Management of craniofacial abnormalities

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A number of congenital and acquired conditions can affect the skull, face and jaws resulting in a wide range of craniofacial abnormalites that commonly present at birth or in early infancy. This article aims to outline the aetiology, pathogenesis, diagnosis and principles of management of those conditions that commonly present to a craniofacial unit namely the craniosynostoses.

Craniosynostosis is premature fusion of one or more of the craniofacial sutures. Management of the patient, their condition and their family is often complex and requires a multidisciplinary approach. The pathogenesis, diagnosis, management and description of the more common craniosynostosis syndromes will be discussed in this article.

Craniosynostosis affects approximately 1 in 2500 children (Johnson, 2003). It may be simple involving one suture, or compound involving two or more sutures. The condition may be primary, or less commonly, secondary to a failure of brain growth or an underlying metabolic or haematological disorder (e.g. hyperthyroidism, thalassaemia). The majority present as isolated non-syndromic craniosynostoses, but some will be part of a recognizable syndrome often associated with deformities of other parts of the body and are termed syndromic craniosynostoses (*Figure 1*).

AETIOLOGY

The fields of molecular biology and genetics have recently contributed significantly to the understanding of the cause of craniosynostosis, particularly the syndromic variety. Mutations in the genes encoding fibroblast growth factor receptors 1, 2 and 3 (FGFR-1, FGFR-2, FGFR-3), MSX2 and TWIST have been identified in syndromic craniosynostosis (Johnson, 2003). The cause of isolated craniosynostosis is less clear, although environmental factors are recognized (e.g. drugs, maternal infection). Some may be associated with chromosomal abnormalities, and defects in the FGFR-3 gene have been identified in a proportion of isolated coronal suture synostoses.

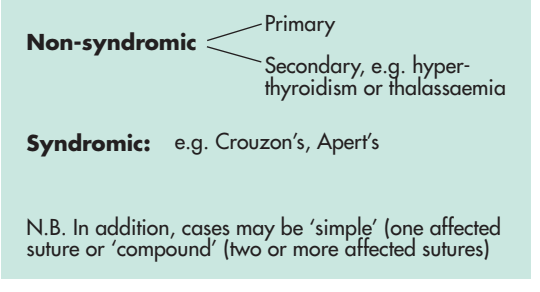

Figure 1. Classification of craniosynostoses

PATHOGENESIS

Normal skull growth is driven by the growth of the underlying brain. The brain reaches approximately 80% of its adult size by the age of 3 years (Muhling, 1999). Premature fusion of the cranial sutures leads to a restriction in growth perpendicular to the direction of the suture (Virchow's Law) (Virchow, 1881). Compensatory growth occurs at other sutures to accommodate the enlarging brain, resulting in characteristic abnormalities of the skull.

The exact sequence of events leading to premature fusion is not known. Virchow (1881) originally believed the primary event occurred at the fused suture itself. Later it was suggested that the cranial vault sutures fuse secondary to a primary

TABLE 1.
Craniofacial Conditions

Craniosynostosis – Non-syndromic and syndromic (craniofacial dysmorphism): e.g. Crouzon's, Apert's, Saethre-Chotzen syndromes
Cranio-orbital-nasal deformities: e.g. hypertelorism, frontonasal dysplasia
Facial clefts
Encephalocoeles
Otocephalic syndromes : e.g. Hemifacial microsomia, Teacher Collins
Craniofacial trauma
Tumours

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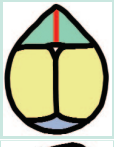
TABLE 2.
Common craniosynostoses

Term	Affected suture	Shape of skull	View from above
Scaphocephaly	Sagittal	Boat-shaped skull, long antero-posterior and narrow transverse dimension	
Brachycephaly	Bilateral coronal	Short AP and broad transverse skull, also usually taller	
Trigonocephaly	Metopic	Triangular shaped forehead, pointed in the midline, often with deficient supra-orbital rims laterally	
Plagiocephaly	Unilateral coronal	Twisted or skewed skull, often with asymmetric forehead	
Occipital plagiocephaly	Lamdoid (rare) or deformational (common)	Skewed or flattened shape to back of head +/- compensational changes to forehead	

anomaly at the cranial base sutures (Moss, 1959). More recently the role of abnormal signals from the underlying dura in maintaining suture patency was discovered by Levine et al in 1998.

