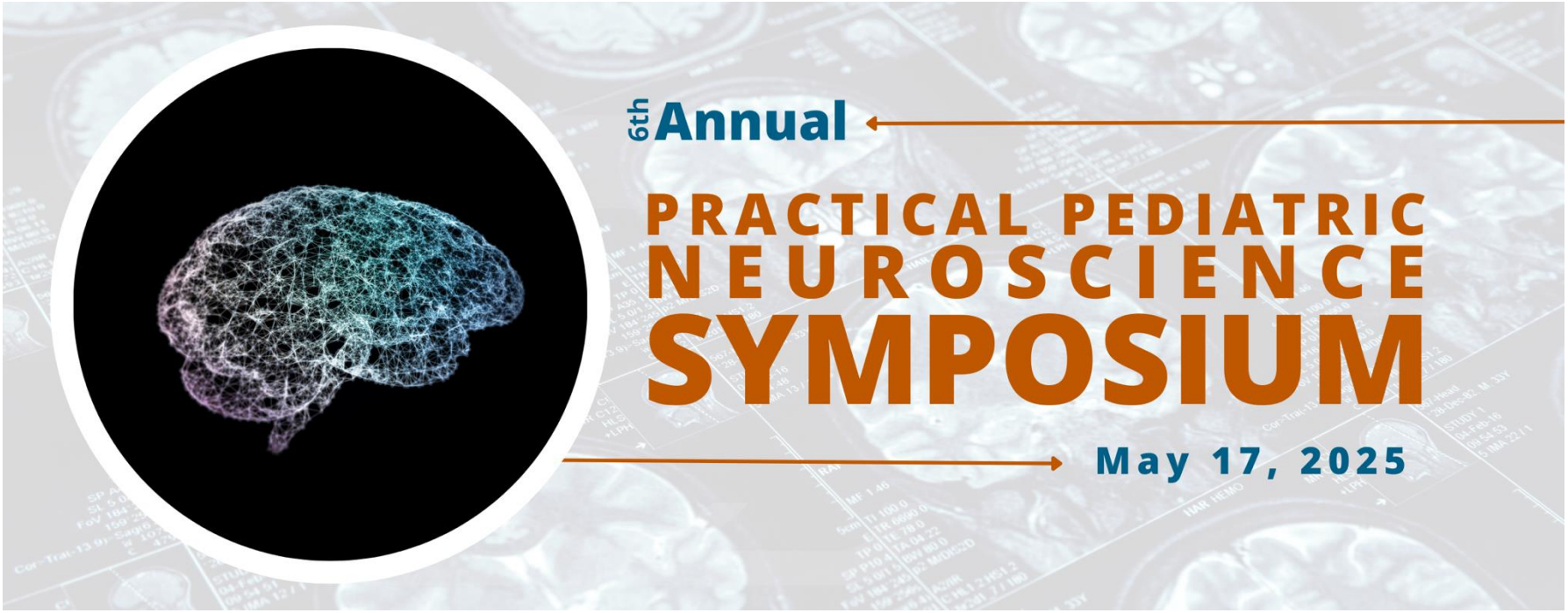

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Pediatric Neurosurgical Disease

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The video of the following presentation has been made available for free by UT Health Austin Pediatric Neurosciences at Dell Children's at:

https://youtu.be/vlCp9QeN2OM?feature=shared

To see all event presentations, please use this link for the 6th Annual Practical Pediatric Neuroscience Symposium presentation playlist:

https://youtube.com/playlist?list=PLPPnZ7QxWdeR-cpRyBnU2C2eEVh1bW1YR&feature=shared

Interprofessional Continuing Education

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Elias Rizk, MD, PhD

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Pediatric Neurosurgical Disease

Introduction

- Importance of pediatric neurosurgery in primary care
- High prevalence of neurosurgical conditions in children
- Common conditions: macrocrania, craniosynostosis, craniospinal dysraphism
- Role of pediatricians in early identification and referral
- Advances in diagnostic imaging and surgical techniques, improving outcomes

Macrocrania Overview

- Definition: head circumference > 95th percentile or rapidly increasing
- Importance of distinguishing between benign and pathological causes
- Three primary causes:
  - Hydrocephalus: accumulation of cerebrospinal fluid (CSF) leading to increased intracranial pressure
  - Benign extra-axial fluid collections of infancy (BEFI): self-limiting CSF accumulation in subarachnoid spaces
  - Familial (Weaver curves)
- Other rare causes include:
  - Genetic disorders: neurofibromatosis, achondroplasia, Canavan's and Alexander's diseases
  - Tumors: intracranial neoplasms causing mass effect
  - Trauma: post-traumatic subdural hematomas

Hydrocephalus Pathophysiology

- CSF produced at a rate of 0.3 mL/min, primarily by the choroid plexus
- Normal flow pathway:
  - Lateral ventricles → foramen of Monro → third ventricle → aqueduct of Sylvius → fourth ventricle → subarachnoid space → venous system via arachnoid granulations
- Hydrocephalus results from an imbalance in CSF production and absorption
- Two major types:
  - Communicating hydrocephalus: impaired CSF absorption due to meningitis, hemorrhage, or idiopathic causes
  - Non-communicating hydrocephalus: obstruction in the ventricular system, commonly at the aqueduct of Sylvius or foramina of Monro

Clinical Presentation of Hydrocephalus

- **Infants:**
  - Rapid head growth, full anterior fontanelle, widened sutures
  - Prominent scalp veins, poor feeding, irritability, vomiting
  - Developmental delays, hypertonia, sunsetting eyes (limited upward gaze)

