

## **Craniosynostosis**

### **Diagnosis, Functional Aspects and Surgical Management**

Craniosynostosis may be defined as the premature closure or fusion of the calvarial sutures occurring intra-uterine or shortly after birth. The calvarial sutures are lines of growth lying between the various bones of the skull. While there are a large number of sutures in the vault and base of the skull which can be involved, this discussion will be concentrate on the common ones affecting the calvarial vault. These are six in number namely the **sagittal suture** which runs longitudinally down the midline of the skull between the anterior and posterior fontanels, the **metopic suture** which runs longitudinally from the anterior fontanel to the area between the eyes, the two **coronal sutures** - one on each side running transversely from the anterior fontanel to the area just behind the orbits and the two **lamboid sutures** which run obliquely downwards from the posterior fontanel to the areas behind the ears.

The exact cause of sutures fusing prematurely in isolated instances is as yet unknown. The overall incidence averages out at approximately 1 in 3000 live births.

The cause in syndromic cases has been determined by molecular genetics in a large number of syndromes. The majority are related to abnormalities of the Fibroblast Growth Factor Receptors resulting in upregulation or increased activity leading to premature fusion of the involved sutures.

Under normal circumstances the growth of the individual skull bones occurs at right angles to the growing sutures. If a suture fuses prematurely the skull fails to grow at right angles to the involved suture(s). Importantly normal adjacent sutures respond to this growth restriction by increasing their activity and there is thus generally a compensatory growth in a direction parallel to the involved suture.

It is the failure of normal growth at right angles to the suture and the excessive compensatory growth at other sutures which gives rise to the classical skull shapes associated with the craniosynostoses. The typical shapes can be used clinically to predict the site of the abnormal suture. Because skull growth is most rapid during the first two years of life and continues to adulthood the presence of an abnormal or non functioning suture gives rise to a progressive deformity which is most rapidly progressive in infancy but has the potential to progress until growth is completed in adulthood. As mentioned there are sutures in the base of the skull as well which may be affected or, more frequently in the isolated synostoses, may grow compensatorially causing distortion of the lower orbits and the face at a later stage.

This has led to the inclusion of the term "craniofacial synostosis" in the vocabulary of these conditions.

The synostotic conditions are usually divided into the Isolated Craniosynostoses which are generally not genetically determined ( as far as yet known) and the Craniofacial Synostosis Syndromes (further divided into Craniofacial Dysostosis and the Acrocephalosyndactyly syndromes.) which have a definite genetically defined basis. In the syndromic cases it is the typical craniofacial appearance in combination with the associated midface and limb anomalies which allow clinical classification into the well recognized syndromes. Molecular genetic analysis now can confirm the diagnosis.

The accompanying table documents the various diagnostic features of the individual synostoses according to the individual areas of the craniofacial skeleton as well as the general morphology. The key clinical features of the major syndromes are listed below.

#### **Crouzons Syndrome (Craniofacial Dysostosis)**

Described in 1912. 1;100 000 births. Autosomal dominant with full penetration and variable expression. 1/3 -1/2 cases are sporadic Craniostenosis, exorbitism and midface retrusion without acral deformities. Brachycephaly most common but can be any other. Exorbitism- severity a function of number of involved sutures and degree of maxillary hypoplasia. Variety of other ophthalmological complications can be present. Severe midface retrusion can result in nasopharangeal obstruction. Class III malocclusion with anterior open bit occurs. Dental crowding with high arched constricted maxilla common. Occasional cleft palate. Mental

retardation thought to be indicator of underlying embryological defect rather than raised ICP (which does develop in neglected cases)

### **■ Apert's Syndrome (Acrocephalosyndactyly Type I)**

Described in 1906. Between 1;100 000 and 1;317 000 live births. Autosomal dominant with full penetrance and variable expressivity Craniostenosis, exorbitism, midface retrusion and Complex syndactyly of hands and feet. Brachy- and turri-cephally common. Exorbitism more severe than with Crouzon's Hypertelorism and antimongoloid slants Class III aloccclusion, anterior open bite. Crowded V-shaped arches 1/3 have soft palate cleft. Hand and feet - complex syndactyly of digits 2,3,4. 1,5 conjoined or separate

Three types

I - Obstetrician's hand - most common 2,3,4 joined 1,5 separate

II - Mitten hand 2,3,4,5 joined 1 separate

III - Hoof Hand Rarest Most severe. All joined

Axial skeletal defects common

Mental retardation common.

