Craniosynostosis

Jerome M. Volk III, M.D.
Assistant Professor-LSUHSC Department of Neurosurgery
Children’s Hospital of New Orleans Department of Neurosurgery
Disclosure

- Nothing to disclose
Infant Skull Deformity

- Sagittal synostosis
- Metopic synostosis
- Normocephaly
- Bilateral coronal synostosis
- Unilateral coronal synostosis
- Lambdoid synostosis

Mild, nonsurgical problem of cosmetic concern only

Deformational posterior plagiocephaly
Epidemiology

- Craniosynostosis is seen in every 1 out of 2,000-2,500 births.
- Most commonly only single suture synostosis is seen.
- White non-Hispanic mothers have increased frequency.
Pathophysiology

• The etiology is not completely understood, and is most commonly sporadic.

• Advanced paternal age and maternal smoking have been implicated.

• Certain genetic mutations have been associated with craniosynostosis.
  – *FGFR1-3*, *NELL1*, *MSX2*
Surgical Indication

• The two main surgical indications are:
  – Correct skull shape for aesthetic and psychosocial considerations.
  – Make sure there is adequate space for normal brain growth to occur.

• There is a low percentage risk of increased intracranial pressure in single suture synostosis (4-14%).
  – That percentage is much higher in multi-suture synostosis (47-67%)
Sagittal synostosis

- Sagittal synostosis is the most common form of craniosynostosis.
  - Accounts for 53-60% of cases
- Patients have a narrow elongated head shape (Scaphocephaly).
  - Biparietal narrowing
  - Occipital Bullet
  - Delayed anterior fontanelle closure
  - Perisutural ridging
Sagittal Synostosis
Metopic Synostosis

• Known as trigonocephaly.
• The metopic suture normally closes around 9 months of age, but can close as early as 3 months.
• 3rd most common type:
  – Accounts for 4-10% of cases.
Metopic synostosis
Metopic Synostosis

• Involves:
  – ridging along the metopic suture.
  – posterior displacement of the supraorbital rims
  – hypotelorism
  – flattening of the frontal bones
  – anterior displacement of the coronal suture
  – temporal narrowing
Metopic Synostosis
Metopic Synostosis
Coronal Synostosis

• 2\textsuperscript{nd} most common type
  – Accounts for 17-29% of cases
  – Unilateral coronal synostosis is more common than bilateral
Coronal Synostosis
Coronal synostosis

- Unilateral Coronal synostosis (Anterior Plagiocephaly)
  - Flattening of the frontal bone
  - Raised supraorbital rim
  - Nasal radix deviation
  - Anterior ear displacement
  - Harlequin deformity (raised sphenoid wing)
Coronal Synostosis

- Bilateral Coronal Synostosis (Brachycephaly)
  - Shortened anterior fossa
  - Widened biparietal diameter
  - Frontal towering (Turricephaly)
  - Recessed supraorbital rims
  - Flattened occiput
Coronal Synostosis

Scalp Incision site
Osteotomy
Stenosed right coronal suture
Lamboid synostosis

- Least common type
  - Accounts for less than 5% of cases
- Posterior Plagiocephaly
  - Trapezoid shape
  - Posterior/inferior displacement of ipsilateral ear
Positional Plagiocephaly

- Positional Plagiocephaly:
  - Parallelogram shape
  - Anterior ipsilateral ear displacement
  - Anterior ipsilateral frontal bossing
Positional Plagiocephaly

- Joint Guidelines Committee of the American Association of Neurological Surgeons (AANS) and the Congress of Neurological Surgeons (CNS) and American Academy of Pediatrics (AAP)

- Imaging:
  - Clinical examination is recommended for the diagnosis of plagiocephaly, and imaging is rarely necessary, except in cases in which clinical diagnosis is equivocal.
Positional Plagiocephaly

• Joint Guidelines Committee of the American Association of Neurological Surgeons (AANS) and the Congress of Neurological Surgeons (CNS) and American Academy of Pediatrics (AAP)

• Repositioning:
  – Repositioning is an effective treatment for deformational plagiocephaly.
    • Although there is Level 1 evidence that PT is more effective.
Positional Plagiocephaly

- Joint Guidelines Committee of the American Association of Neurological Surgeons (AANS) and the Congress of Neurological Surgeons (CNS) and American Academy of Pediatrics (AAP)

- Physical Therapy:
  - Physical therapy is recommended over repositioning education alone for reducing prevalence of infantile positional plagiocephaly in infants 7 weeks of age (Level I).
  - Physical therapy is as effective for the treatment of positional plagiocephaly and recommended over the use of a positioning pillow in order to ensure a safe sleeping environment and comply with American Academy of Pediatrics recommendations (Level I).
Positional Plagiocephaly

- Joint Guidelines Committee of the American Association of Neurological Surgeons (AANS) and the Congress of Neurological Surgeons (CNS) and American Academy of Pediatrics (AAP)

- Helmet Therapy:
  - Helmet therapy is recommended for infants with persistent moderate to severe plagiocephaly after a course of conservative treatment (Level II).
  - Helmet therapy is recommended for infants with moderate to severe plagiocephaly presenting at an advanced age (Level II).
Syndromic Craniosyntosis

- Represents fewer than 5% of cases
- Raised ICP is common
- Can require more than one surgery
- Must have a team approach to treatment
  - Airway abnormalities
  - Cognitive delay
  - Orthopaedic abnormalities
  - Genetic abnormalities
Syndromic Craniosynostosis
Crouzan’s Syndrome

- First described in 1912
- Associated with autosomal dominant inheritance pattern
  - *FGFR2* gene
- Increased paternal age is a risk factor
- Normal intelligence
- 1.5 per 100,000 per year
Crouzan’s Syndrome

- a “beaky” nose
- a recessed frontal region due to bicoronal synostosis
- prominent eyes (exorbitism) due to the combined recession of the infra- and supraorbital regions
- retruded maxilla

- Extra-cranial findings:
  - Cervical vertebral fusion
  - Ankylosis affecting particularly the elbows
- Monitor for increased ICP.
Apert’s Syndrome

- First described in 1906
- 1.5 per 100,000 per year
- Autosomal dominant inheritance
  - FGFR2 gene
- Developmental and learning difficulties
Apert’s Syndrome

- Tall and shortened from front to back (turribrachycephaly)
- Midfacial (maxillary) retraction
- Proptosis
- Downward cant to the palpebral fissures
- Hypertelorism

- Extra-cranial findings:
  - Syndactyly (fingers and toes)
  - Visceral and cutaneous abnormalities
  - Cervical spine fusion
- Initially just the coronal sutures are fused, but by 2 years of age all the sutures are fused.
Muenke Syndrome

- 1 per 30,000 per year
- Autosomal dominant inheritance
  - FGFR3 gene
- Synostosis typically affects either one or both coronal sutures
- Raised ICP is uncommon
- Learning difficulty is not uncommon
Pfeiffer Syndrome

• 1 per 100,000 per year
• Autosomal dominant inheritance
  – $FGFR1$ & $FGFR2$ gene
• Suture fusions that range from bicoronal synostosis alone to pan-synostosis
• Digital abnormalities
  – Usually shortened curved thumbs and great toes
- Pfeiffer Type I
  - Bicoronal synostosis
  - Midface retrusion
  - Digital abnormalities
  - Can have unaffected neurocognitive development
- **Pfeiffer Type II and III**
  - Severe midface and frontal retrusion
    - Airway obstruction
    - Ocular protrusion (corneal damage)
  - Shortening of the skull base and crowding of the posterior fossa (lamboid synostosis)
    - Increased risk of hydrocephalus
Thank you