Clinical Presentation of Hydrocephalus

- **Older children:**
  - Symptoms of increased intracranial pressure (ICP): morning headaches, nausea, vomiting
  - Vision changes, lethargy, difficulty in school performance
  - Papilledema, sixth nerve palsy, behavioral changes

Diagnosis of Hydrocephalus

- **Imaging Studies:**
  - Cranial ultrasound (if fontanelle is open)
  - CT scan: quick assessment of ventricular enlargement
  - MRI: gold standard for detailed visualization of ventricles and associated anomalies
  - Cine-flow MRI: evaluates CSF flow dynamics, useful for diagnosing aqueductal stenosis
- **Lumbar Puncture Considerations:**
  - Typically avoided in cases of suspected increased ICP due to risk of herniation

Treatment Options for Hydrocephalus

- **Medical Management:**
  - Limited role: acetazolamide and furosemide reduce CSF production but are not long-term solutions
- **Surgical Options:**
  - **Ventriculoperitoneal (VP) Shunting:**
    - Most common treatment
    - Catheter diverts excess CSF from the ventricles to the peritoneal cavity
  - **Endoscopic Third Ventriculostomy (ETV):**
    - Creates an alternate CSF drainage pathway between the third ventricle and subarachnoid space
    - Preferred for obstructive (non-communicating) hydrocephalus

Complications of Hydrocephalus Treatment

- **Shunt-related complications:**
  - Malfunction (30-40% failure rate within the first year)
  - Infections: Staphylococcus epidermidis most common pathogen (3-29%)
  - Overdrainage: causes subdural hematomas, postural headaches
- **ETV risks:**
  - Failure in younger children (<2 years old)
  - Risk of vascular injury, hormonal disturbances (diabetes insipidus)

Benign Extra-Axial Fluid Collections of Infancy (BEFI)

- Self-limiting accumulation of CSF in subarachnoid spaces
- Clinical features:
  - Rapid head growth between 3 and 18 months
  - No signs of raised intracranial pressure
  - Normal development or mild transient delays in motor skills
- Diagnosis:
  - CT/MRI showing symmetric subarachnoid space enlargement
  - Differentiation from subdural hematomas or chronic SDH
- Management:
  - Regular monitoring, resolves by 2 years
  - Rarely requires intervention unless associated with other conditions

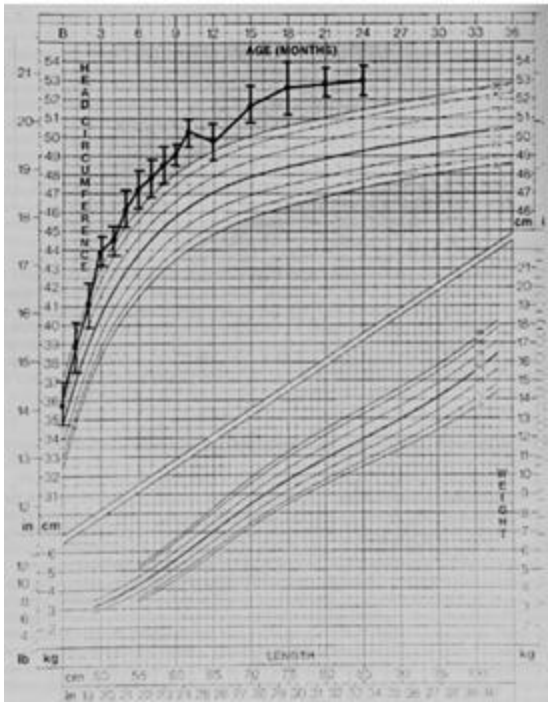
Benign Extra-Axial Fluid Collections of Infancy (BEFI)

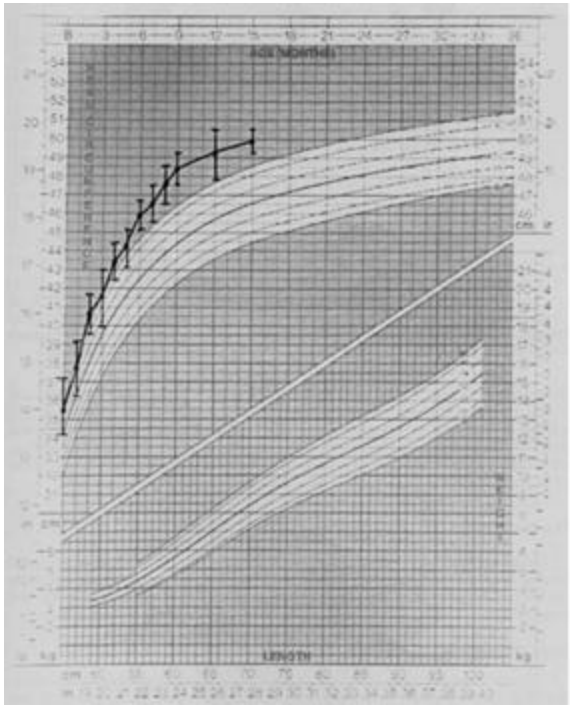
- BEFI vs subdural fluid collections or hematomas of child abuse
- Subdural hematomas that follow child abuse are of mixed density or are slightly more hyperdense than CSF and compress the adjacent sulci or produce mass effect on the ventricles or cerebral hemispheres
- MR imaging is extremely useful in questionable cases

Benign Extra-Axial Fluid Collections of Infancy (BEFI)