Whatever the true underlying cause, the resultant disturbance in growth in the cranial vault and base leads to abnormalities of both shape and volume of not only the skull, but deformity in the face, notably the forehead, orbits and midface.

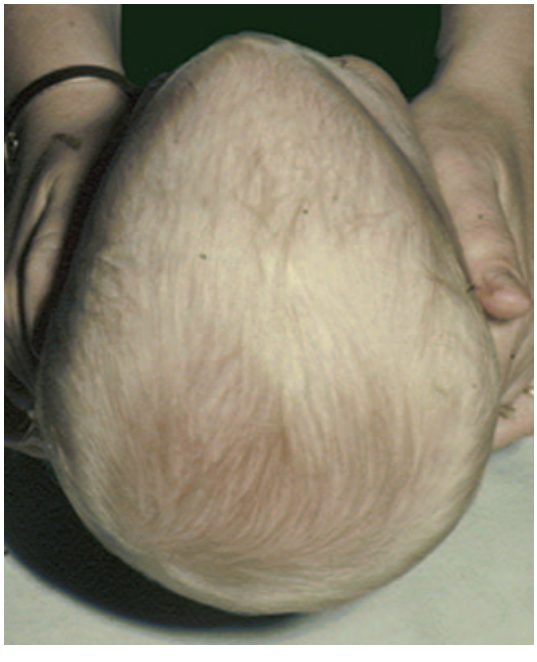


Figure 2. Trigonocephaly.

Abnormalities of shape lead to cosmetic and subsequent psychological sequelae, whereas abnormalities of volume give rise to functional problems in addition to their cosmetic effects.

DIAGNOSIS

Clinical diagnosis of craniosynostosis is mainly on the basis of head shape and clinical appearance. Involved sutures may feel ridged when palpated. While severe cases may occasionally be diagnosed antenatally, the majority of cases present at birth or in the first few months of life as growth occurs and the deformity comes to the attention of concerned parents or professionals.

The syndromic craniosynostoses are often apparent at birth because of the characteristic facial appearance or associated systemic abnormalities. The skull deformity in all craniosynostoses is usually classified in terms of the involved suture and resultant head shape (*Table 2*).

The following terms describe premature fusion of the affected sutures:

- One coronal suture – frontal plagiocephaly (plagio = skewed)
- Both coronal sutures – brachycephaly (brachy = short)
- Metopic suture – trigonocephaly (trigono = triangular) (*Figure 2*)
- Sagittal suture – scaphocephaly (scapho = keel-shape) (*Figure 3*)
- Lambdoid suture – occipital plagiocephaly
- Multiple sutures – most commonly seen in the syndromic synostoses. Various shapes are possible including the very severe coloverleaf (trilobed) skull when all the sutures are affected.

Although there are over 100 recognized syndromes with craniosynostosis as a feature, the most common syndromes are described below (Gorlin, 1990).

Crouzon's

Crouzon's affects only the face and skull and is usually characterized by bilateral coronal synostosis leading to brachycephaly. Involvement of the cranial base gives rise to the typical appearance of midface retrusion, exorbitism, apparent mandibular prognathism and there may be mild hypertelorism. Normal intellect is usual.

Apert's

Although the facial appearance is similar to Crouzon's it is usually more severe and turribrachycephaly ('turri' = tower) with a skull vault that is short from front to back but tall in the vertical plane is typical. Syndactly of the feet and hands is a major diagnostic criterion. Cleft palate is often an associated condition and the midface

retrusion can result in respiratory embarrassment, sleep apnoea and middle ear conditions. Intellectual impairment has been reported in up to 30% of cases.

Pfeiffer

Pfeiffer also affects the face, skull, hands and feet. It is characterized by brachycephaly, midface retrusion and often mild hypertelorism. The thumbs and great toes are broad with occasional soft tissue syndactly of the hands.

Saethre-Chotzen

Saethre-Chotzen is characterized by mild forehead and midface retrusion with low hairline and eyelid ptosis.