### **■ Saethre- Chotzen Syndrome (Acrocephalysyndactyly III)**

Described 1931 & 1932 Autosomal dominant full penetrance, variable expressivity (often mild) Craniostenosis, low frontal hairline, eyelid ptosis, deviated nasal septum, lacrimal duct stenosis fingerprint abnormalities and brachydactyly. Usually acrocephalic Midfacial - minimal midfacial retrusion. No class III malocclusion, but anterior open bite common Strabismus is common. Simple incomplete syndactyly between 2,3 digits hands and feet. Not obligatory for diagnosis. Short stature and spinal defects occur. Radio-ulnar synostosis. Most studies intellect normal.

### **■ Pfeiffer's Syndrome (Acrocephalosyndactyly V)**

Described 1964. Autosomal dominant full penetrance, variable expressivity. Craniostenosis with variable maxillary protrusion. All features of Apert's but milder. Partial soft tissue syndactyly usually 2,3 occasionally 3,4. Broad thumbs and great toes are pathognomonic. Turribrachycephaly. Variable midface abnormalities as for Crouzon's. Variety of axial skeletal deformities can occur. Most have normal/low average intellect. Vertebral fusion abnormalities occur in 30% of Pfeiffers, 38% of Crouzon's and 71% of Apert's. Cervical Xray of these children should thus be routine pre-op due to the implication of fusions on intra-operative airway control.

## **■ Form Vs Function**

In the vast majority of cases of single suture craniosynostosis the compensatory growth of the normal sutures is generally sufficient to allow the developing brain to grow without causing raised pressure. However, in a certain percentage of cases (somewhere between 10-15% of single suture synostoses and possibly over 50% of syndromic cases) the restriction is such that the pressure within the skull rises (so-called raised intracranial pressure) and this may cause functional problems in terms of development if left untreated. In addition there is evidence accumulating that there may be local pressure effects, cerebral perfusion deficits and changes in brain metabolism underlying the involved sutures which may be corrected by surgery. The overall benefit of craniofacial surgery in terms of form and function in the craniofacial dysostoses such as Apert's and Crouzon's syndromes, as well as in multiple suture synostosis, is seldom questioned. The role of surgery in isolated single suture craniosynostosis is less well defined. It is often misperceived as being merely for 'cosmetic' benefit alone. The question of "Form vs. Function" in these

patients arises. Direct comparison of reports in the literature is difficult due to generally low numbers and differences in systematic data presented as well as differences in definitions. It has been documented however that whilst the benefit in terms of "Form" is substantial, single suture craniosynostosis is a complex clinical entity with more at stake than purely the "cosmetic" appearance of the child. Psycho-social development and function (including potential 'Learning Disability' and 'Behavioural Deficits') as well as the risk of 'subclinical' raised intracranial pressure are coming to the fore. The possibility of a reversible underlying cerebral hypoperfusion defect is becoming more evident. These factors apply even more so in the syndromic cases.

### Surgical Indications

There are thus a number of reasons why surgery is generally indicated in craniosynostosis:-

- 1) For the treatment of an established deformity.
- 2) To attempt to prevent the significant progression of a developing deformity.
- 3) To relieve established raised intracranial pressure
- 4) To decrease the risk of developing raised intracranial pressure or other functional pressure related/cerebral perfusion effects.
- 5) To protect the eyes

The type of surgery required is frequently determined by the degree of the deformity and the underlying sutures involved.