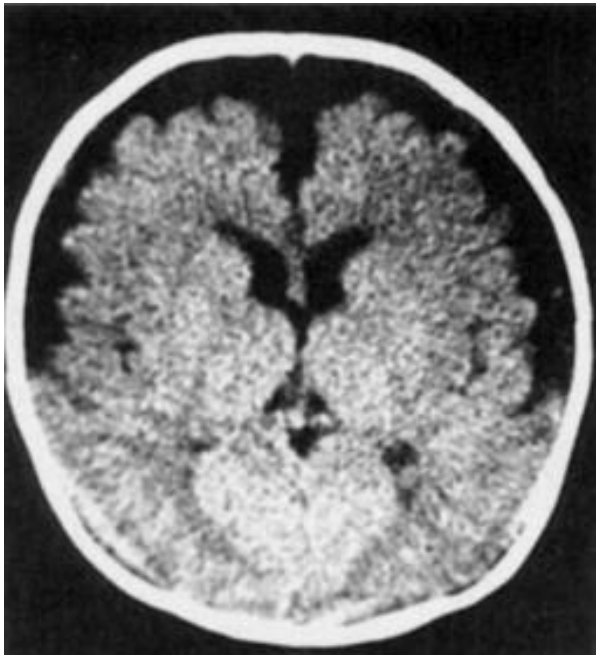
- Treatment is unnecessary in essentially all cases because CSF absorption is corrected when adequate CSF absorption capacity develops, usually by 2 years of age
- Infants with BEFI should be watched closely during the first 18 to 24 months with frequent head circumference measurements and developmental assessments
- Ventricular shunting is reserved for exceptional patients

Head circumference growth chart for a cohort of infants with BEFI

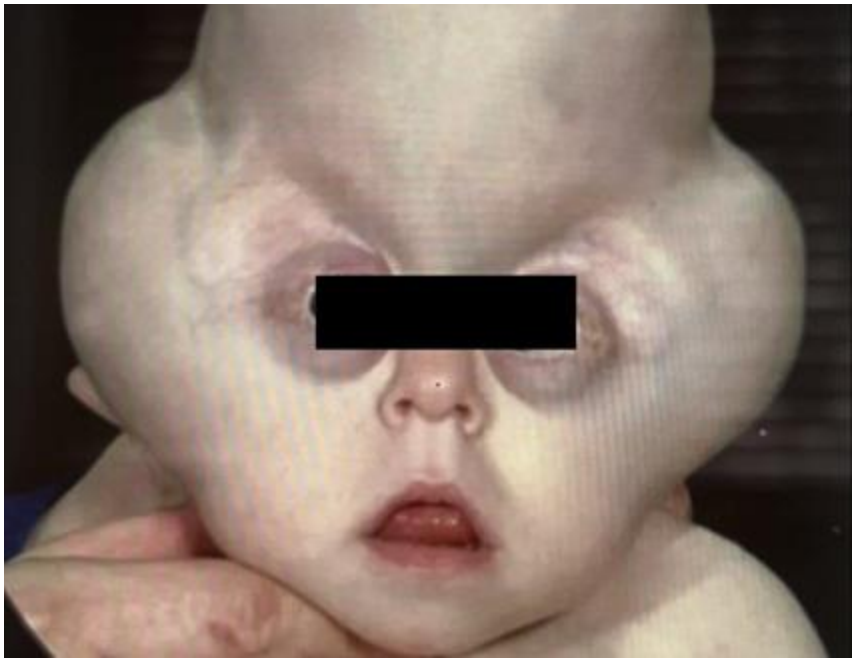




CT scan characteristics of BEFI



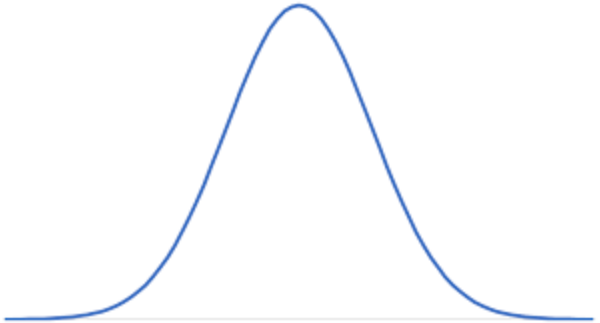
Microcrania Overview



- Head circumference of less than the fifth percentile for age:
  - Genetic predisposition
  - Disordered brain development, prenatal or postnatal brain insults
  - Craniosynostosis (Kleeblattschadel)

What To Do With the Fontanelle

- The normal time of fontanelle closure depends on the patient's gestational age at birth.
- The posterior fontanelle usually cannot be palpated after two months of age.
- The anterior fontanelle generally closes between 10 and 24 months of age.
  - The fontanelles of premature infants tend to close later.
- Growth of fibrous tissue over the fontanelle precedes closure by several months and may be difficult to distinguish from true closure on physical examination.
  - 5 to 9 months – 3 percent
  - 10 to 12 months – 21 percent
  - 13 to 15 months – 35 percent
  - 16 to 18 months – 60 percent
  - 19 to 21 months – 84 percent
  - 22 to 24 months – 89 percent



What To Do With the Fontanelle

- Early closure of the anterior or posterior fontanelle is not uncommon in an otherwise normal child
- Other causes of early closure:
  - Craniosynostosis
  - Hyperthyroidism
  - Hypophosphatasia
  - Hyperparathyroidism
- Decreased head circumference (or decreasing percentile) indicates a problem

