Headlines
Craniofacial Support

CROUZON SYNDROME

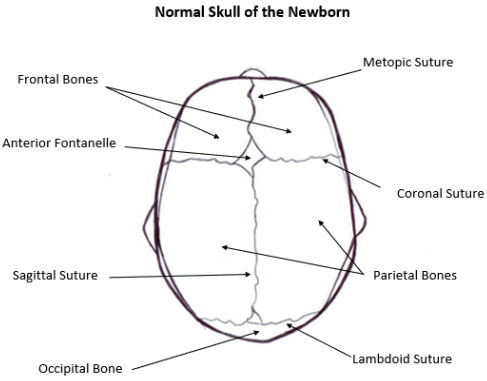
An introductory guide for parents and carers

If your child has just received a diagnosis of Crouzon syndrome, you may be feeling a bit overwhelmed. There can be a lot of information to take in, particularly at a time when you may only just be getting used to having a new baby. This leaflet is intended to help you understand your child's condition and the treatment that you may be offered.

Crouzon syndrome is the most common type of syndromic or 'complex' craniosynostosis. However, although it is thought to occur in around 1 in 25,000 births, it is still very rare.

The skull is made up of several plates of bone which meet at gaps (sutures), called the sagittal, coronal, metopic and lambdoid sutures. The sutures allow the bones of the skull to overlap slightly so that the baby's head can pass through the birth canal. The sutures also enable the skull to expand to accommodate the brain, which grows rapidly during the first two years of life.

Normally, sutures join (fuse) during during late childhood into early adulthood, when brain growth has finished. When a child has craniosynostosis, the sutures fuse too early, usually before birth. It can affect one suture or several.



In Crouzon syndrome, one or 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 (a condition known as 'proptosis'). The arrangement of teeth (dentition) is also affected.

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. They affect how certain cells in the body – including bone cells – grow, divide and die.

The gene mutation can be inherited (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, there is a 50% chance their child will inherit it.

What are the symptoms of Crouzon syndrome?

Children with Crouzon syndrome have a characteristic appearance because of 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.

Although the head shape is often the most striking initial feature, there are a range of other symptoms. If multiple sutures are involved, then surgery may be required when the baby is quite young, to prevent intracranial pressure (a build up of pressure within the skull).

Hydrocephalous (which is when cerebrospinal fluid (CSF) is stopped from circulating or being re-absorbed) can also arise, but this is something that will be closely monitored.

Failure of the midface bones to grow properly can affect breathing because the airway is narrow, and eyes may be affected because they are not protected by the orbits and eyelids.

The bones of the spine in the neck area (cervical spine) can also be affected,so they will be carefully monitored as your baby grows, to detect any problems such as 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 because of the misshapen upper jaw. However, intelligence is not usually affected – most children with Crouzon syndrome are of completely normal intelligence.

Treatment and surgery

Because Crouzon syndrome is such a rare condition, treatment is best delivered at a specialist centre where a multidisciplinary team approach can be taken. There are 4 Designated Highly Specialised Craniofacial Units in England and 1 in Scotland – further details of these services and how to contact them are available on the Headlines website.

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.

Once you have been referred, your baby will usually undergo a number of investigative tests. These may include MRI and CT scans and a review by a clinical geneticist.

Treatment will be individualised according to how Crouzon syndrome has affected your baby. It may involve surgery on the skull early on in life, or treatment of any breathing or eye protection concerns.

Orthodontic treatment using braces will usually 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.

All children with Crouzon syndrome will be monitored regularly to ensure that any problems are identified quickly and treatment offered promptly.

What does the future hold for my child?

Children born with Crouzon syndrome in the UK today benefit from an array of recent advances in surgical techniques and treatments. The outlook for individual children depends on the severity of their symptoms and the impact the condition has on functions such as breathing, vision and hearing.

Children with Crouzon syndrome will require long term monitoring, particularly during periods of growth in childhood and adolescence, but surgery tends to be completed by the time children are in their early twenties and the growth of the face is complete.

Some children and families benefit from psychological support at various stages throughout childhood and adolescence, and into adulthood. However, with the right support, most children born with Crouzon syndrome will go on to do well at school and college.

Headlines produces a number of other leaflets covering different aspects of craniosynostosis and treatment. Please email helpline@headlines.org.uk for further information.

Headlines is the only national charity supporting those affected by craniosynostosis and rare craniofacial conditions. As well as providing information for parents, carers and families, we also offer a confidential helpline, a member magazine and regular newsletters, conference & information days, opportunities for members to meet, and an Annual Family Weekend.

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