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Headlines

Craniofacial Support

Glossary of Terms associated with Craniosynostosis

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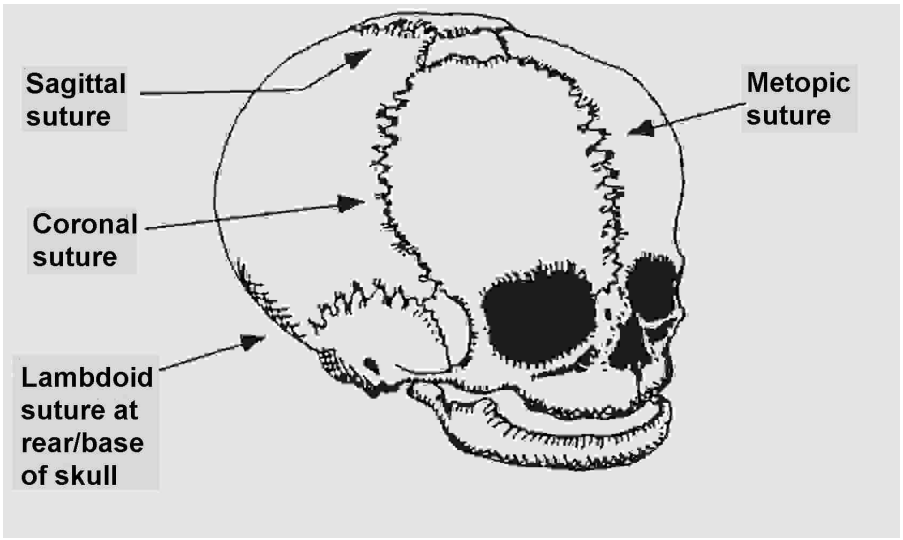
Turricephaly 'tower shaped head' caused by multiple or late fused Coronal sutures.

Unicoronal Synostosis where only one of the Coronal sutures is prematurely fused. There is a failure of adequate front to back growth on the side of the fused suture, a flattening of the brow and elevation of the upper part of the eye and eyebrow area and the ear tends to be pulled forward. There is frequently compensatory growth in the cheek on the fused side. This is traditionally called Plagiocephaly.



The information in this leaflet has been taken from various sources. It is provided as a layman's introduction to the terms you may come across. For more detailed information, please consult other leaflets (see back page) and other Medical sources. The advice of one of the Supra Regional Hospitals or other Medical Practitioner should be sought in all individual cases. It should be noted that procedures at different Hospitals may vary.

This list is not exhaustive and other terms may be added in the future. (December 2001)

Acanthosis Nigricans a pre-malignany skin disorder with hyperkeratosis and hyperpigmentation.

Acrocephaly 'topmost skull' caused by fused multiple sutures.

Allele one of two or more alternative forms of a gene, only one of which can be present in a chromosome.

Anterior front

Apert Syndrome first described in the early 1900's, predominantly affects the head, face and limbs. It may also affect brain development. Common features:

- regressed mid-face and shallow eye sockets, this may result in a small or constricted airway and the appearance of a large lower jaw. Seen from the side the face has a concave appearance and the shallow eye sockets result in prominent eyeballs. The arrangement of teeth is also affected.
- the abnormal skull shape may result in raised intracranial pressure and require surgery to protect the restricted brain.
- fusion of fingers and toes, this can vary from fusion of the centre three digits to complete fusion of the bones and nails of all five digits.
- limited movement of the shoulders and elbows can occur.
- the skull in Apert's patients tends to be short from front to back referred to descriptively as Brachycephaly.

Autosome any chromosome that is not a sex chromosome and occurs in pairs in diploid cells, **Autosomal** (adjective)**.**

Bicoronal Synostosis premature fusion of both Coronal sutures causing inadequate growth at the front and back on both sides of the skull. The skull becomes excessively wide and short front to back. This is traditionally called Brachycephaly.

Brachycephaly a wide skull which is short front to back. Caused by premature fusion of both Coronal sutures.

Callotasis the process of stretching the callus (tissue) that forms between the ends of a bone that has been divided. It is achieved by means of an external adjustable frame attached to the bone in the procedure for mid-face advancement. The elongated callus consolidates to form new bone.