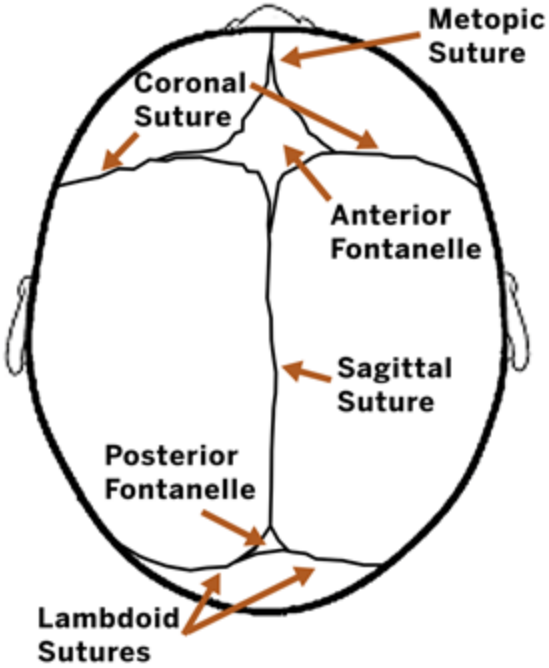
What To Do With the Fontanelle

- Delayed closure
  - Normal variation
  - Congenital hypothyroidism
  - Primary megalencephaly
  - Increased intracranial pressure
  - Down syndrome
  - Rickets

Craniosynostosis Overview

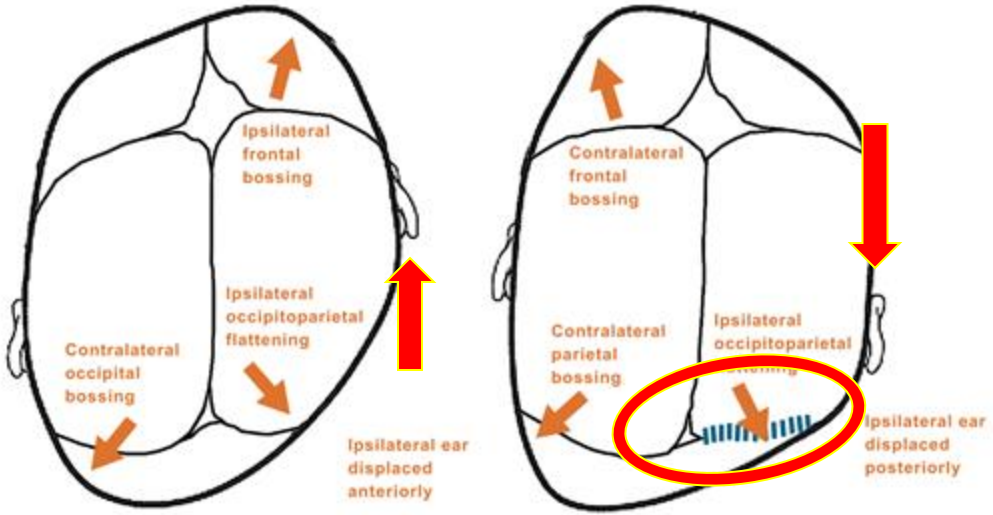
- Definition: premature fusion of cranial sutures leading to abnormal skull shape
- Types:
  - Primary: isolated suture fusion
  - Secondary: associated with syndromic conditions (Apert, Crouzon, Pfeiffer syndromes)
- Clinical concerns:
  - Skull deformity
  - Potential for increased ICP in multisutural involvement

