

■ Crouzon Syndrome (*Headlines factsheet*)

■ Introduction:

Crouzon syndrome is an inherited syndrome of craniofacial dysmorphism, or abnormal craniofacial appearance, which was originally described in 1912 and is now well recognised. It is thought to occur in 1 in 25,000 births.

Children who have Crouzon syndrome have a range of problems of variable severity, from mild facial symptoms causing a primarily cosmetic concern, to severe symptoms affecting breathing feeding, vision and brain development.

The child with Crouzon syndrome usually enters a co-ordinated programme of care involving many different clinical specialities integrating their various expertise, which often continues from birth to the later teenage years. In addition, there is an on-going programme of research into many aspects of Crouzon and its related syndromes, to constantly investigate and update the services that the specialist teams provide.

■ The Child with Crouzon Syndrome:

Crouzon syndrome predominantly affects the appearance of the head and face.

The **skull**, or *calvarium*, is made up of flat plate-like cranial bones which are connected by seam-like joints, or cranial *sutures*. There are many such sutures, but the most clinically important are the *metopic* and *sagittal* sutures, running from front to back and interrupted by the anterior fontanelle (soft spot); and the paired *coronal* and *lambdoidal* sutures. These run from side to side, the coronal sutures run from the anterior fontanelle to the temple and the lambdoidal from the posterior fontanelle to the back of the base of the skull. Neighbouring cranial bones are thus mobile against each other, and this first allows normal birth, and then growth of the brain inside the skull without restriction.

During normal childhood and into adulthood, the sutures fuse, becoming bone in a seemingly pre-programmed fashion, protecting the brain within. The growth of the face and skull are, of course, closely integrated, and facial bones are also joined by sutures.

In Crouzon syndrome, either or both of the skull and face may be affected. In the skull, the cranial sutures may fuse prematurely, and this is called *craniostenosis* or *craniosynostosis*. This alters the pattern of skull growth, and thus the shape of the skull, sometimes with consequences for the developing brain. There are characteristic skull shapes depending upon the pattern of sutural fusion. Crouzon syndrome may involve any combination of cranial sutures, most commonly including the coronal and sagittal sutures. Common terms for the resulting head shapes are 'brachycephaly', giving a flat forehead, 'scaphocephaly', or boat shaped skull and 'turriccephaly' - tower shaped skull; and these may result in different pressures upon the growing brain.

Craniosynostosis usually begins during pregnancy or the first year of life and is complete by three years of age. Raised intracranial pressure may become a clinical concern.

In the **face**, the commonest features are a regressed mid-face and *shallow orbits* (eye sockets), which may be present at birth or become more evident as the childhood progresses. The arrangement of the **teeth**, or dentition, is also affected, and this requires specialist orthodontic care; which is dependent on good general dental care at home. Rarely, there may be **palate** problems. Seen from the side, the face has a concave appearance, and the shallow orbits result in prominent eyeballs or '*proptosis*'.

■ Clinical problems and programme of care:

The child with Crouzon syndrome may thus have a range of clinical problems. Although the head shape is often the most striking initial feature, from the outset the major concerns are the ease of breathing and potential feeding problems.

The regressed mid-face, or 'maxillary hypoplasia', results in a small larynx and pharynx behind the nose and mouth. This restricts the passage of air into the trachea (windpipe) and lungs, and causes respiratory distress,

particularly at night when snoring and snuffling can interrupt sleep. The degree of airway obstruction and quality of sleep is assessed by a sleep study, and if necessary, treatment takes the form of CPAP (Continuous Positive Air Pressure) devices at home or surgical intervention. Similarly, the passage of food is restricted, and regurgitation may result in aspiration of food into the lungs.

The shallow orbits and proptosis may threaten the cornea, or surface of the eyeball, with '*exposure keratitis*'; and surgical measures may become necessary to protect the exposed eyes. All children have regular ophthalmic review, as other ocular problems may uncommonly occur. Ear, nose, and throat (ENT) follow-up is also recommended, as some children have hearing difficulties, and grommets may be advised to treat chronic infections and improve hearing. The common surgical approaches are given in the table below.

The abnormal skull shape may require surgery to protect the constricted brain and help relieve raised intracranial pressure, which is most commonly revealed by headaches or visual changes identified by the ophthalmologist. The aim of this surgery, called cranioplasty, is to provide a more normal head shape and increase the volume of the skull. Examples of cranioplasty include '*frontal*' and '*fronto-orbital advancement*', '*vault expansion*', and '*frontal*' or '*posterior remodelling*'. Pressure is thus taken off the growing brain. Another surgical method of reducing intracranial pressure is to insert a ventriculoperitoneal shunt, and either or both may be used at different times during childhood, as the skull shape of Crouzon syndrome may be only one of many contributing factors to raised intracranial pressure.

At the current time it is impossible to predict how the skull and face abnormalities will progress as the child grows, and surgical decisions are made in the light of circumstances as they arise. In addition, 'variability of expression' characterises the condition - some children may have mild mid-face problems only, others may have craniosynostosis only, whilst others may have severe craniosynostosis and mid-face regression together. Surgery may be indicated for cosmetic reasons only, or for the more serious conditions described.

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Non-surgical aspects:

Developmental delay in the absence of raised intracranial pressure in Crouzon syndrome is rare. The psychology and language therapy teams have many means of identifying and treating developmental delay early, and their important role in the overall care of the child is emphasised.

Though many new cases are spontaneous, Crouzon syndrome can run in families. When this happens it does so in a 'dominant' manner. The genetic basis of the syndrome is one of the recent research discoveries. Although the inheritance is 'dominant' it must be remembered that the 'expression' of the disease in the child is variable, and most children of a Crouzon family will not be severely affected. The geneticist will advise about risk in subsequent generations of a family.

Summary:

Crouzon syndrome is an inherited syndrome affecting craniofacial growth and development. The care of the child with Crouzon syndrome is multi-disciplinary, involving the co-ordinated expertise of many clinical teams. Surgical care is staged throughout life from infancy to late adolescence, and may follow the following pattern as clinical circumstances arise:

Operation	Age	Indication
Cranioplasty	Infancy	Skull expansion and remodelling for cosmetic benefit and to relieve intracranial pressure
Shunt surgery	Childhood	Neurosurgical operation to reduce intracranial pressure

Facial advancement	ChildhoodAdolescence	To protect the eyes, protect against breathing difficulty, and provide cosmetic benefit. Often preceded and followed by a programme of orthodontics.
Choanal dilatationGrommet insertionBone-anchored hearing aid	Childhood	ENT procedures to improve the airway, treat chronic ear infection, aid hearing.
Squint surgeryTarsorrhaphy	Childhood	To correct ocular squint and improve vision. Tarsorrhaphy may be used to protect against exposure damage to surface of eye.