Each case needs to be individually assessed in terms of functional indications and indications in terms of established or progressive deformity. Whilst the surgery required is fairly extensive, if performed in an established unit it is regarded as safe with acceptable risk in terms of the benefits achieved. These "benefits versus risks" are best discussed by the individual surgeons based on the individual features and indications in each child.

It has been established that it is safe to remove segments of bone in the area of the involved sutures, to reshape segments of bone and change their position with predictable survival of the bone fragments and with reliable healing of the bone and soft tissue.

This discussion will be limited to the upper craniofacial skeleton with an emphasis on the general concepts and techniques. Specific details of approaches to syndromic conditions are covered in a separate talk by another speaker

### Surgical options

Essentially the principals involve one or more of the following approaches

Resect

Release

Remodel

"Redirect"

### **Types of Surgery**

## Strip Craniectomy

## FOAR (Fronto-Orbital Advancement and Remodelling)

## Lateral Releases

## Posterior Remodelling

## Posterior Release (Staging)

## Total Calvarial Remodelling

Sagittal suture synostosis is traditionally treated in the early phases by removal of some of the bone overlying the abnormal midline suture, the so called "strip craniectomy". This may be combined with various other ancillary procedures such as "plication" or tightening of the bones laterally to encourage the development of a broader, shorter head with growth. In established cases in older children more extensive reshaping of the skull may be required.

In the anterior synostoses (namely metopic, bicoronal and unicoronal synostosis) the aim is to recreate a symmetrical forehead and orbital rim and to "release" the area of the involved suture and there by allow more normal growth of the skull. The mainstay of this type of surgery is the "fronto orbital advancement and remodelling" procedure whereby the upper aspects of the orbits are freed and advanced unilaterally or bilaterally as appropriate and a more symmetrical forehead is reshaped from the existing or adjacent bone.

As mentioned lamboid sutures synostosis is uncommon and the surgery is aimed at preventing progressive deformity by releasing the suture and remodelling the posterior skull. This is referred to as Posterior Remodelling

In certain cases particularly the severe syndromics in infancy, the rate of progression of the calvarial deformity particularly in the anterior skull as well as the occurrence of raised pressure in total synostosis makes early intervention unavoidable. To avoid the risk of recurrent surgery being required anteriorly an initial posterior release can be performed to increase intracranial volume and decrease the anterior "driving force" with a FOAR performed later. This staging technique can also be employed in late presenting cases where bone thinning may be such that anterior stability may be difficult to achieve if a primary FOAR is performed. Initial posterior release allows the anterior bone to thicken after pressure is released allowing a stage FOAR.

Irrelevant of the sutures involved if there is evidence of secondarily raised intracranial pressure then more extensive skull vault surgery may be required in order to expand the volume of the skull and thus relieve the pressure. This can be performed laterally if anterior contour is acceptable, or combined with a repeat FOAR or Total remodelling inf necessary.

It is important to note, once again, that while this is major surgery it is generally classed as safe if performed by a multidisciplinary team i.e. a neurosurgeon with a plastic surgeon and/or a maxillofacial surgeon in an established unit.

Finally, it is important to note that because the underlying defect in the growth centre is as yet uncertain the surgery does not necessarily normalise the growth in all cases and there is a tendency in a certain percentage of cases for the condition to recur, or for progression to occur in other areas particularly the face, thus further operation or re-operations may be required in some cases. The predictive factors for this will depend on the sutures involved and the extent and rate of progression of the deformity. Once again it is impossible to give blanket guidelines for this and it should be discussed in detail between the patient/parents and the treating surgeons.

The potential consequences of late referral in terms of functional risks and surgical management are not insignificant . Based on the available evidence a plea is made for early referral for assessment and regular follow-up by an experienced multidisciplinary team. This should minimize the risk of "inappropriate

conservative treatment" as well as minimizing the morbidity and optimizing the results in cases where surgery is appropriate.

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