Cranial Suture Anatomy

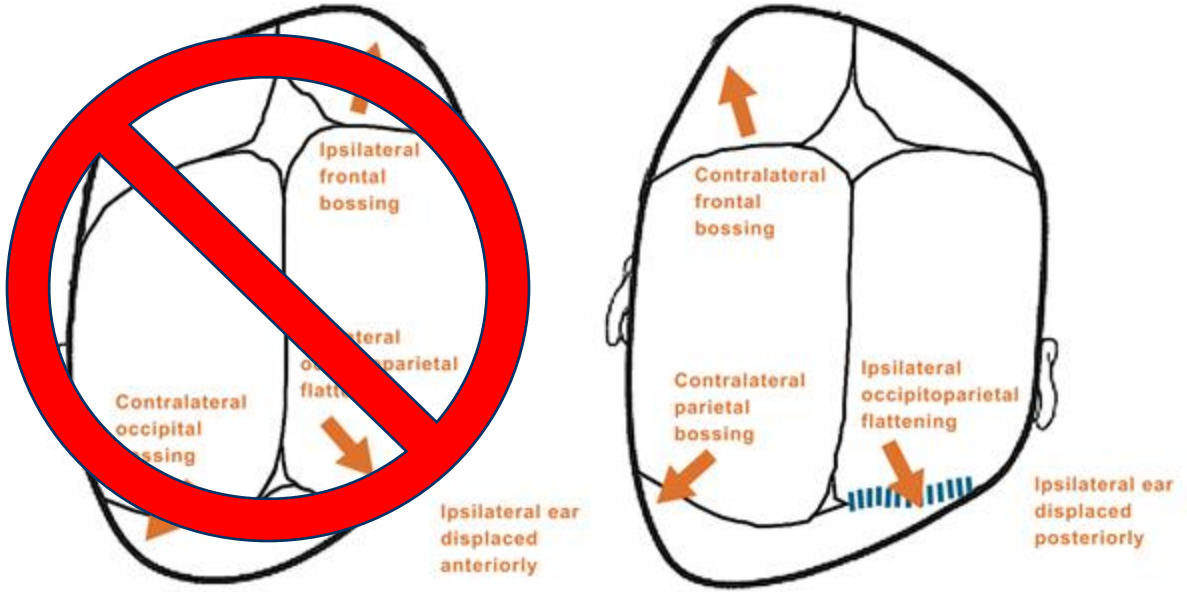


Misshapen Head

Positional Plagiocephaly vs Lambdoid Craniosynostosis

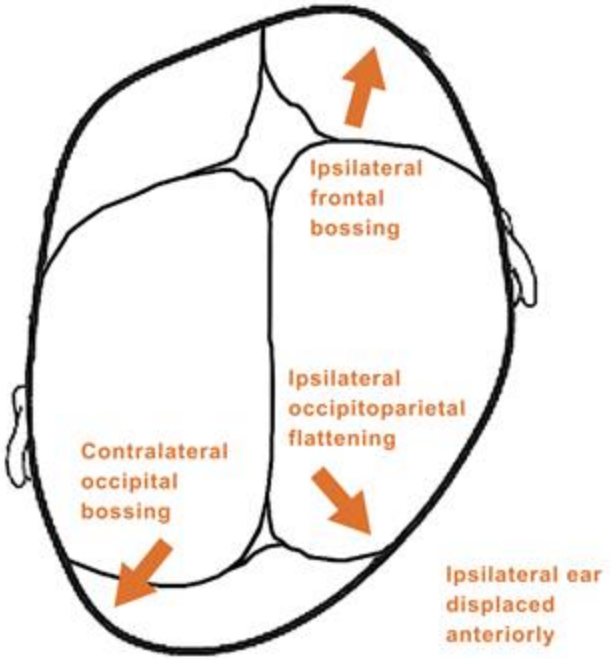


Who Should Get Surgery?



Positional Plagiocephaly

- Incidence of 5-48% at 1 year of age
- Typical presentation
  - Rhomboid shape, “box”
  - 80% right sided
  - 80% post-natal
  - Boys:girls – 2:1
- Cause
  - Intrauterine molding
  - Patient preference
  - Torticollis
  - “Back to sleep”



Positional Plagiocephaly – Treatment

- Moderate to severe based on cephalic index
  - Max width/max length x 100
- Molding helmet
  - 23 hrs/day 4-6 months, $3500 = braces

Molding Helmets

- Option for children with moderate–severe deformities
- Literature demonstrates no long-term benefit from helmets vs repositioning
  - Two studies (Netherlands, New Zealand) suggest no additional long-term benefit from helmets
- No credible evidence suggesting an impact on any abnormality other than head shape/cosmesis

Craniosynostosis

- Craniosynostosis is found in both syndromic and nonsyndromic forms
- Nonsyndromic synostosis is more common, occuring in approximately 1 in 2500 live births
  - Sagittal synostosis is most common: 40-60%
  - Lambdoid synostosis is least common: 1-4%

Causes of Craniosynostosis

- Genetic disorders
- Metabolic disorders
  - Hyperthyroidism
- Mucopolysaccharidoses
- Mucolipidosis III
- Hematologic disorders
  - Thalassemias
  - Sickle cell
  - Polycythemia vera

Teratogens

Valproate
DPH
Retinoic acid

Malformations

Microcephaly
Encephalocele
Shunted hydrocephalus
Holoprosencephaly

Genetics of Craniosynostosis Syndromes

- Mutations of fibroblast growth factor receptor (*FGFR*) gene
  - Crouzon, Apert, Jackson-Weiss
- Mutations of *TWIST* gene
  - Pfeiffer, Saethre-Chotzen
- Mutations of *MXS2* (homeobox gene)
  - Boston craniosynostosis
- *FGFR*, *MXS2* involved in osteogenic growth and development
- Both expressed in developing calvarium and in osteogenic front of calvarial sutures

Detailed Analysis of Craniosynostosis Types

- **Brachycephaly**: bilateral coronal synostosis (broad, flat head)
- **Dolichocephaly**: sagittal synostosis (long, narrow head)
- **Trigonocephaly**: metopic synostosis (triangular forehead)
- **Plagiocephaly**: unilateral coronal or lambdoid synostosis (asymmetric skull)
- Diagnosis and imaging modalities
- Surgical interventions and timing for optimal outcomes

Common Syndromic Presentations

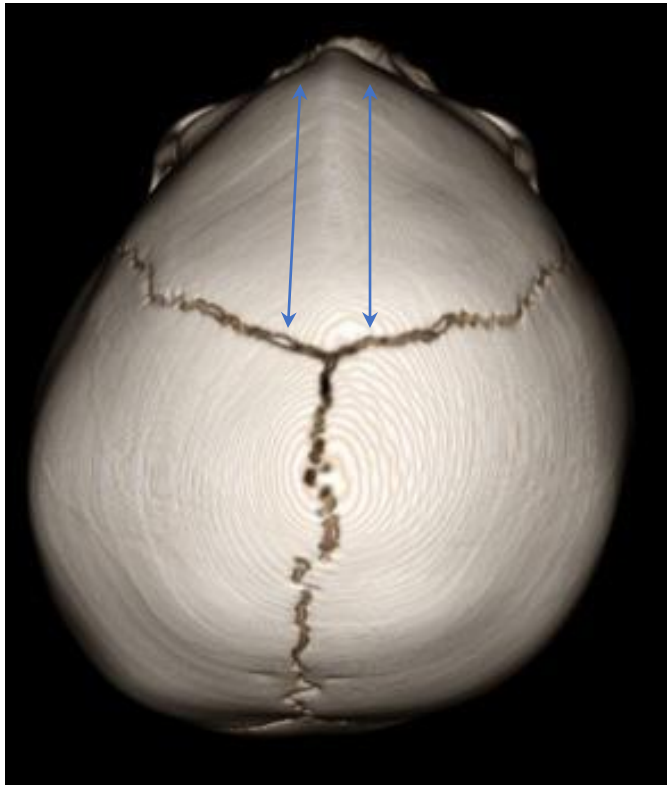
Syndrome	Inheritance	Sutures	Additional Characteristics
Crouzon	AD	Bicoronal	Midface hypoplasia, proptosis
Apert	AD	Bicoronal	Syndactyly, mitten hands, mental deficiency
Saethre-Chotzen	AD	Bicoronal	Ptosis, low-set hairline, syndactyly, brachydactyly, normal thumbs, broad toes
Pfeiffer	AD	Bicoronal	Broad thumbs and great toes, mild syndactyly
Jackson-Weiss	AD	Bicoronal	Broad first metatarsal and fused tarsal bones, normal thumbs

