CRANIOSYNOSTOSIS
Pathological condition that results from premature fusion of one or more sutures in the cranial vault; associated with a deformity of the vault and cranial base.
Development

- Bones of the cranium
- Skull base and the calvarial vault
- Growth of skull bones
- Expanding growth of the brain.
Brain Growth

- At term has nearly 40 percent of his or her adult brain volume,
- And this increases to 80 percent by three years of age.
- Continues to grow until the age of 12 yrs
The cranium

Mature suture closure occurs by 12 years of age, but completion continues into the third decade of life and beyond.

<table>
<thead>
<tr>
<th>Type of suture/fontanelle</th>
<th>Time to closure</th>
</tr>
</thead>
<tbody>
<tr>
<td>Metopic suture</td>
<td>Nine months to two years (may persist into adulthood)</td>
</tr>
<tr>
<td>Coronal, sagittal, lambdoid sutures</td>
<td>40 years</td>
</tr>
<tr>
<td>Anterior fontanelle</td>
<td>Nine to 18 months</td>
</tr>
<tr>
<td>Posterior fontanelle</td>
<td>Three to six months</td>
</tr>
<tr>
<td>Anterolateral fontanelle</td>
<td>Three months</td>
</tr>
<tr>
<td>Posterolateral fontanelle</td>
<td>Two years</td>
</tr>
</tbody>
</table>
HISTORY AND PATHOGENESIS

- Otto
- Stahl  Hyrtl
- 1851, Virchow
Suture Growth

- Sutures allow growth perpendicular to them.
- Growth at suture lines is related to brain growth.
Virchow classified the different types of skull deformity. Van der Klaauw, in 1946, and Moss, in 1959, described the cranial base source of abnormal physical stress leading to dural abnormalities that yielded premature sutural fusion.
Animal studies

The cranial vault abnormalities typical of synostosis can be produced with experimental fusion of developing cranial vault sutures.

L.C Lane

Lannelogue-1890

Tessier
Incidence

- One per 1,800 to 2,200 live births
- Males: Sagittal and metopic stenosis
- Females: Coronal

Theories of Craniosynostosis

- Sommering (1839)

- Virchow (1821) and Otto (1830)

- Jane JA
Theories of Craniosynostosis

- Moss (1959)
  - Described functional matrix theory. According to this theory, cranial base abnormality was the primary pathological process and cranial vault suture abnormality was secondary as cranial base mature embryologically before cranial vault.

- Persson (1979)
  - Cranial vault suture pathology may be primary in the development of synostosis leading to cranial base and facial deformity.

- Marsh and Vannier (1986)
  - Following cranioplasty in patients with individual suture craniosynostosis, surgery altered only the cranial vault structure, the previously developed cranial base abnormalities were not ameliorated.
Familial Non syndromic Craniosynostosis

Syndromic craniosynostosis

- Less common (20 percent)
- More than 150 syndromes with craniosynostosis have been identified.
- Multiple sutures are involved.
- Autosomal dominant

Etiology

- Sporadic in most instances
Risk factors

- White maternal race
- Advanced maternal age
- Male infant sex
- Maternal smoking
- Residence at high altitude
- Nitrosatable drugs (e.g., nitrofurantoin, chlordiazepoxide, chlorpheniramine)
- Certain paternal occupations (e.g., agriculture and forestry, mechanics, repairmen)
- Fertility treatments.

Pathophysiology

- Cranial sutures - fibrous joints
- Abnormal osteoblastic activity - observed in cultures of synostotic bone
- Decreased growth rate
- Decreased alkaline phosphatase production
- Increased levels of osteocalcin
  - platelet-derived growth factor
  - epidermal growth factor
• Fibroblast growth factor and fibroblast growth factor receptor (FGFR) regulate fetal osteogenic growth—
  • Expressed in cranial sutures in early fetal life.
  • Mutations in the gene coding for FGFR1 → Pfeiffer's disease.
  • FGFR2 → Apert's syndrome and Crouzon's disease.
<table>
<thead>
<tr>
<th>Classification</th>
<th>Descriptions</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Primary</strong></td>
<td></td>
</tr>
<tr>
<td>Simple</td>
<td>Nonsyndromic: sagittal, coronal, metopic, lambdoid</td>
</tr>
<tr>
<td>Compound</td>
<td>Nonsyndromic: bicoronal</td>
</tr>
<tr>
<td>Syndromic</td>
<td>Crouzons disease, Apert's syndrome, Pfeiffer's disease, Saethre-Chotzen syndrome</td>
</tr>
<tr>
<td><strong>Secondary</strong></td>
<td></td>
</tr>
<tr>
<td>Metabolic disorders (e.g., hyperthyroidism)</td>
<td></td>
</tr>
<tr>
<td>Malformations (e.g., holoprosencephaly, microcephaly, shunted hydrocephalus, encephalocele)</td>
<td></td>
</tr>
<tr>
<td>Exposure of fetus (e.g., valproic acid, phenytoin)</td>
<td></td>
</tr>
<tr>
<td>Mucopolysaccharidosis (e.g., Hurler's syndrome, Morquio's syndrome)</td>
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</tbody>
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Types

- Scaphocephaly (Sagittal synostosis)

Derived from the Greek words scaphos, meaning boat, and kephali, meaning head.

