Editorial

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Editorial on Pfeiffer Syndrome

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Editorial

Pfeiffer syndrome is a rare genetic disorder characterized by premature fusion of certain skull bones and other birth defects in the hands and feet. The abnormality of the skull bones causes changes in the shape of the face and head. Most affected individuals also have differences to their midface and conductive hearing loss. This syndrome also affects bones in the hands and feet. It's also referred to as acrocephalosyndactyly type V, ACSV, craniofacial-skeletal-dermatologic syndrome, and Noack syndrome [1-3].

Types of Pfeiffer

Type I Pfeiffer: People usually have a normal lifespan and typical intelligence.

Types II and III Pfeiffer: People have more severe defects that can impair brain development and function.

Symptoms

The primary features of Pfeiffer include craniosynostosis and turribrachycephaly. Additional features may include a high, full forehead; underdeveloped midfacial regions (midface hypoplasia); widely spaced eyes (ocular hypertelorism); an underdeveloped upper jaw (hypoplastic maxilla), with a prominent lower jaw; and dental abnormalities. Bulging eyes, High forehead, Protrusion of the eyes, Hearing loss (in over 50% of those affected), brachydactyly, syndactly, Cloverleaf-shaped head are some major symptoms.

Causes

Pfeiffer syndrome type I is associated with mutations in genes known as FGFR1 and FGFR2. Pfeiffer syndrome type II and type III are associated with mutations in FGFR2.

Diagnosis

The diagnosis of Pfeiffer syndrome is based on clinical findings. Pfeiffer syndrome is diagnosed by the presence of the characteristic birth defects. If the diagnosis is uncertain, genetic testing can be done to identify changes in the FGFR1 and/or FGFR2 genes.

Treatment

There is no treatment that can reverse Pfeiffer syndrome, but treatments are available that address each individual's specific symptoms. Treatments may be needed from a variety of specialists, including surgeons, pediatricians, otolaryngologists (ENT specialists), neurologists, or others. Reconstructive surgeries can help overcome some of the physical defects associated with the syndrome. Specific therapies for Pfeiffer syndrome are symptomatic and supportive. In some cases, reconstructive surgery may be performed to help correct ear malformations and/or specialized hearing aids may be used to improve conductive hearing loss. Genetic counseling is recommended for affected individuals and their families. In addition, thorough clinical evaluations may be important in family members of diagnosed individuals to detect any symptoms and physical characteristics that may be potentially associated with Pfeiffer syndrome.

References

1 https://rarediseases.org/rare-diseases/pfeiffer-syndrome/

2 https://medlineplus.gov/genetics/condition/pfeiffer-syndrome/#resources

3 https://www.medicinenet.com/pfeiffer_syndrome/article.htm#what_is_the_life_expectancy_of_an_individual_with_pfeiffer_syndrome