Treatment

- Goal of treatment is complete release of fused suture and correction of compensatory skull changes
- Timing of surgery is important to outcome
  - Early surgery utilizes the rapid brain growth potential
    - birth = 400g -> six months = 800g -> two and a half years = 1600g

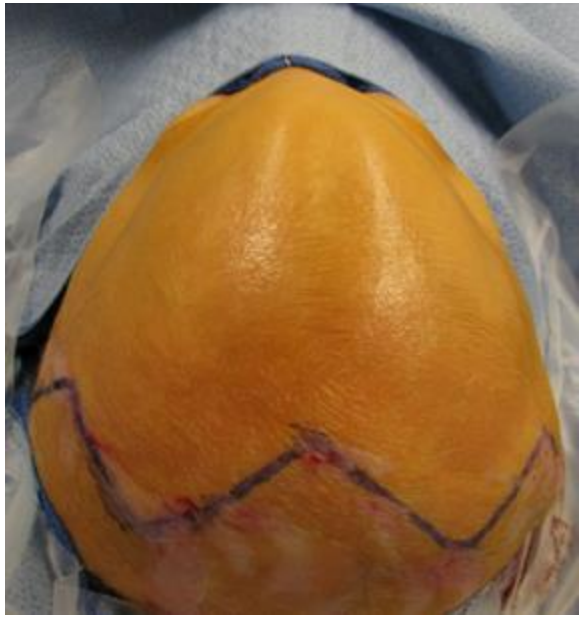
Metopic Synostosis

- Earliest suture to fuse at 6-8 months
- Clinical findings
  - Trigonocephaly (keel-shaped skull)
  - Metopic ridging
  - Bitemporal narrowing
  - Biparietal expansion
  - Hypotelorism

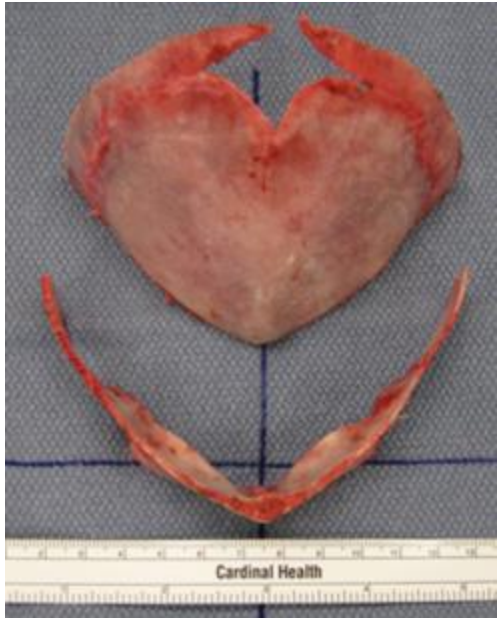


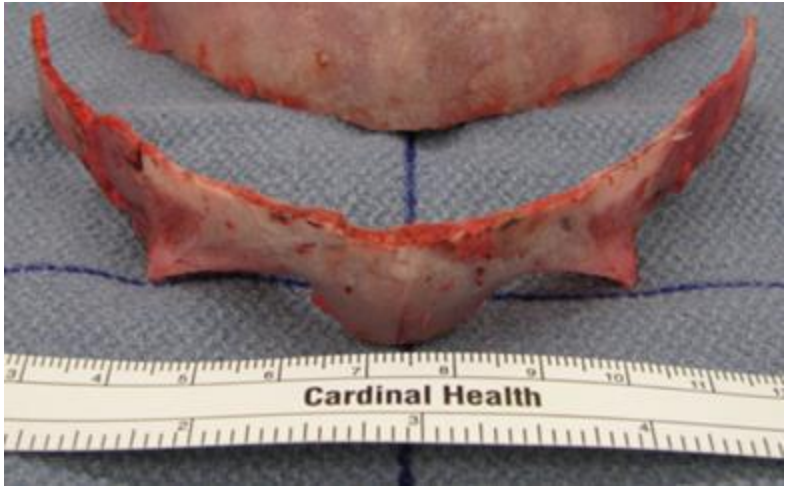
Metopic Synostosis – Treatment

- Surgical goal is maximal anterolateral expansion and correction of bitemporal narrowing

