



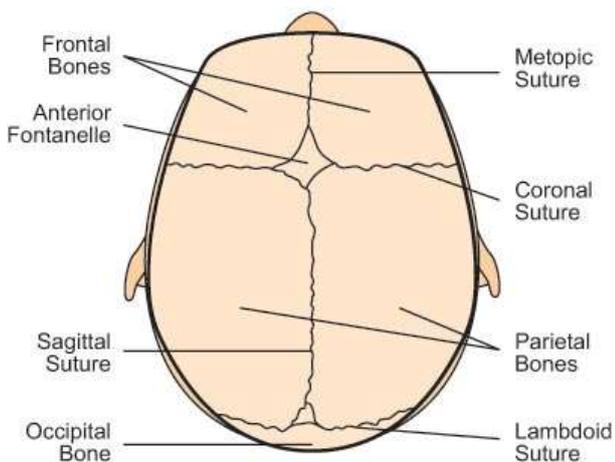
Great Ormond Street Hospital for Children NHS Foundation Trust

## Crouzon syndrome: information for families

**Crouzon syndrome is the most common type of complex craniosynostosis. It is named after the doctor who first described it in the early 20th century. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Crouzon syndrome.**

The skull is made up of several 'plates' of bone which, when we are born, are not tightly joined together. The seams where the plates join are called 'sutures'.

**Normal Skull of the Newborn**



As we grow older, the sutures gradually fuse (stick) together, usually after all head growth has finished. When a child has craniosynostosis, the sutures fuse before birth. It can affect one suture or several.

When more than one suture is affected, it is called 'complex craniosynostosis'. This may happen as part of a syndrome (collection of symptoms often seen together), and so may be referred to as 'syndromic' as well.

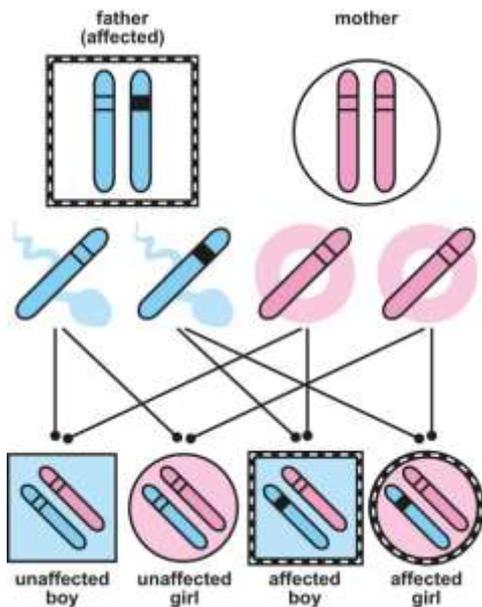
In Crouzon syndrome, both coronal sutures fuse before birth and other sutures may be affected

too, making the skull misshapen. The bones in the midface are also affected, as the cheekbones and upper jaw do not grow in proportion to the rest of the skull. The bones around the eyes (orbits) are wider spaced and shallower than usual, causing the eyes to bulge outwards.

### What causes Crouzon syndrome?

Crouzon syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected genes as the Fibroblast Growth Factor Receptor 2 (FGFR2) gene and FRGR3. Some of these genes may also be involved in Pfeiffer syndrome. They affect how certain cells in the body – including bone cells – grow, divide and die.

The gene mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). If it is inherited, it is passed on in an autosomal dominant manner – this means that if one parent is affected, half their children will inherit the condition.



## What are the symptoms of Crouzon syndrome?

Children with Crouzon syndrome have a characteristic appearance due to the problems with the skull plates fusing and the midface bones not growing in proportion. The degree of fusing and underdevelopment varies from child to child and can be mild or severe.

If the skull plate fusion is severe, pressure can build up inside the brain (raised intracranial pressure) which will require urgent treatment.

There is also an increased risk of developing hydrocephalus. Hydrocephalus occurs when cerebrospinal fluid (CSF) is stopped from circulating or being re-absorbed. The CSF builds up within the ventricles (cavities) of the brain resulting in increased pressure on the brain.

Failure of the midface bones to grow can affect breathing as the airway is narrow.

Vision may also be affected as the eyes are not protected by the orbits and eyelids. These problems will also need treatment early in life. The bones of the spine in the neck area (cervical spine) can also be affected, causing a condition called Chiari malformation, where the base of the brain is squeezed.

Hearing problems are also common due to narrowed ear canals, as are problems with overcrowding of the teeth due to the misshapen upper jaw. Most children with Crouzon syndrome are of normal intelligence.

## How is Crouzon syndrome diagnosed?

As children with Crouzon syndrome have a characteristic appearance, no specific diagnostic tests are needed. Imaging scans, such as x-ray, CT or MRI may be suggested to monitor bone growth before, during and after treatment and to detect hydrocephalus. Genetic tests may be suggested to identify the mutated genes particularly if future pregnancies are planned. Hearing and vision checks will be needed throughout childhood.

## How is Crouzon syndrome treated?

As Crouzon syndrome can affect various areas of the body, 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, ophthalmologists (eye specialists), audiologists (hearing specialists), dentists and orthodontists, geneticists, psychologists and speech and language therapists with other specialists brought in as needed.

Depending on the severity of the skull fusion, treatment soon after birth may be needed if pressure inside the skull is raised, breathing problems are severe or there is a risk of eye damage.

In many cases, initial skull re-shaping surgery takes place within the first few years of life. This will involve cutting through the fused sutures in the skull and re-shaping them to give a more normal skull shape.

Further surgery to improve the midface problems will usually be carried out in late childhood, most commonly using a rigid external distraction (RED) frame to gradually pull the affected bones forward over a period of weeks and months. The eye sockets will also be re-shaped so that the eyes sit more deeply inside the skull so that the eyelids can close fully to protect the eyes.

Chiari malformation (if present) can be treated by widening the opening at the base of the skull). Orthodontic treatment using braces will be suggested to improve overcrowding and speech. Hearing aids may also be needed.

As the bone continues to grow during childhood and adolescence, further surgery may be needed to make corrections to the skull shape and midface area.

## 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](http://www.headlines.org.uk)

Changing Faces is another organisation that offers help and support to anyone living with a condition that affects their appearance. Visit their website at [www.changingfaces.org.uk](http://www.changingfaces.org.uk) or telephone their helpline on 0845 4500 275.

## What is the outlook for children and young people with Crouzon syndrome?

The outlook for children born with Crouzon 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 early twenties and the growth of the face is complete.

Some children and families benefit from psychological input at various stages throughout childhood and adolescence. Children are of normal intelligence so usually do well at school, college and university.