Treacher Collins syndrome

Summary

- Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial anomalies and hearing loss.
- In most cases, the child’s intelligence is unaffected.
- Generally, reconstructive surgery has good results and helps the person to have a good quality of life.

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 anomalies 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 unaffected.

Treacher Collins syndrome is either inherited or caused by a new change in a gene at the time of conception. There is no cure, but skull and face (craniofacial) surgery can improve speech and reduce some of the more severe craniofacial anomalies.

Treacher Collins syndrome is also known as mandibulofacial dysostosis or Franceschetti syndrome.

Symptoms of Treacher Collins syndrome

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 misshaped 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.

Cause of Treacher Collins syndrome

A change in the gene TCOF1 causes up to 93 per cent of cases of Treacher Collins syndrome. 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 gene change is unknown. In other cases, Treacher Collins syndrome caused by the altered TCOF1 gene is inherited as an autosomal dominant trait. This means that the child inherits the affected gene from a parent with Treacher Collins syndrome, and an unaffected copy of the gene from their other parent.

Another two per cent of cases are thought to be caused by changes in the POLR1C and POLR1D genes.

Exactly how the gene changes cause facial anomalies is not known. They are thought to cause cell death
Pattern of inheritance of Treacher Collins syndrome

An affected parent has a 50 per cent chance (one in two) of passing the affected gene on to each child they have. This is a chance event and cannot be altered. The severity of facial anomalies in a child who inherits the altered gene cannot be predicted.

Complications of Treacher Collins syndrome

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

- **feeding problems** – a cleft palate prevents the baby from sucking and swallowing
- **breathing problems** – a small jaw and average-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** – may be caused by a cleft palate, conductive hearing loss or both
- **learning problems** – may be caused by hearing loss or deafness. Treacher Collins syndrome generally does not affect a child’s intelligence
- **dry eye syndrome** – may occur when there are not enough tears to keep the eyes moist and comfortable. Complications of dry eye syndrome include recurrent eye infections
- **psychological problems** – may include low self-esteem and depression caused by social stigma.

Diagnosis of Treacher Collins syndrome

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 based on clinical features alone.

Treatment for Treacher Collins syndrome

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 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 for Treacher Collins syndrome

Depending on the severity of the craniofacial anomalies, 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.
Genetic counselling and Treacher Collins syndrome

If your child has been diagnosed with Treacher Collins syndrome, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Treacher Collins syndrome and what causes it, how it is inherited, and what a diagnosis means for your child’s health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Treacher Collins syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

The Genetic Support Network of Victoria (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Treacher Collins syndrome.

Where to get help

- Your GP (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, or Tel. 1300 368 835 outside Victoria
- Victorian Clinical Genetics Services (VCGS), Royal Children’s Hospital Tel. 1300 118 247
- Genetic Support Network Victoria Tel. (03) 8341 6315
- The Murdoch Children’s Research Institute Tel. (03) 8341 6200

This page has been produced in consultation with and approved by:

Victorian Clinical Genetics Services (VCGS)

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