Treacher Collins' Syndrome

Synonyms: mandibulofacial craniosynostosis; mandibulofacial dysostosis, Berry-Treacher Collins syndrome, Franceschetti-Zwahlen-Klein syndrome, Thomson complex

Treacher Collins' syndrome is a disorder of craniofacial development. There is much clinical and genetic variation. Most have normal intelligence.

Genetics

Treacher Collins' syndrome is an autosomal dominant disorder with variable expression. The critical region is at chromosome 5q31.3-32. More than half of all cases are thought to be new mutations because there is no family history of the disease. It is suspected to be a ribosomopathy, where a genetic abnormality causes impaired ribosome biogenesis and function, resulting in a specific clinical phenotype.

Epidemiology

Incidence is approximately 1 in 50,000 live births.

Presentation

- The main features are antimongoloid palpebral fissures, coloboma of the lower eyelids, eyelash malformations, molar defects, preauricular hair displacement, flat cheekbones (hypoplastic zygomatic arches), micrognathia and 'fish-like' facial appearance.
- The common features are macrostomia, low-set ears and ear defects, high-arched palate, nasal deformity, teeth malocclusion, open bite and conductive hearing loss.
- Other abnormalities (cleft palate, colobomas of the upper lid, hypertelorism and mental retardation) are infrequent.
- In severely affected patients the airway is compromised by the mandibular deficiency, glossoptosis and choanal atresia. Sleep apnoea and sudden infant death syndrome may occur.

Differential diagnosis

- Oculo-auriculovertebral dysplasia
- Goldenhar's syndrome.
- Nager's acrofacial dysostosis (similar facial features).

Investigations

- The earliest possible diagnosis is by chorionic villus sampling (if there is a family history).
- Diagnosis may also be made at midtrimester antenatal ultrasound.
- Postnatally, diagnosis is essentially made on clinical features. A thorough assessment must be made for all associated features, especially those affecting breathing, and complications, eg conductive hearing loss.

Management

The spectrum and degree of deformities are extensive and therefore the nature and intensity of management are also very variable.

General points

- Affected children and their families may need a great deal of support.
- Hearing and speech: hearing aids, speech therapy.

Surgical

- In severe cases the airway must be evaluated and secured from birth. Either positioning alone or tracheostomy is required to manage the airway, and a gastrostomy required for feeding.
Operative correction of cleft palate may be necessary. Operations of choanal atresia or mandibular lengthening are performed at the age of 2 to 3 years or later. The timing of bone and soft tissue reconstruction will vary but bone reconstruction should usually precede soft tissue corrections:

- Autogenous tissues, e.g., ribs or iliac bone, should be used and synthetic materials avoided.
- Soft tissue reconstruction includes correction of lower eyelid coloboma, and ear reconstruction.

### Complications

- Feeding difficulty.
- Hearing loss.
- Speech delay.
- Hearing and speech difficulties may lead to educational difficulties.
- Self-awareness and bullying resulting from the affect on appearance may lead to psychological difficulties.

### Prognosis

Most become normally functioning adults with normal intelligence. Careful attention should be given to any hearing problem in order for the child to realise their full potential.

### Prevention

Genetic counselling is recommended for prospective parents with a family history of Treacher Collins’ syndrome.

### Further reading & references

- Treacher Collins-Franceschetti Syndrome, Online Mendelian Inheritance in Man (OMIM)

1. Tolarova MM;et al; Mandibulofacial Dysostosis (Treacher Collins Syndrome), eMedicine, Nov 2009

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