Syndromic craniosynostosis

- Craniosynostosis in Saudi Arabia: A Retrospective Analysis of Subtype Patterns, Syndromic Risk, and Postoperative Outcomes
- Identification of a Novel FGFR2 Gene Mutation (c.514_515delinsCT, p.Ala172Leu) in a Chinese Neonate With Apert Syndrome: A Case Report
- Comparison of Postoperative Complications and Reoperation Rates of Le Fort I Osteotomies Using Demineralized Bone Matrix (DBM) or Autogenous Bone Grafts in Patients with Orofacial Clefts and Craniofacial Malformations
- Identification and characterization of short-chain dehydrogenase/reductase 3 (DHRS3) deficiency, a retinoic acid embryopathy of humans
- Cranial Bone Changes Associated With Intracranial Hypertension in Apert Syndrome: Insights for Early Surgical Intervention
- Syndromic Craniosynostosis: The Hidden Burden of Comorbidities on Surgical Outcomes
- Facial asymmetry in syndromic craniosynostosis patients undergoing midface surgery compared to a large general population
- Impact of SMAD6 Variants on Neurodevelopment in Craniosynostosis

Craniosynostosis can occur as an isolated event resulting in nonsyndromic craniosynostosis, or it can occur in conjunction with other anomalies in well-defined patterns that make up clinically recognized syndromes.

Etiology

Syndromic Craniosynostosis Etiology.

Diagnosis

Syndromic Craniosynostosis Diagnosis.

Syndromic Craniosynostosis Complications

Syndromic Craniosynostosis Complications.

Treatment

Patients with syndromic craniosynostoses are much more complicated to care for, requiring a multidisciplinary team to address all of their needs effectively. These are typically genetic in nature and may demonstrate autosomal dominant, autosomal recessive, and X-linked patterns of inheritance. Although busy tertiary care centers will encounter a broad range of syndromes, the more commonly identified craniosynostosis syndromes seen by plastic surgeons include Crouzon, Saethre-Chotzen, Apert, Pfeiffer, and Muenke syndromes. These variably share some common features in

addition to craniosynostosis including exophthalmos, midface hypoplasia, cranial base anomalies, abnormal facies, and limb anomalies. In fact, the craniofacial features of the various syndromes can be so similar that the digital anomalies may be the sole differentiating physical finding to allow a clinical diagnosis.

Case series

Seventy-six patients with sCS (35 female [46.1%] and 41 male [53.9%]), with a mean age of 4.5 years (range 0.2-19.2 years), were compared with 86 control subjects (38 female [44.2%] and 48 male [55.8%]), with a mean age of 6.4 years (range 0.1-17.8 years). Untreated sCS patients < 1 year old had lower CBF than control subjects. In older age categories, CBF normalized to values observed in controls. Graphical analyses of CBF by age showed that the normally expected peak in CBF during childhood, noted at 4 years of age in control subjects, occurred at 5-6 years of age in patients with sCS. Patients with longitudinal pre- to postoperative CBF measurements showed significant increases in CBF after surgery.

Untreated patients with sCS < 1 year old have lower CBF than control subjects. Following vault expansion, and with age, CBF in these patients normalizes to that of control subjects, but the usual physiological peak in CBF in childhood occurs later than expected [1)].

A total of 6 patients with syndromic craniosynostosis underwent endoscopic suturectomy followed by helmet therapy during the study period. Of these, 3 patients were male. The involved syndromes included Crouzon, Pfeiffer, Jackson-Weiss, Muenke, Saethre-Chotzen, and craniosynostosis-3 (n = 1 each). The patients underwent endoscopic surgery at a median age of 2.1 months (range 0.9-4.1 months). The median estimated blood loss was 30 ml (range 20-100 ml), with 2 patients requiring a transfusion. The median length of stay in the hospital was 1.5 days (range 1-4 days), and the median follow-up was 29.0 months (range 16.8-81.7 months), with 1 patient (16.7%) requiring an open revision. Three patients (50%) were classified as Whitaker Category I at the last follow-up. The patients for whom additional open surgery was performed or recommended (Whitaker Category IV) were the oldest patients in the cohort, ranging from 2.6 to 4.1 months at the time of surgery. CONCLUSIONS This series demonstrates that endoscopic surgery can be sufficient to treat syndromic craniosynostosis without subsequent open calvarial remodeling over a median follow-up period of at least 2 years. The findings suggest that younger age at the time of endoscopic surgery may be an important factor in determining the sufficiency of this procedure. Even among patients who require subsequent open calvarial remodeling, early endoscopic surgery may allow for growth and development of the brain and skull while delaying the need for open remodeling until the patient is older and can better tolerate the procedure [2)].

1)
Doerga PN, Lequin MH, Dremmen MHG, den Ottelander BK, Mauff KAL, Wagner MW, Hernandez-Tamames JA, Versnel SL, Joosten KFM, van Veelen MC, Tasker RC, Mathijssen IMJ. Cerebral blood flow in children with syndromic craniosynostosis: cohort arterial spin labeling studies. J Neurosurg Pediatr. 2019 Dec 27:1-11. doi: 10.3171/2019.10.PEDS19150. [Epub ahead of print] PubMed PMID: 31881544.
2)
Hersh DS, Hoover-Fong JE, Beck N, Dorafshar AH, Ahn ES. Endoscopic surgery for patients with syndromic craniosynostosis and the requirement for additional open surgery. J Neurosurg Pediatr. 2017 May 5:1-8. doi: 10.3171/2017.2.PEDS16710. [Epub ahead of print] PubMed PMID: 28474983.