Most common 45-50%
Plagiocephaly

- Greek word plagios, meaning oblique or sloping, and corresponds to unilateral coronal synostosis.
- 10-20% (Posterior plagiocephaly corresponds to lambdoid synostosis.)
- 1.3%
Left coronal synostosis
Ipsilateral frontal bossing

Ipsilateral ear displaced anteriorly

Ipsilateral occipitoparietal flattening

Contralateral occipital bossing

Trigonocephaly

- Derived from the Greek word trigonos, meaning triangular
- Metopic synostosis
- 5 → 10% incidence
Trigonocephaly
Trigonocephaly
Brachycephaly

- Both coronal sutures
Bilateral coronal synostosis results in a prominent frontal bone, flattened occiput, and anterior displacement of the skull vertex.

Oxycephaly

- Oxys, meaning sharp, and is a high, conical head with sharp bossing in the region of the anterior fontanelle
- Coronal and sagittal sutures results in an abnormally high conical head shape
- Encountered in syndromic types.
Cloverleaf skull deformity
(Triphyllocephaly)
Frequency

- Sagittal: 45%-50%
- Unilateral coronal: 15%
- Metopic synostosis: 5%
- Lambdoid: 1.3%
CRANIOSYNOSTOSIS SYNDROMES

- 10-20\% of cases
- Autosomal Dominant
- Linked to Chromosome 10
- Multi-sutural, complex case
- If a suture is fused, check hands, feet, big toe, and thumb
Crouzon’s

- Autosomal dominant pattern.
- One of every 25,000 live births.
- 5 percent of cases of craniosynostosis.

Clinical findings

- Brachycephaly
- Significant hypertelorism, proptosis, maxillary hypoplasia, beaked nose

Intracranial anomalies

- Hydrocephalus
- Chiari 1 malformation,
- and hindbrain herniation (70 percent).
• Normal intellect
  • Normal extremities
  • 5% have Acanthosis nigricans
  • 30% have progressive hydrocephalus
Apert’s - “Crouzon’s with Hand Involvement”

Syndactyly
PFEIFFER SYNDROME

- 1 in 2 lakhs
- Clover leaf skull in 20%
- Broad thumbs, broad great toes
- Intelligence is reported to be normal
CARPENTER SYNDROME

- Autosomal recessive.
- Syndactyly of feet.
- Sagittal and lambdoid suture closes first coronal last.
- Cardiac abnormalities.
Diagnosis
Passage of the head through the birth canal deforms the head. This shape is retained for 2-3 weeks postnatally.

Early diagnosis is important. The brain grows rapidly during this period. Delay only worsens the deformity of the head shape.
Detailed history

- Birth, sleeping position.
- Head tilt, torticollis deformational plagiocephaly.
- Family history: Abnormal head shape or multiple systemic problems (e.g., cardiac, genitourinary, musculoskeletal).
- Detailed history.
Clinical Exam

• If a suture is fused, check hands, feet, big toe and thumb
FUNCTIONAL CONSEQUENCES

Intra cranial hypertension

- Complicates one third of cases
- Principal indication of surgery
- ICP monitoring
- Syndromic forms → 30%
- 17% single suture
- Causes
  - Abnormal venous drainage
  - Respiratory obstruction
  - Chiari malformations
Hydrocephalous

- 4% to 18%
- Communicating
- Causes
- Cerebral maldovelopment
- Brain atrophy
- Abnormal csf circulation
- Venous outflow obstruction
- Hind brain herniation
- Aqueductal stenosis
Respiratory Abnormalities

- Syndromic craniosynostosis
- Manifest during sleep
- Maxillary hypoplasia, choanal stenosis, tonsillar hypertrophy
- Nasal stents, tonsillectomy or tracheostomy
- Nocturnal CPAP
- Surgical correction of midfacial hypoplasia
Feeding

- Abnormalities of palatal shape and movement
- Disordered dentition
- Dental malocclusion
- Nasogastric tube or gastrostomy
Vision

- Chronically raised ICP-papilloedema-optic atrophy (Crouzon syndrome)
- Shallow orbits
- Primary optic atrophy: compression, traction
- Early craniectomy
Plain Films

- Simple and inexpensive,
- Absent or line of increased density
- Harlequin appearance → coronal
- Cannot differentiate Lambdoid synostosis and deformational plagiocephaly (plagiocephaly without synostosis).

