormalities

Charcot-Marie-Tooth: motor and sensory neuropathy, nephritis

CHARGE syndrome: acronym for the set of congenital features: Coloboma of the eye, Heart defects, Atresia of the nasal choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, and Ear abnormalities and deafness.

Chondrodysplasias, e.g. achondroplasia

Crouzon Syndrome: craniosynostosis, maxillary hypoplasia, shallow orbits

DiGeorge sequence: cardiac defects, Thymus hypoplasia and/or T cell-mediated immunodeficiency, and hypocalcemia and/or absence of parathyroids– (part of deletion 22q11 spectrum)

Down Syndrome

Ehlers-Danlos syndrome: synthesis of collagen defects, characterized by hypotonia, ocular abnormalities, joint hypermobility

Friedreich ataxia: spinocerebellar, resulting in progressive gait ataxia

Goldenhar syndrome: incomplete development of the ear, nose, soft palate, lip, and mandible (part of the oculo-auriculo-vertebral spectrum)

Hemifacial microsomia: abnormal development of the lower half of the face, most commonly the ears, the mouth and the mandible (part of the oculo-auriculo-vertebral spectrum)

Hunter syndrome (mucopolysaccharidosis II): a lysosomal storage disease characterized by progressive intellectual impairment, death between 10 and 15 years

Hurler syndrome (mucopolysaccharidosis I): a lysosomal storage disease characterized by coarse facial features, skeletal malformations, recurrent otitis media, hepatosplenomegaly, and macroglossia, developmental delay, death by 10 years

Jervell and Lange-Nielsen syndrome: variant of long QT syndrome (see below)

Klinefelter syndrome (XXY): hypogonadism, infertility

Large Vestibular Aqueduct Syndrome: enlargement of vestibular aqueduct in the inner ear

Long QT syndrome: prolongation of QT on ECG, syncope, and sudden death

Neurofibromatosis II (NF2): tumours of the central and peripheral nervous system, including non-malignant vestibuloschwannomas

Noonan syndrome: short stature, characteristic facial features, hypotonia, cardiac abnormalities

Norrie syndrome: retinal detachment, possible mental retardation

Ohdo syndrome: mental retardation, congenital heart disease, blepharophimosis/ptosis, hypoplastic teeth

Osteogenesis imperfecta: disorder of type I collagen metabolism characterized by bone fragility

Osteopetrosis: increased osseous density due to defects in osteoclastic resorption

Pendred syndrome: goitre and hypothyroidism

Pfeiffer syndrome: craniosynostosis

Pierre Robin sequence: craniofacial abnormalities

Refsum syndrome: phytanic acid storage disease characterized by microcephaly, severe developmental delay, hypotonia, hepatomegaly, retinitis pigmentosa and dysmorphic facial features

Saethre-Chotzen Syndrome: craniofacial anomalies including variable craniosynostosis

Stickler syndrome: flat midface, cleft palate, myopia with retinal detachment and cataracts, musculoskeletal findings

Treacher Collins syndrome: craniofacial abnormalities

Turner syndrome: XO genotype characterized by short stature, infertility, renal abnormalities, chronic otitis media

Usher syndrome: retinitis pigmentosa and vitiligo

Velocardiofacial Syndrome: (part of 22q11 deletion spectrum) typical characteristics include cardiac abnormality (especially Fallot's Tetralogy), abnormal facies, thymic aplasia, cleft palate, hypocalcemia

Waardenburg Syndrome: white forelock, heterochromia of irises

Primary reference:

Toriello, H., Reardon, W., & Gorlin, R., eds. *Hereditary Hearing Loss and its Syndromes*, 2nd edition. New York, Oxford University Press, 2004.

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