

# TREACHER COLLINS

## Description

Though the features of Treacher Collins syndrome have wide variability between people, it is most noticeably characterized by abnormalities of the head and face. These include: down slanting eyes with notched lower lids; underdeveloped cheek and jawbones, prominent nose, broad mouth and characteristically small chin with steep lower jaw angle. Ears are malformed or prominent and hair may extend towards the face; hearing loss is also possible. A minority of those affected with Treacher Collins syndrome may have: cleft lip and/or palate, heart defects, and strabismus (crossed eyes).

## Prevalence/Causes

Treacher Collins syndrome occurs in approximately 1:50,000 live births. It is a genetic condition that is either inherited from a parent affected with the syndrome or occurs spontaneously as a first-time genetic mutation. An affected parent has a 50% chance of passing the condition on to a child.

## Treatment

Airway, feeding and hearing difficulties are the three main areas requiring immediate treatment. A small jaw may impede newborn breathing and feeding. A tracheostomy (breathing hole) may be required. Patients with Treacher Collins syndrome may undergo multiple surgeries, some of which are cosmetic. These surgeries include: outer ear, repair of eyelid deformities or cheek or jaw enhancements.

## Associated Conditions

- Sleep apnea, difficulty breathing at night that results in frequent wake ups and restless sleep, may be a concern and a sleep study should be done.

- Treacher Collins children may have cleft lip and/or palate (hole in the lip or palate) which results in impaired feeding ability.
- Most children have a 40% hearing loss in each ear due to outer and middle ear abnormalities.
- Thumbs may be abnormally small or absent
- Dry eyes can lead to infection