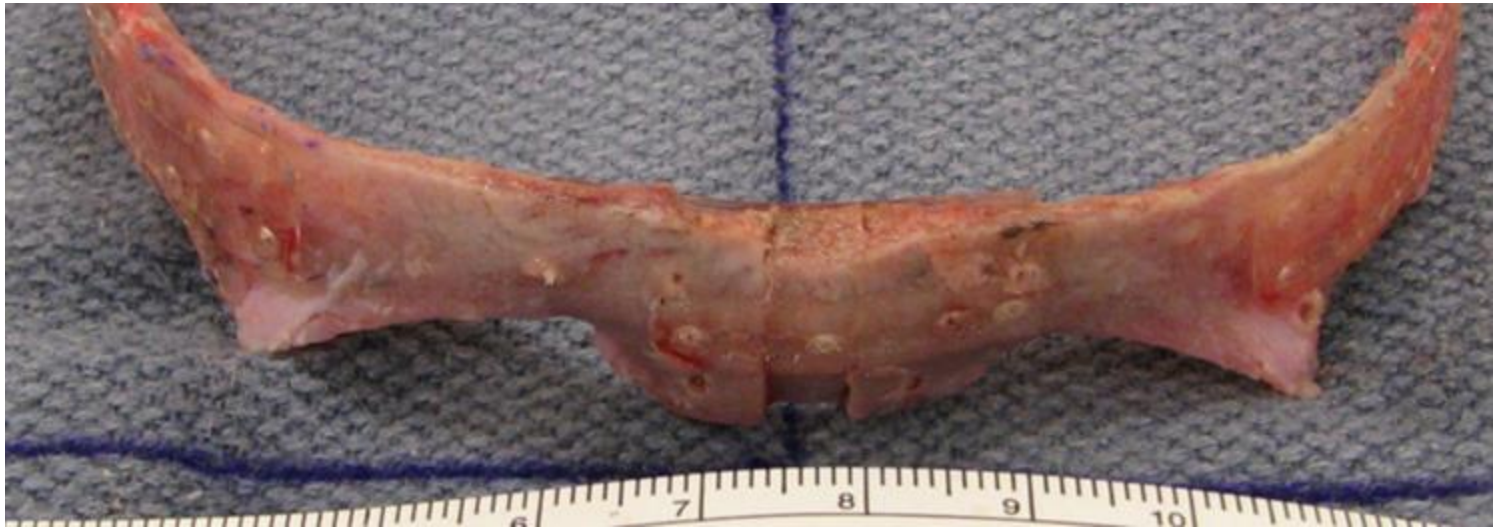


Metopic Synostosis – Treatment

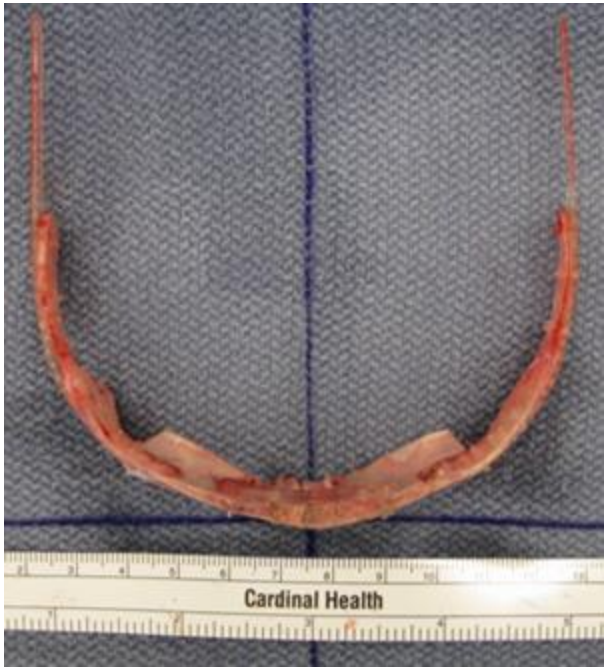


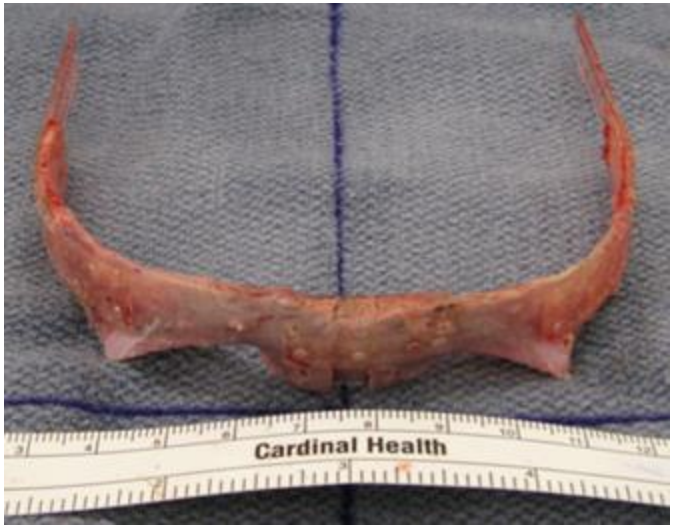


Supraorbital Bandeau Reshaping

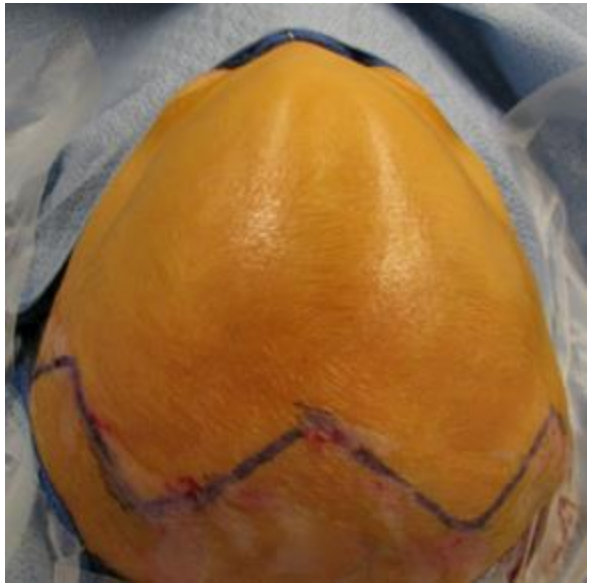


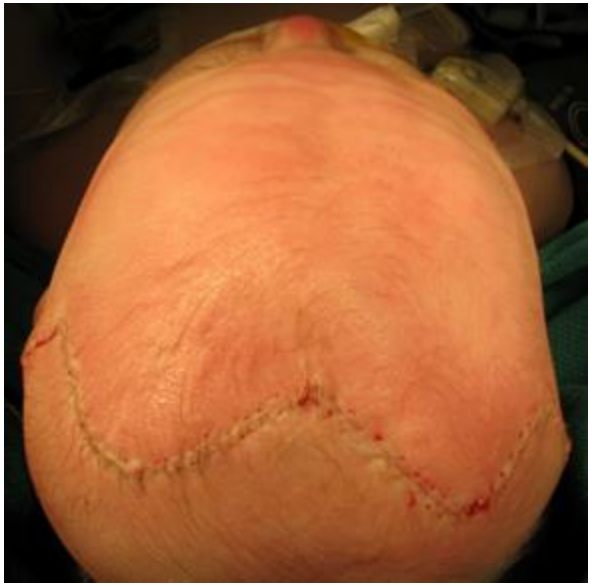
Metopic Synostosis – Treatment





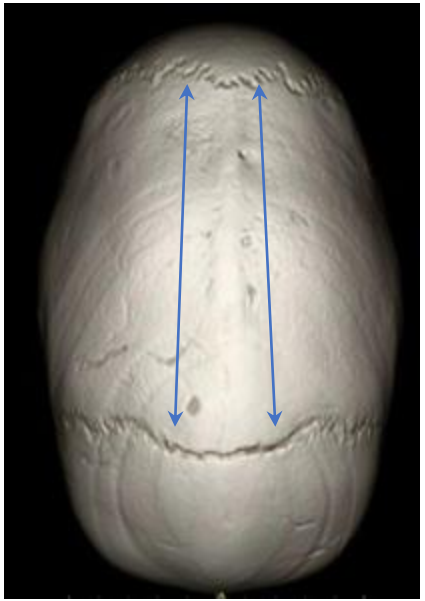
Metopic Synostosis – Treatment



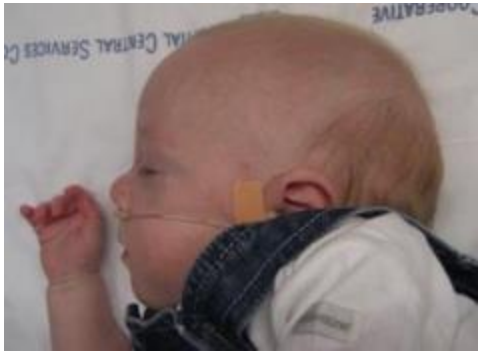


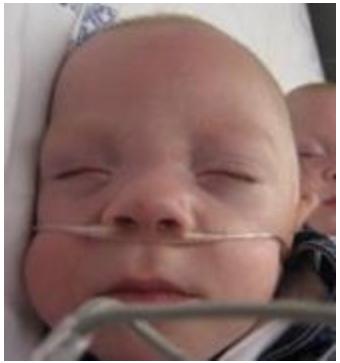
Sagittal Synostosis