Single Suture Craniosynostosis In the vast majority of cases the compensatory growth of the normal sutures is generally sufficient to allow the developing brain to grow without problem. However, in some cases the restriction is such that the pressure within the skull rises (see Intracranial Pressure).

There are a number of reasons why surgery may be indicated

- for the treatment of an established disfigurement
- to attempt to prevent the progression of a developing disfigurement
- to relieve raised Intracranial Pressure
- to decrease the risk of developing raised Intracranial Pressure or other functional pressure-related affects.

Skull the skeleton of the head and face, which is made up of 22 bones. It can be divided into the cranium, which encloses the brain, and the face (including the lower jaw).

Strip Craniectomy removal of some of the bone overlying a fused suture, eg the Sagittal suture.

Supra Regional Craniofacial Units the four NHS Hospitals in England & Wales which are funded by the National Specialist Commissioning Advisory Group (NSCAG) to provide Craniofacial Surgery and associated Care. These are

- Alder Hey Hospital, Liverpool
- Birmingham Children's Hospital
- Radcliffe Infirmary, Oxford and
- Great Ormond Street Hospital, London.

Suture (in anatomy) a type of immovable joint, found particularly in the skull, that is characterized by a minimal amount of connective tissue between the two bones. The cranial sutures include the coronal suture, between the frontal and parietal bones; the Lambdoidal suture, between the parietal and occipital bones; and the Sagittal suture, between the two parietal bones.

Syndactyly congenital webbing of the fingers. Adjacent fingers are joined along part or all of their length. They may be joined only by skin, or the bones of the fingers may be joined.

Trigonocephaly 'triangle-shaped head' has the vault of the skull sharply angled just in front of the ears, giving the skull a triangular shape caused by premature closure of the Metopic suture.

Plagiocephaly 'skew or oblique head' traditionally reserved for craniosynostoses involving either one Coronal or one Lambdoidal suture. It is sometimes used to describe other conditions not involving Craniosynostosis which cause a similar shaped head, (see Deformational Plagiocephaly)

Plication tightening of the bones of the skull laterally to encourage development of a broader, shorter head with growth.

Posterior rear

Proptosis forward displacement of the eyes, generally caused by the eye sockets being too small.

Ptosis (blepharoptosis) drooping of the upper eyelid.

Recessive describing a gene (or its corresponding characteristic) whose effect is shown in the individual only when its allele is the same, i.e. when two such alleles are present (the double recessive condition). Many hereditary diseases are due to the presence of a defective gene as a double recessive. They are said to show autosomal recessive inheritance

Sagittal Suture the suture which runs longitudinally down the midline of the skull between the anterior and posterior fontanels.

Sagittal Synostosis premature fusion of the Sagittal suture. This causes lack of growth across the head and excessive growth front to back. This is traditionally called Scaphocephaly.

Saethre-Chotzen Syndrome was described in in the early 1930's, its diagnosis is difficult.
Common features:

- abnormal shaped head, it is usually the coronal sutures that are affected. The abnormal skull shape may result in raised intracranial pressure and require surgery to protect the restricted brain. Usually one or both of the Coronal sutures are affected.
- droopy eyelids
- short fingers, joining of the fingers, and broad toes rarely causing any problems.
- fusion of bones in the neck rarely causing problems but care must be taken during anaesthetics.

Scaphocephaly 'boat-shaped head' an abnormally long and narrow skull due to premature closure of the Sagittal suture along the top of the skull.

Callus the tissue formed between bone ends when a fracture is healing. It initially consists of bloodclot, small blood vessels and connective tissue, which develops into cartilage and then calcifies to form bone. Callus formation is an essential part of the process of healthy union in a fractured bone.

Calvarial Sutures are lines of growth lying between the various bones of the skull.

Cephalic Index a measure of the shape of a skull, the ratio of the greatest breadth, multiplied by 100, to the greatest length of the skull.

Coronal Suture the sutures, one on each side, running transversely from the anterior fontanel to the area just behind the eyes.

Cranial Index the same measurement as the Cephalic Index, but measured on the bare skull.

Craniosynostosis the premature closure or fusion of the Calvarial sutures occurring in the uterus or after birth.