Functional Problems

Failure of cranial vault and base growth causes deficits in cranial, orbital and midface volume. In turn, this may lead to raised intracranial pressure (ICP), orbital proptosis, airway compromise and dental malocclusion.

Raised ICP occurs in 13% of single suture synostoses, but is more common in the syndromes and multiple suture synostosis (Renier et al, 2000). A history of failure to thrive, feeding difficulties and failure to reach normal developmental milestones may indicate underlying raised ICP. Its presence is one of the indications for surgery, with the aim of increasing intracranial volume and therefore decreasing ICP.

In severe syndromic cases with reduced orbital volume, proptosis may be severe and rarely may even prevent eye closure and corneal protection. This requires urgent management to prevent visual loss.

In severe midface retrusion (e.g. Apert's) there may be a threat to the airway particularly in neonates. Sleep apnoea or recurrent respiratory infections may occur in the older child. Other functional problems caused by associated abnormalities make full multidisciplinary team (MDT) assessment mandatory.

DIFFERENTIAL DIAGNOSIS

Deformational plagiocephaly

Deformational plagiocephaly (DP) is the most important and the most common differential diagnosis in children presenting with abnormal head shape. Babies' skulls are mouldable by external pressure or abnormal muscular forces. Since the introduction of the 'Back to Sleep' (APP Taskforce on Infant Positioning and SIDS, 1992) campaign there has been an increased incidence of posterior plagiocephaly as a result of positional moulding with the baby favouring

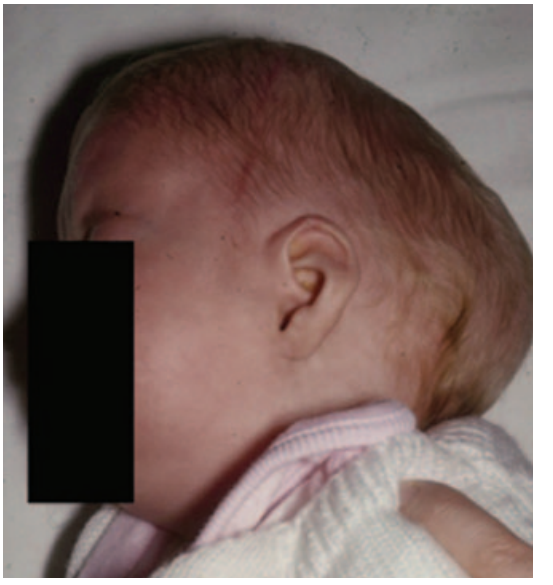


Figure 3. Scaphocephaly.

one side over the other during sleep. Torticollis and, more rarely, spinal deformity or ocular squints can cause DP and should be excluded. True lamdoid synostosis is rare and as the treatment of each condition is different, recognition of DP is extremely important.

The head should be viewed from above with particular reference to its shape and the position of the ears (Huang et al, 1998). In DP and lamdoid synostosis the most striking deformity occurs in the posterior skull rather than in the frontal area, while in frontal plagiocephaly the converse is true. With DP the skull is, in effect, pushed forward on one side leading to forehead prominence and a more anterior ear position on the side of the occipital flattening (*Figure 4*). Conversely, in lambdoid synostosis there is lack

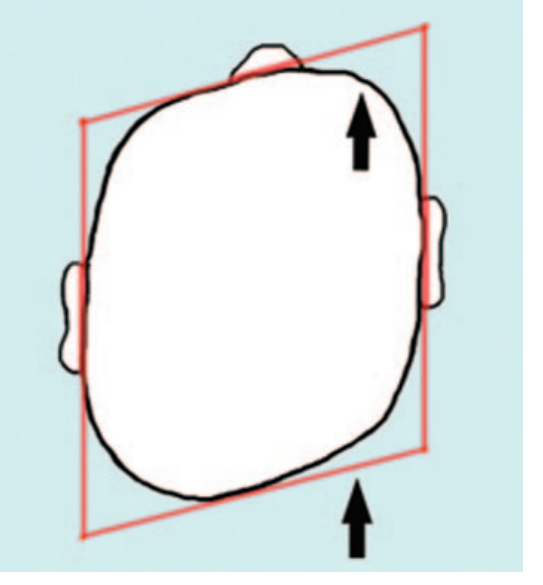


Figure 4. Positional Plagiocephaly.

