Treacher-Collins syndrome

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Treacher-Collins syndrome (also known as mandibulofacial dysostosis) and where to get help.

What is Treacher-Collins syndrome?

Treacher-Collins syndrome is a congenital (present at birth) condition affecting the bones and tissues in the face. Early in pregnancy, the cheekbones, jaw and eye sockets do not develop properly. This causes a characteristic appearance, with downward drooping eyes and a small jaw.

What causes Treacher-Collins syndrome?

Treacher-Collins syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified three genes affected: TCOF1 which is the most common gene mutated as well as the genes POLR1C and POLR1D. These genes are responsible for the formation of proteins that play an important role in how bone and tissue cells develop early in pregnancy.

The gene mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). If the TCOF1 or POLR1D gene mutation is inherited, it is passed on in an autosomal dominant manner – this means that a child only has to inherit the faulty gene from one parent to develop the condition. If the mutation is on the POLR1C gene, this is passed on in an autosomal recessive manner – this means that a child has to inherit the faulty gene from both parents to develop the condition.

What are the signs and symptoms of Treacher-Collins syndrome?

The symptoms of Treacher-Collins syndrome are very variable – ranging from mild to severe.

Children with Treacher-Collins syndrome have a characteristic appearance due to the problems with their cheekbones, jaw and eye sockets forming. If the jaw is very small and/or underdeveloped, breathing difficulties may become apparent soon after birth. The jaw problems may also cause feeding difficulties. Some children also have a cleft (hole) palate and coloboma – a notch in the lower part of the eye. The ears are often affected in children with Treacher-Collins syndrome, either being absent or very small (microtia). If the internal structures of the ear are also underdeveloped, this can lead to hearing impairment.

How is Treacher-Collins syndrome diagnosed?

As children with Treacher-Collins syndrome have a characteristic appearance, no specific diagnostic tests are needed. Imaging scans, such as x-ray or MRI may be suggested to monitor bone growth before, during and after treatment. Imaging may also be needed to examine the internal structures of the ear alongside hearing tests to diagnose hearing loss.

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How is Treacher-Collins syndrome treated?

As Treacher-Collins syndrome can affect various areas of the head and face, treatment is best delivered at a specialist centre where a multidisciplinary team approach can be taken. The multidisciplinary team will usually comprise craniofacial (skull and face) surgeons, neuro (brain) surgeons, ear, nose and throat (ENT) surgeons, audiologists (hearing specialists), dentists and orthodontists, geneticists and speech and language therapists with other specialists brought in as needed.

Initially, stabilising a child's breathing problems will require treatment. For some children, these are so severe that they need a tracheostomy – artificial opening into the windpipe – to allow them to breathe. Other children may only need breathing support at night. Feeding problems may be helped with enteral feeding – a feeding tube or gastrostomy directly into the stomach bypassing the mouth and throat. Cleft palate repair (if required) will also be carried out in the first year of life.

Later in childhood, the underdeveloped jaw will require treatment – often with bone grafts and jaw distraction. This is a long term treatment but has good results for the majority of children. Sometimes the cheek bones need to be reconstructed using bone grafts or implants. Ear reconstruction can also be carried out in later childhood, either creating a new ear modelled on the other ear or the parents' ears if both are absent. Ear reconstruction only improves the appearance of the ear, it will not improve function. Children with hearing impairment will need support in terms of hearing aids or cochlear implant as well as speech and language therapy.

As the bone continue to grow during childhood and adolescence, further surgery may be needed to make minor corrections to face shape. Final corrections are usually complete between the ages of 16 and 20 years.

What happens next?

The outlook for children born with Treacher Collins syndrome is variable depending on the severity of their symptoms and the impact it has on bodily functions such as breathing, vision and hearing. They will require long term monitoring, particularly during period of growth in childhood and adolescence, but surgery tends to be completed by the time the child is in their mid-twenties. Some children and families benefit from psychological input at various stages throughout childhood and adolescence.

Further information and support

Headlines – the Craniofacial Support Group – is the main support organisation in the UK for families of children and young people affected by a craniofacial disorder. Visit their website at www.headlines.org.uk.

Changing Faces is another organisation that will be able to offer help and support to anyone living with a condition that affects their appearance. Visit their website at www.changingfaces.org.uk or telephone their helpline on 0845 4500 275.

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