

Treacher Collins syndrome

Treacher Collins syndrome is a genetic disorder that affects growth and development of the head. It prevents the skull, cheek and jawbones from developing properly, causing facial defects and hearing loss. About one child in every 50,000 is affected. Problems range in severity from mild to very severe. In most cases, the child's intelligence is normal.

Treacher Collins syndrome is either inherited or caused by a new genetic mutation at the time of conception. There is no cure, but skull and face (craniofacial) surgery can improve speech and create a more normal appearance. Treacher Collins syndrome is also known as mandibulofacial dysostosis or Franceschetti syndrome.

Symptoms

Symptoms and signs range from barely noticeable to severe and disabling. Typically, the characteristics of a person with Treacher Collins syndrome may include:

- Cleft palate
- Small jawbone (micrognathia)
- Disproportionately large mouth (macrostomia)
- Small or absent cheekbones
- Large and pointed nose
- Droopy misshapen eyes with notched lower lids
- Absent lower eyelashes
- Absent floor of the eye sockets
- Overgrowth of scalp hair onto the cheeks
- Low-set, misshapen, small or absent ears
- Deformities of the ear canal
- Conductive hearing loss or conductive deafness, caused by malformations of inner ear structures.

A faulty gene

The particular gene that causes Treacher Collins syndrome is called TCOF1. This gene, located on chromosome 5, is responsible for facial development. In about half of all cases, TCOF1 spontaneously changes at conception but what triggers the mutation is unknown.

In other cases, Treacher Collins syndrome is inherited as an autosomal dominant trait. This means that the affected child inherits the faulty gene from an affected parent and the correct copy from the other.

Researchers have so far discovered about 50 different changes of TCOF1 that can cause Treacher Collins syndrome. Exactly how the faulty gene causes the facial defects is not known. It is thought to cause cell death (apoptosis) of facial bone, cartilage and soft tissue during weeks 3–8 of fetal development. The apoptosis causes the characteristic facial defects.

Pattern of inheritance

An affected parent has a 50 per cent chance (1 in 2) of passing it on to each child. This is a chance event and cannot be altered. The severity of facial abnormalities in a child who inherits the altered gene cannot be predicted.

Possible complications

Complications depend on the severity of the condition. Generally, Treacher Collins syndrome may lead to complications including:

- **Feeding problems** – the cleft palate prevents the baby from suckling and swallowing.
- **Breathing problems** – the abnormally small jaw and normal-sized tongue can interfere with breathing and lead to sleep apnoea. A tracheostomy (surgical opening in the windpipe) may be needed in severe cases.
- **Speech problems** – caused by the cleft palate, conductive hearing loss or both.
- **Learning problems** – caused by hearing loss or deafness. Treacher Collins syndrome generally does not affect the child's intelligence.
- **Dry eye syndrome** – not enough tears to keep the eyes moist and comfortable. Complications of dry eye syndrome include recurrent eye infections.
- **Psychological problems** – including low self-esteem and depression caused by social stigma.

Diagnosis

Tests used to diagnose Treacher Collins syndrome may include:

- Physical examination
- Medical history
- Genetic testing – this is often not necessary as the diagnosis can be made easily on clinical features alone.

Treatment

Treatment depends on the severity of the condition but may include:

- **Genetic counselling** – for the individual or the whole family, depending on whether the condition was inherited or not.
- **Hearing aids** – usually helpful in the case of conductive hearing loss.
- **Dental work** – including orthodontic work to help correct the child's malocclusion ('bad bite').
- **Speech therapy** – to improve the child's communication skills. Speech pathologists also work with people who have trouble swallowing food or drink.
- **Surgery** – a number of operations are available that can improve function and appearance.

Surgery options

Depending on the severity of the defects, surgical options may include:

- Reconstruction of lower eyelids
- Closure of cleft palate
- Repair of absent facial bones including cheekbone and lower eye socket
- Ear reconstruction
- Re-positioning of the lower jaw (orthognathic surgery)
- Rhinoplasty to improve the look of the nose.

The timing of these operations depends on the child's age and stage of development. For example, eye surgery may be performed during infancy, ear reconstruction between the ages of five and seven years, and facial surgery after adolescence when the face has stopped growing. Generally, reconstructive surgery has good results and helps the person to have a good quality of life.

Where to get help

- Your doctor
- Paediatrician
- Dentist
- Orthodontist
- Craniofacial (skull and face) surgeon

- Genetic counselling services – available at most large public maternity hospitals
- Speech Pathology Australia Tel. (03) 9642 4899
- Genetic Health Services Victoria, Royal Children’s Hospital Tel. (03) 8341 6200
- Genetic Support Network Victoria Tel. (03) 8341 6315
- The Murdoch Children’s Research Institute Tel. (03) 8341 6200

Things to remember

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- In most cases, the child’s intelligence is normal.
- Generally, reconstructive surgery has good results and helps the person to have a good quality of life.

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Genetic Health Services Victoria

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