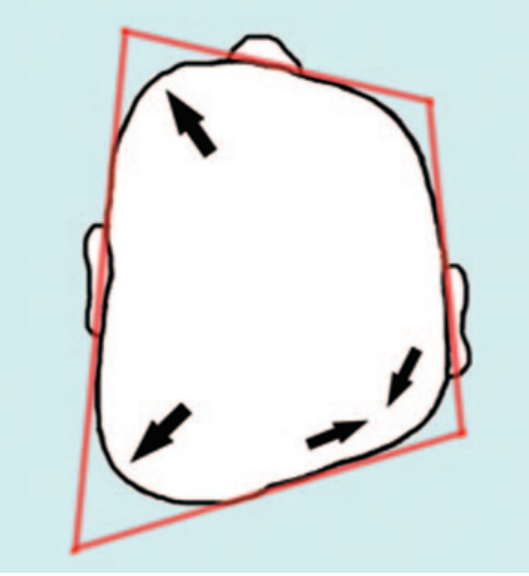


Figure 5. True Lamdoid Synostosis.

of growth leading to apparent pulling back of the ear on the affected side with compensatory forehead prominence occurring on the contraleral side (*Figure 5*). Not all patients conform to these patterns and diagnosis based on clinical examination alone can sometimes be difficult. The presence of a ridged suture on palpation makes the diagnosis of lamdoid synostosis more likely.

The appearance of DP improves as the child develops and is able to sit up reducing the pressure on the flattened occiput, and hair growth will lessen the apparent deformity. Treatment with moulding helmets is controversial and there is currently a lack of good evidence to support their use. They are popular in the US and are being used increasingly in the UK. They are available commercially but not generally available in the NHS, and are currently under review by the National Institute for Clinical Excellence.

TABLE 3.
Members of the multidisciplinary team commonly involved in the management of craniofacial synosotoses

Neurosurgeon
Maxillofacial surgeon
Plastic surgeon
Paediatric anaesthetist
Paediatrician
Intensive care nurse
Clinical nurse specialist
Geneticist
Psychologist
Speech and language therspist
Orthodontist
Paediatric dentist
Ear nose and throat surgeon
Ophthalmologist

Investigations

Plain radiographs may show a sclerotic line and lack of sutural lucency at the site of a fused suture in an infant. 'Copper-beating' of the skull may also be evident in cases of chronically raised ICP. ICP monitoring may be indicated in selected cases where clinical signs of raised ICP are equivocal.

Computerized tomography (CT) not only identifies fused sutures but also defines the skull deformity. Three-dimensional images and more recently the ability to produce accurate models using stereolithography has aided in the surgical planning of the more complex cases.

MANAGEMENT

The National Service Commissioning Advisory Group (Department of Health, 1998) designated four centres in England for the treatment of craniosynostosis (Birmingham, Great Ormond Street, Liverpool and Oxford). A number of non-designated centres provide paediatric craniofacial services and while arrangement for service provision are controversial, there is general agreement that the more complex syndromic cases should be managed in one of the designated centres.

Due to the complex nature of craniofacial anomalies, the expertise of multidisciplinary teams is required (*Table 3*). The problems associated with craniosynosotosis may continue well into adulthood and patients commonly require input from the various members of the team from birth to maturity and beyond.

Neonatal

Severe midface retrusion associated with airway compromise may require urgent management in the form of nasopharyngeal intubation or tracheostomy. On rare occasions ocular proptosis needs urgent correction if inability of the eyelids to close around the cornea threatens sight. Feeding difficulties may need to be addressed, otherwise little physical intervention is required at this stage. Psychological support for the family is, however, crucial and ongoing. Genetic counselling should be offered to parents planning further pregnancies.

Infant

The two main indications for surgical correction of craniosynostosis are to prevent the sequelae

of raised ICP (namely impairment of neurological development, optic atrophy and hydrocephalus) and to improve aesthetics to prevent the long-term psychological impact of altered appearance.

