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The Medical Bulletin

In Syndrome

Treacher Collins Syndrome

Synonyms :

- Mandibulofacial dysostosis
- Franceschetti-Zwalen-Klein syndrome
- Treacher Collins-Franceschetti syndrome

Definition :

Treacher Collins syndrome is a rare autosomal dominant disorder of craniofacial development.

Etiology :

- Due to mutation of TCOF1, POLR1B, POLR1C or POLR1D genes.
- Most commonly TCOF1 - autosomal dominant, rarely autosomal recessive.
- Mutation in TCOF1 causes reduced amount of Treacle protein which causes reduced number of cranial neural crest cells migrating to 1st & 2nd pharyngeal arches.

Prevalance : 1 in 10,000 - 50,000

Clinical Features :

- Abnormalities – symmetrical & congenital.
- Otological – absent external auditory canal, auricular tags,pits,fistula, dysplastic ears & conductive hearing loss.
- Opthalmological - downward slanting palpebral fissures, coloboma, absence of eyelashes & hypertelorism.
- Other – flat malar region, hypoplasia or absence of zygomatic bones, hypoplasia of the mandible (Micrognathia or Retrognathia), cleft/high arched palate & macro/microstomia.

Diagnosis :

- Genetic testing for TCOF1.
- Prenatal USG - Hypoplasia of the maxilla and zygomatic bone, Micrognathia, Cleft palate, Malformed or absent ears.

Treatment :

- Multidisciplinary approach depending on age.
- Airway obstruction – Mandibular Distraction osteogenesis.
- Cleft palate – Palatoplasty at 9 – 12months of age.
- Hearing loss - Prosthetic auricular reconstruction and bone-assisted hearing aid (BAHA) placement.
- External ear reconstruction at 8years of age.
- Reconstruction of cheek & maxilla at 5 – 7years of age.
- Orthognathic surgery at 13 -18years of age.
- Nose reconstruction at 18years of age.

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