Crouzon Syndrome was originally described in 1912, and causes a range of problems of variable severity, from mild facial symptoms of a mainly cosmetic nature to symptoms affecting breathing, feeding, vision and brain development and predominantly affects the appearance of the head and face.
Common features:

- regressed mid-face and shallow eye sockets, which may be present at birth or become more evident later. Seen from the side, the face has a concave appearance and the shallow eye sockets result in prominent eyeballs, arrangement of teeth is also affected.
- the abnormal skull shape may result in raised intracranial pressure and require surgery to protect the restricted brain.
- It may involve any combination of cranial sutures, most commonly including the coronal and Sagittal sutures.
- The skull can be referred to descriptively as Brachycephaly, Scaphocephaly or Turricephaly.

Deformational Plagiocephaly a 'skew or oblique head' with causes not associated with Craniosynostosis (also called **Occipital Plagiocephaly**.
Dentition type, number and arrangement of teeth.

Diploid describing cells, nuclei, or organisms in which each chromosome except the Y sex chromosome is represented twice.

Distraction increasing the distance between two points. In mid-face advancement procedures callus (tissue) can be stretched by increasing the distance between pins attached to the bone (see callotasis).

Dolichocephaly having a relatively 'long skull', generally due to the premature fusion of the Sagittal suture.

Dominant (in genetics) describing a gene (or its corresponding characteristic) whose effect is shown in the individual whether its allele is the same or different. If the allele is different it is described as recessive and its effect is masked. In genetic diseases showing autosomal dominant inheritance, the defective gene is dominant and will therefore be inherited by 50% of the offspring (of either sex) of a person with the disease. It will always be expressed in these offspring (since the normal allele inherited from the unaffected parent is recessive)

Hydrocephalus an abnormal increase in the amount of cerebrospinal fluid within the ventricles of the brain. In association with Craniosynostosis this may lead to raised Intracranial Pressure.

Hyperkeratosis thickening of the outer layer of skin.

Hyperpigmentation areas of the skin become darker than the surrounding skin.

Hypotelorism the eyes being closer together than normal

Intracranial Pressure the pressure within the skull. Raised Intracranial pressure may lead to functional and developmental problems, if left untreated

Lambdoid Sutures the sutures which run obliquely downwards from the posterior fontanel to the areas behind the ears.

Lambdoid Synostosis premature fusion of a Lambdoid suture causes the opposite back of the head to bulge

Metopic Suture the suture which runs longitudinally from the anterior fontanel to the area between the eyes.

Metopic Synostosis premature fusion of the Metopic suture with failure of adequate transverse growth in the forehead and compensatory growth across and at the back of the head. This gives a central ridge of the forehead with a pinched look above the brows. The eyes tend to be close together (Hypotelorism). This is traditionally called Trigonocephaly.

Mid-face Advancement A procedure utilising an adjustable frame which allows the mid part of the face to be moved forward. This is done to improve airways, eye sockets etc (see **Callotasis** and **Distraction**).

Muenke Syndrome was identified in 1996 and is generally diagnosed as the result of a specific genetic blood test.
Common features

- distortion in skull shape by one or both of the Coronal sutures being fused
- fingers may sometimes be short, slightly crooked or webbed without affect on their function
- hearing loss and mild to moderate learning difficulties

Occipital Plagiocephaly a 'skew or oblique head' with causes not associated with Craniosynostosis (also called **Deformational Plagiocephaly**).

Occiput the back of the head.

Orbit eye socket.

Oxycephaly 'sharp skull' caused by fused multiple sutures.

Parietal Bones the pair of bones forming the top and sides of the cranium (see Skull).

Pfeiffer Syndrome was originally described in 1964 and has a range of symptoms varying in severity, from mainly cosmetic facial symptoms to those affecting breathing, feeding, vision and brain development. It predominantly affects the appearance of the head and face. The hands and feet are also involved and may require surgical care.
Common features

- a regressed mid-face and shallow eye sockets, resulting in prominent eyeballs, this may become more evident as the child grows.
- the hands and feet are involved to a variable degree. The thumbs and big toes are broad and deviated towards the midline. There may be mild soft tissue webbing or syndactyly, between the second, third and fourth digits of either or both hands and feet. The digits may be short and misshapen, with consequences for an adequate grip (hand) or footwear (feet).