The timing of surgery needs to balance the dangers of operating in small infants and the risk of post-surgical re-stenosis with early surgery, against the increasing risk of raised ICP and progressive deformity if surgery is delayed (Renier et al, 2000). Corrective surgery is generally carried out at 6 months for isolated scaphocephaly, and 12–18 months for other forms of craniosynostosis.

The principles of surgery consist of:

- Excision of the affected suture, leaving a bony gap which allows for future unrestricted expansion of the bones on either side as the underlying brain grows.
- Remodelling of the deformed area of skull into a normal shape
- Moving the bones into a normal position, thereby restoring normal volume and shape as a basis for future growth.

These general principles are adapted to each specific condition.

Scaphocephaly is usually treated with wide excision of the sagittal suture and out-fracturing of the temporal and parietal bones to increase the width of the skull. More extensive remodelling procedures are described and these may be used in early surgery. However if diagnosis is delayed and/or surgery is carried out late these more extensive procedures are usually required.

In cases of craniosynostosis involving abnormal forehead shape (namely trigonocephaly, plagiocephaly and brachiocephaly) removal, remodelling and replacement of both frontal bones is required. This includes bone in the supraorbital region and orbital roof that is removed as a separate segment (*Figure 6*). This is known as fronto-orbital advancement and remodelling.

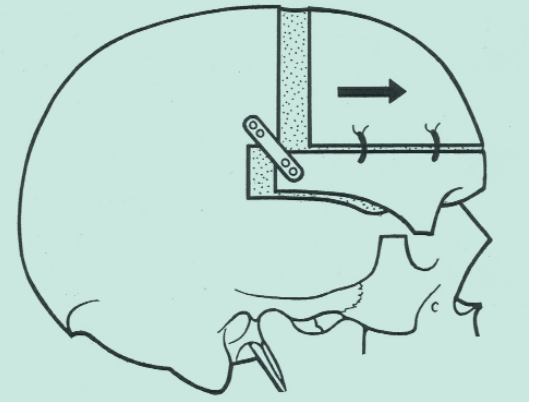


Figure 6. Fronto-orbital advancement to correct anterior cranial and forehead deformity.

Childhood

Throughout childhood general physical and mental development should be monitored. Speech and language development is extremely important and therapists have a large role in the MDT. Paediatric dental and orthodontic care is commenced at this stage. Lack of volume in the midface may lead to Eustachian tube and middle ear dysfunction resulting in deafness.

Craniofacial surgery to address midface deformity in syndromic patients can be considered at this stage. This may be indicated for psychological reasons or functional airway problems such as obstructive sleep apnoea. Various midface osteotomies can be performed to correct midface retrusion, exorbitism and hypertelorism. More recently, distraction osteogenesis is increasingly replacing conventional surgery for correction of midface deformity (*Figures 7–9*). Psychological counselling for both patients and parents is of enormous value in helping families cope with the major changes in appearance associated with midface correction.

Adolescence/Adulthood

Midface surgery does not always fully address misalignment of the upper and lower jaws. Continued growth of the mandible in association with poor growth of the maxilla following midface surgery may result in a deterioration of dental occlusion requiring a combination of

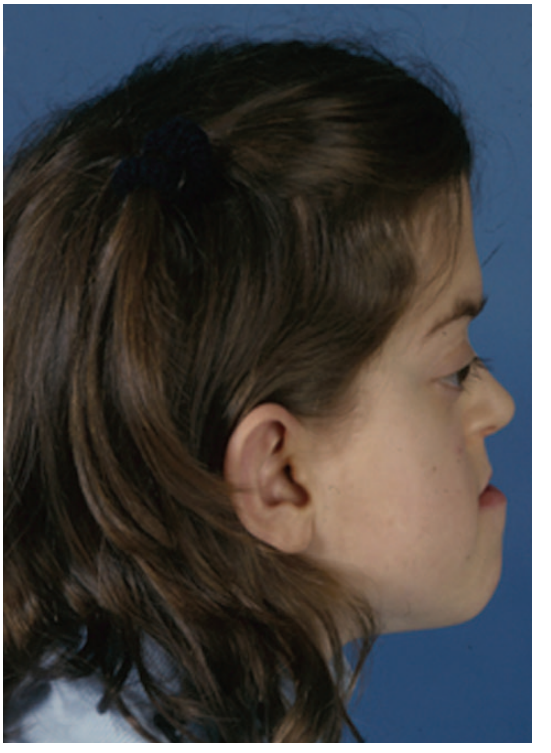


Figure 7. Crouzon's syndrome in a nine year old girl prior to osteogenesis distraction.

