

CRANIOSYNOSTOSIS

Craniosynostosis is a condition that children are either born with or develop. It literally means fusion of the skull bones and may occur singularly or as part of a genetic syndrome.

At birth, the human skull is not a solid mass of bone. Rather, soft spots or sutures separate the seven bones that comprise the skull. These separations allow for brain growth. When one or more of these sutures close prematurely, the result is called a craniosynostosis condition. The deformity that results depends on which suture(s) is involved.

What actually happens is that as a baby grows, the brain increases rapidly in size. When the suture(s) fuse, there is no room for the brain to develop at that area. This growth inhibition is compensated for by overgrowth in another area resulting in an abnormally shaped skull.

Types of Craniosynostosis:

(a) *Isolated* craniosynostosis is non-inherited and usually involves only one suture.

- *Sagittal synostosis* refers to premature closing of the midline suture that runs from front to back of the skull. This results in an elongated and narrow skull shape called *scaphocephaly*.

- *Metopic synostosis* refers to the early closing of the forehead suture. This causes a triangular forehead with closely placed eyes or a deformity called *trigonocephaly*.
- *Unilateral coronal synostosis*, closure of the coronal suture on one side, produces flattening of the forehead and the brow on the affected side. The opposite side forehead therefore becomes excessively prominent. This cranial asymmetry is called *plagiocephaly*.

(b) *Craniofacial Synostosis Syndromes* are comprised of premature closing of *multiple* sutures and are associated with abnormalities of the face and extremities. These syndromes tend to be inherited.

- Crouzon syndrome, one of the most common craniofacial conditions, results from multiple premature suture closings. Abnormal skull shape, bulging eyes and underdevelopment of mid-face and upper jaw characterize this syndrome.
- Apert syndrome (bilateral coronal suture synostosis) is characterized by exorbitism (bulging eyes) and mid-face and jaw underdevelopment. In addition, Apert syndrome presents with fusion of fingers and toes and severe acne.
- Pfeiffer Syndrome is characterized by a short, pointed head. It is similar to Apert but with milder hand deformities.
- Carpenter Syndrome is extremely rare and involves the sagittal and lambdoid sutures and possibly the coronal suture(s). Head shape is dramatically uneven. It is also characterized by: congenital heart disease, obesity, extra fingers or toes, genital abnormalities and short stature.

- Saethre-Chotzen Syndrome is also extremely rare. This syndrome is characterized by a small head, mildly fused fingers and/or toes with abnormalities of the skin in these areas. Short stature, often mild to moderate mental retardation and facial abnormalities may occur.

Treatment

Isolated craniosynostoses: Surgical treatment is recommended after 6 months – earlier surgery is not usually required since there is no increased cranial pressure. The surgical procedure, *fronto-orbital advancement with cranial vault remodeling* releases the fused suture and reshapes the skull.

Craniofacial syndromes: A craniofacial treatment team needs to do a comprehensive evaluation and form a treatment plan based on the age of the patient.

- Newborns – attention is paid to performance of vital functions (i.e. breathing, feeding, intracranial pressure and vision). If there is a significant airway problem then a tracheostomy may be required. Acute feeding problems may require a gastrostomy (stomach tube). Hydrocephalus (fluid on the brain) is treated via a ventriculoperitoneal (VP) shunt. Severe pressure on the brain requires removal of a segment of bone to enlarge the skull and relieve the pressure.
- 6-12 months – Fronto-orbital advancement with cranial vault reshaping is most often recommended. The brow and forehead are advanced, the skull reshaped and enlarged to relieve pressure and

protect the optic nerve. This procedure increases the size of the eye sockets (orbits) to reduce tendency to exorbitism (bulging eyes).

- 3-8 years old – It is during this developmental span that mid-face advancement is undertaken to correct the severe underdevelopment of the mid-face and upper jaw (responsible for a sunken facial appearance and under bite). Mid-face advancement should improve the airway, correct the malocclusion (under bite) and improve the contour of the mid-face.
- 8-18 years old – During the adolescent growth spurt, the mandible (lower jaw) undergoes significant growth so that the lower jaw and teeth may wind up in front of the upper highlighting the need for a second mid-face advancement procedure, or lower jaw surgery, or both.

Associated Conditions

Increased intracranial pressure: When the skull does not expand sufficiently to accommodate the rapidly growing brain, possible brain damage and/or visual loss may occur.

Hydrocephalus: Excess fluid in and around the brain is not uncommon in craniofacial synostoses syndromes. Like intracranial pressure, it may require treatment (placing a shunt to divert excess fluid into the abdomen) to prevent brain damage.