— To visualize all the sutures, special Waters views must be taken.
Ultrasound

- Noninvasive
- More effective than plain skull radiographs in detecting fused sutures
- Accuracy depends on a reliable and experienced operator.
CT Scan

- Standard for the complete visualization of the skull and cranial sutures.
- Detailed anatomy of the calvaria and the brain parenchyma.
- Document effect of corrective surgery.
MRI

- Complex craniosynostosis
- Improved definition of intracranial soft tissue structures
- Hindbrain herniation
- Identify sites of respiratory obstruction
Radio isotope scanning

- Diminished uptake → complete fusion
ICP monitoring

- Clinically occult – Majority
- Radiological signs inconclusive
- Deciding nature and timing of surgery
- Features of ↑ ICT
  - Mean pressure > 15 mm Hg
  - Raised baseline value
  - Prolonged plateau wave
Management

Surgery vs. Conservative Management
Goal

- Normalization of deviated appearance, growth and function of skull
- Keep the suture open till brain growth is complete
- Rarely achieved
Indications
Timing of surgery

Early operation (3-6 months)

journal of pediatric neurosciences


Pediatric craniofacial surgery for craniosynostosis: YN Anantheswar¹, NK Venkataramana²
¹ Department of Plastic Surgery, Manipal Hospital, Kengeri, Bangalore, India
² Advanced Neuroscience Institute, BGS Global Hospital, Kengeri, Bangalore, India
Management Considerations in the Treatment of Craniosynostosis
John A. Persing, M.D.
New Haven, Conn.
Plastic and Reconstructive Surgery • April 2008
Early frontocranial remodelling is performed between 2 and 4 months for brachycephalies. Other operated on between 6 and 12 months of age. For syndromal craniofacial synostosis, two-step operation: forehead advancement first, facial advancement later, to avoid the risk of frontal osteitis.

Late intervention

- Closer the cranium is to the adult size, the less overcorrection for reconstruction and the better the ultimate skull shape.
- Higher risk of recurrent deformity
- Surgical correction more complex
Basic mechanisms

- Passive reshaping: Generous removal of bone
- Strip craniectomy
- Morcellation
- Active reshaping: Frontoorbital advancement, Cranial vault reshaping
Incision

- Zigzag bicoronal incision
- Prevents parting of the hair along a straight line
- Scar tends to spread less due to redistribution of the forces.
- Incision begins slightly anterior and superior to the helix of the ear.
- Electrocautery is used cautiously.
Sagittal craniosynostosis

- Correction of scaphocephaly
- Frontal bossing and occipital protrusion
- Initial surgical procedures included a narrow-strip craniectomy, which reduced restenosis.
- Wider and more extensive craniectomy did not address frontal bossing and occipital bathrocephaly. This relied on the growing brain to correct these deformities.
More extensive cranial vault remodeling with barrel-stave osteotomy - Spring assisted cranioplasty - With the advent of endoscopes in neurosurgery, extended-strip craniectomy is performed and the patient is placed in a custom-made molding helmet to correct the frontal bossing and bathrocephaly.

Rapid recovery of the child and diminished need for blood transfusion,
Bilateral coronal stenosis

- Extended bicoronal craniectomies with reconstruction of the forehead.
- The supraorbital bar or brow is reshaped and advanced forward with the forehead.
- The reconstructed forehead and brow are rigidly fixed to the nose and lateral orbits with microplates.
Metopic stenosis

- Objectives:
  - Increase in width of the bifrontal diameter,
  - Increase in volume of the anterior cranial fossa, and
  - Normalization of frontal bone shape.

- The frontal bones and the frontoorbital bones are excised and transferred to the side assembly.
- The frontoorbital bar is advanced to create an appropriate brow position.
- The interdacyon distance is increased by placing a bone graft between the 2 halves of the frontoorbital bars.
Syndromic craniosynostosis

- Initial fronto-orbital and cranial vault remodeling,
- A midface advancement procedure with or without distraction (Le Fort III or monobloc),
- Secondary orthognathic surgery to correct any dentofacial deformities (Le Fort I, mandibular osteotomies).
Conservative Therapy for Deformational Plagiocephaly

• Re-positioning
• If no improvement by 6 months...
Long Term Follow-Up

- Speech
- Genetic Counseling
- Feeding/Swallowing
- Ophtho
Thank you