Craniofacial Anomalies

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KEYWORDS

• Craniosynostosis • FGFR mutations • Scaphocephaly • Dolichocephaly • Trigonocephaly • Brachycephaly • Plagiocephaly • Minimally invasive surgery

KEY POINTS

- Craniofacial anomalies are common (1:2000 for isolated suture synostosis) but syndromic craniosynostosis is relatively rare.
- FGFR mutations underlie most syndromic craniosynostosis, whereas nonsyndromic synostosis frequently involves a variety of genetic and environmental risk factors.
- Single-suture synostosis is often an isolated finding, and intracranial hypertension, developmental delays, and strabismus, though less likely in isolated synostosis, are more frequent in multisuture and syndromic forms of craniosynostosis.
- Minimally invasive approaches are most successful when done between ages 3 and 6 months, and often require many months of molding-helmet therapy postoperatively.
- Surgery after age 8 to 9 months usually requires open cranial vault reconstruction whereby the skull is surgically osteotomized; bone grafts are reshaped and repositioned, then fixated in anatomically improved and often overcorrected positions.

Videos related to pediatric craniofacial surgical procedures (Video 1: Open approach for sagittal synostosis subtotal cranial vault recon without bone grafting and Video 2: Depicts endoscopic-assisted strip craniectomy with wedge craniectomy) accompany this article at http://www.facialplastic.theclinics.com/

OVERVIEW

Craniosynostosis, the premature closure of cranial sutures, may be isolated or nonsyndromic, affecting 1 in 2000 live births[1] (typically single-sutured), or syndromic, affecting 1:30,000 to 1:100,000 live births[2] (frequently multisutured), and primary or secondary. Multifactorial, genetic, and environmental influences may be involved. Regardless of etiology, the fused suture(s) typically cause(s) a restriction in skull growth and subsequent skull characteristics, skull base, and asymmetries and disturbances at the time of facial growth. Three theories have been proposed:

1. Virchow's theory that sutural fusion precedes other events
2. Moss's idea that primacy lies with primary skull-base growth restriction preceding synostosis
3. Others have suggested that brain growth restriction is the primary factor that subsequently leads to premature fusion[3]

It is now generally accepted that underlying mechanical force signaling pathways and cytokines mediate cranial suture patency, and premature fusion is thought to occur secondary to mutations in these genes.[4] Secondary synostosis is evidenced in both microcephalic and overshunted individuals, and in certain hematologic disorders.[5]

The majority or craniofacial growth and development occurs during the first year of life and growth potential, with the brain doubling in size

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by 1 year of life and tripling by 2 years.[6] This rapid growth of the brain and resultant bone growth are determined by complex growth factor pathways regulated by genes that code for fibroblast growth factor receptor (FGFR) and transforming growth factor (TGF)-β, among others, and are at the core of the principles of craniofacial surgery, including the duration and timing of surgical intervention.[7] There are clearly both environmental and genetic factors at play for isolated, single-suture, nonsyndromic forms of synostosis (**Fig. 1**).

PATHOPHYSIOLOGY

Multiple environmental associations have been described, including paternal occupations such as agriculture and forestry, maternal age, exposure to tobacco smoke, and medications, including nitrofurantoin and warfarin use during pregnancy. Malpositioned fetal lie and intrauterine constraint or, at times, metabolic factors can also be associated, including mucopolysaccharidosis, mucolipidosis, rickets, and hyperthyroidosis.[5,8]

MOLECULAR GENETICS

A variety of mutations in transcription derived growth factors, FGFR1,2 and 3 are known to be involved in syndromic craniosynostosis (**Table 1**).[1,7,9,10]

Scaphocephaly

Scaphocephaly (boat-shaped head) (**Fig. 2**) can occur without synostosis; however, it is the most common manifestation of sagittal synostosis in up to 50%[1] of isolated synostosis. Calvarial bone growth is limited perpendicular to the affected sagittal suture, resulting in narrowing of the head transversely, and resultant brain growth anteroposteriorly leads to frontal bossing and/or occipital cupping. Shape may vary depending on the duration or timing of synostosis, whether partial or complete suture involvement, and whether other sutures are involved (**Figs. 3 and 4**).[2,3,11]

Trigonocephaly

Metopic synostosis may result in mild metopic ridging in the midline forehead or a combination of ridging, bitemporal narrowing, and hypotelorism, which together make the forehead appear triangular (**Fig. 5**C, D). The metopic suture fuses as early as age 3 to 6 months and, unlike other cranial sutures, normally disappears; hence, the diagnosis largely depends more on clinical shape and less on computed tomography (CT) findings.[12,13]

Deformational Plagiocephaly

Meaning twisted or slanted, deformational plagiocephaly (DP) refers to flattening of the head.

The most common type of plagiocephaly is positional, namely DP. DP results when the skull is

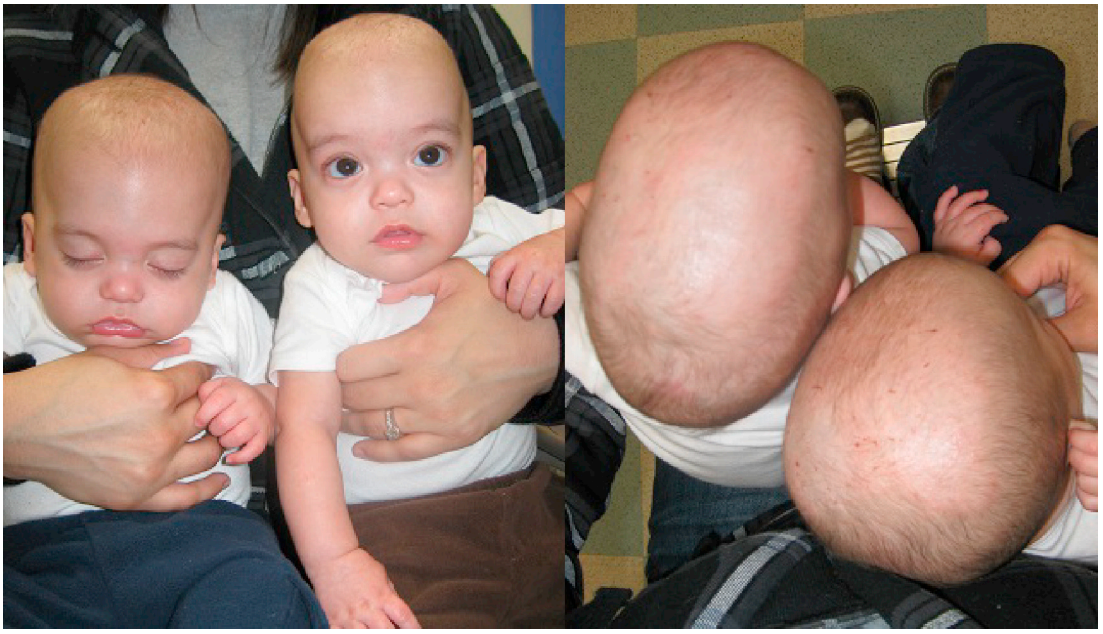

Fig. 1. Four-month-old identical twins, frontal and vertex views. Twin on left lap with sagittal synostosis and scaphocephaly.