



Treacher Collins Syndrome

The syndrome was named after an ophthalmologist called Edward Treacher Collins in 1900. It can also be known by other names such as Berry-Treacher Collins Syndrome, Franceschetti-Klein Syndrome, Franceschetti-Zwahlen Syndrome and Thomson complex. It is a condition that causes facial malformations and severe hearing loss.

Someone with Treacher Collins Syndrome may have malformed cheekbones, chin, nose, jaw and temples. Eyelids are often drooping, seeming not to support the eyes, and there may be a small nick in the lower lid. The ears may be malformed or completely absent. Hairline and palate may also be unusual.

Depressed cheekbones and sloping eyes

The cheekbones can be underdeveloped or absent. This means that the cheek muscles join onto the lower jaw muscles and so cause sloping eyes. This can be corrected by surgery using implants and bone grafts.

Malformed or absent ears

This will vary greatly from case to case. Some may just have small ears; others may have no ears at all. The outer ear may be folded or squashed. The middle ear can also be malformed or missing. Any hearing loss is almost always conductive and can vary from partial to severe.

Corrective surgery will depend on the individual and on which parts of the ear are missing. Artificial ears can be attached using Titanium, a metal that uniquely bonds with bone. Hearing may be improved with hearing aids. Most often a bone conduction hearing aid is used, attached to a headband, or a Bone Anchored Hearing Aid attached directly to the mastoid bone.

Receded chin

The Treacher Collins baby is often born with a much receded lower jaw. This can improve as the child grows.

Hair growth

This again is quite common at birth. The hairline continues across the cheek forming a definite sideburn towards the mouth. This may only be temporary and varies from child to child.

Other possible problems

Breathing problems can occur. In small children and babies breathing can be very noisy. When colds or infections occur the usual problems are sometimes more severe. This is because the roof of the mouth may be high and the nasal passages very small. Snoring is a common problem and may be loud. Eye infections are common in children.

Dental problems may occur because of the smallness of the mouth. A receded jaw could cause overcrowded teeth or an incorrect bite. All this can be improved by orthodontic treatment. Cleft palate or choanal atresia conditions may be present in severe cases.

Is it inherited?

Yes - but you must have the syndrome to pass it on. For those that have the Syndrome there is a 50-50 chance of having children affected by it. The severity of a parent's condition is not an indication of the possible severity of the child. This means that a severely affected parent may have a mildly affected child, and vice-versa.

Each case will be different and must be assessed individually. The psychological effects of Treacher Collins Syndrome are significant due to facial malformations. It is important to have correct consultation on everything involved.

Over half of Treacher Collins Syndrome cases are a result of fresh genetic mutation, not direct inheritance from parents. These children in turn will have a 50-50 chance of passing it on.

The future

It is important to remember that Treacher Collins Syndrome is not progressive. Children do not get worse. In fact many features can be improved by growth and by surgery and other treatment.

Because it can be inherited people with Treacher Collins Syndrome should seek guidance from a qualified genetic counsellor if considering having children.

Treacher Collins Family Support Group

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