

Defining Craniosynostosis

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How is Craniosynostosis classified?

CS is classified in several different ways. Primary CS can be sporadic or part of a syndrome and is thought to be caused by

an abnormal osteoblastic proliferation during osteogenesis. Secondary causes of CS can be wide ranging and include rickets, hypophosphatemia, hyperthyroidism, overshunted hydrocephalus, and positional among others.

CS is also classified as Simple (non syndromic) or Complex (syndromic). Simple cases of CS usually will have only one fused suture while complex cases have multiple fused sutures and have typical phenotypes (Crouzon, Apert, Pfeiffer, Saethre-Chotzen). These can be sporadic or are often inherited on an autosomal dominant fashion.

Identifying Craniosynostosis

- Sutures are fused at birth and may have a ridge.
- Normal skull grows perpendicular to the sutures and can bulge.
- Bones involving the suture flatten.
- An early closed fontanelle doesn't imply CS as long as the head circumference (HC) is normal and the child develops adequately. CS infants never jump HC curves.

Diagnosing specific types of CS

Simple non syndromic infants don't look phenotypically syndromic. They just have abnormal skull shapes:

- **Metopic:** Trigenocephaly (narrow forehead with midline ridge and hypotelorism)
- **Sagittal:** Dolicocephaly or Scaphocephaly: long AP and narrow lateral axis, occipital cupping.
- **Unicoronal:** anterior plagiocephaly; one forehead side is flat. One eye may be higher than the other.
- **Bicoronal:** Usually in syndromic.
- **Brachicephaly:** broad and tall forehead; narrow AP axis.

- **Lambdoid:** posterior plagiocephaly. This may be the most difficult to identify since it can be confused with positional or deformational plagiocephaly.

Four characteristics of each type that help differentiate between them are:

- **Lambdoid:** Rhomboid head shape when viewed from above; suture has a ridge; ipsilateral ear is displaced posteriorly; contralateral occipital/parietal bone is bulged.

Craniosynostosis (CS) is the in-utero fusion of skull sutures (coronal, metopic, sagittal and/ or lambdoid) that prevents normal skull growth and shape.

CS is most easily defined by an abnormal head shape in newborns, having bone flattening, bulging and/or ridging. These infants should be evaluated promptly by a pediatric neurosurgeon.

- **Positional:** ipsilateral ear is displaced anteriorly; torticollis may be present; persistent positioning on affected side; no contralateral bone bulging.

Complex syndromic are hard to miss. They have obvious cranio-facial phenotypes. Common findings are proptosis, brachicephaly, midface hypoplasia. Apert's have syndactyly and mental retardation is common.

Testing prior to referring the patient

The diagnosis is clinical and imaging is reserved mainly for surgery planning by the neurosurgeon in syndromic cases who may have bony defects, hydrocephalus or Chiari malformations therefore, testing prior to referral is unnecessary.

Management and Surgical treatment of CS

Observation by a neurosurgeon is recommended for the first months of life if there is doubt about CS as a diagnosis. If diagnosis is clear, surgical management is typically delayed until the baby is around 4-9 months old, depending on the type of CS. This allows for blood volume to increase and for better bone remodeling during the first year of life.

Surgery has two main purposes, cosmetic and preventive. The first prevents negative psychosocial issues. The later refers to the known risk of developing increased intracranial pressure (ICP) with its consequences like delayed neurocognition and visual deterioration.

Surgical options are cranial vault reconstructions (author's preference), strip craniectomies with or without endoscopic assistance and subsequent use of head helmets. These helmets are worn for 1-6 months and up to 4 sizes may be needed.

CS prognosis

Cosmetic changes are impressive even in the most complex cases; some syndromic patients require multiple surgeries during their first 2 decades of life.

It appears that even simple CS cases have mildly lower neurocognitive scores compared to non-CS subjects preoperatively. These features are not improved after surgery but the deterioration is prevented. The incidence of increased ICP is about 45% in syndromic cases. The incidence is significantly lower in non-syndromic cases but its not known.

Following CS patients

Suspected CS cases should always be referred to pediatric neurosurgery for evaluation. In the primary care setting, be aware of possible signs of increased ICP: head aches, performance deterioration, visual changes and papilledema.

In syndromic patients, a Chiari malformation can cause lower cranial nerve deficits, syringomyelia and head aches. Hydrocephalus should be suspected as well with signs of increased ICP.