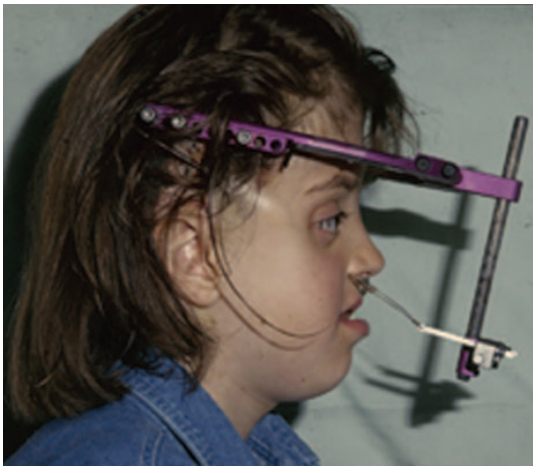


Figure 8. External distractor in place.

orthodontic tooth alignment and jaw (orthognathic) surgery. This is usually performed at 17–18 years of age when skeletal growth has ceased. Any residual nasal deformity is usually treated at this age, and there may be a requirement for revision procedures to enhance the results of previous surgical interventions.

Genetic counselling regarding the risk of the condition occurring in future offspring should be offered to patients.

CONCLUSIONS

Craniofacial synostosis is a relatively rare but potentially serious congenital condition that may occur in conjunction with other recognized abnormalities as part of a syndrome. The complex nature of the condition should be recognized promptly and onward referral to dedicated units that can provide the necessary expertise within an experienced multidisciplinary team is vital.

The complications of raised intracranial pressure and long-term psychological morbidity dictate the majority of management decisions. Further advances in molecular biology should aid identification of the cause of craniosynostosis and may lead to non-surgical therapeutic interventions becoming available. Until such time the use of resorbable osteosynthesis plates for fixation and the introduction of endoscopic surgery may go someway to reducing the potentially serious complications inherent in current surgical management. HM

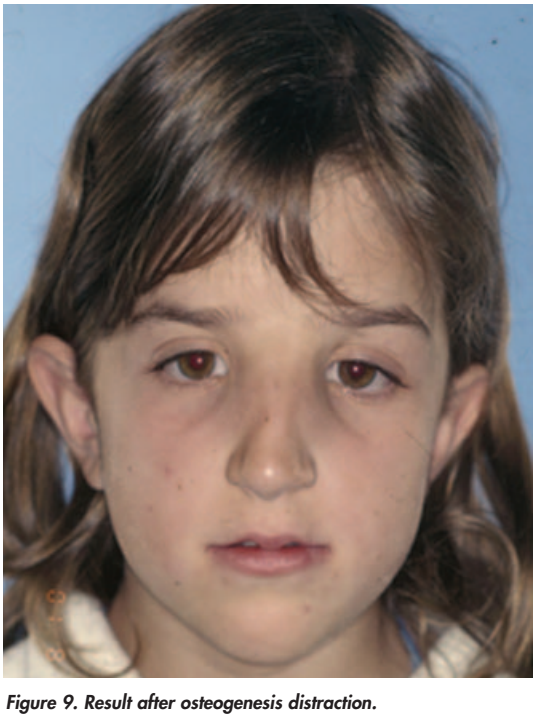


Figure 9. Result after osteogenesis distraction.

Conflict of interest: none

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KEY POINTS

- Craniosynostosis is the premature fusion of one or more craniofacial sutures.
- Craniosynostosis may occur in isolation or as part of a recognized syndrome.
- Craniosynososis is a complex condition that requires care within an experienced multidisciplinary team.
- Surgery aims to reduce the complications associated with raised intracranial pressure and long term psychological morbidity associated with craniofacial deformity.
- Syndromic craniosynostoses appear to be related to abnormalities in the genes coding for fibroblast growth factor receptors TWIST and MSX2.