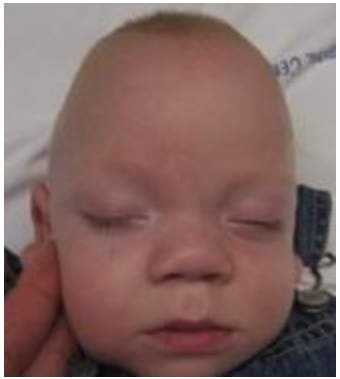
- Most commonly involved suture, 1:2000-1:5000
- Results in scaphocephaly
  - Anteroposterior elongation
  - Sagittal sutural ridging
  - Biparietal narrowing
  - Frontal and occipital bossing



Sagittal Synostosis vs NICUcephaly





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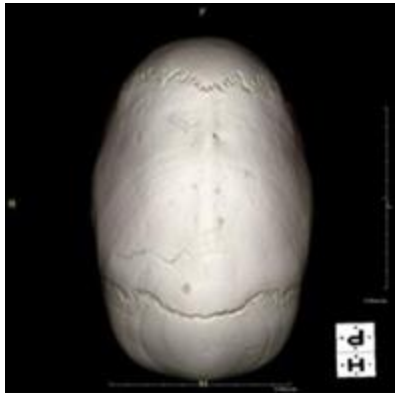
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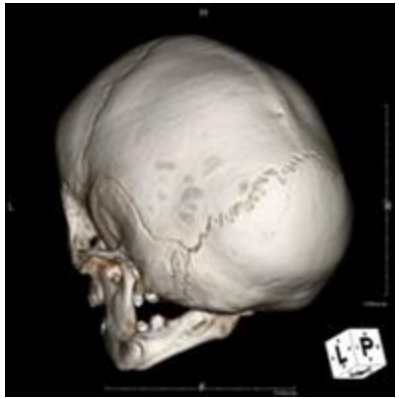
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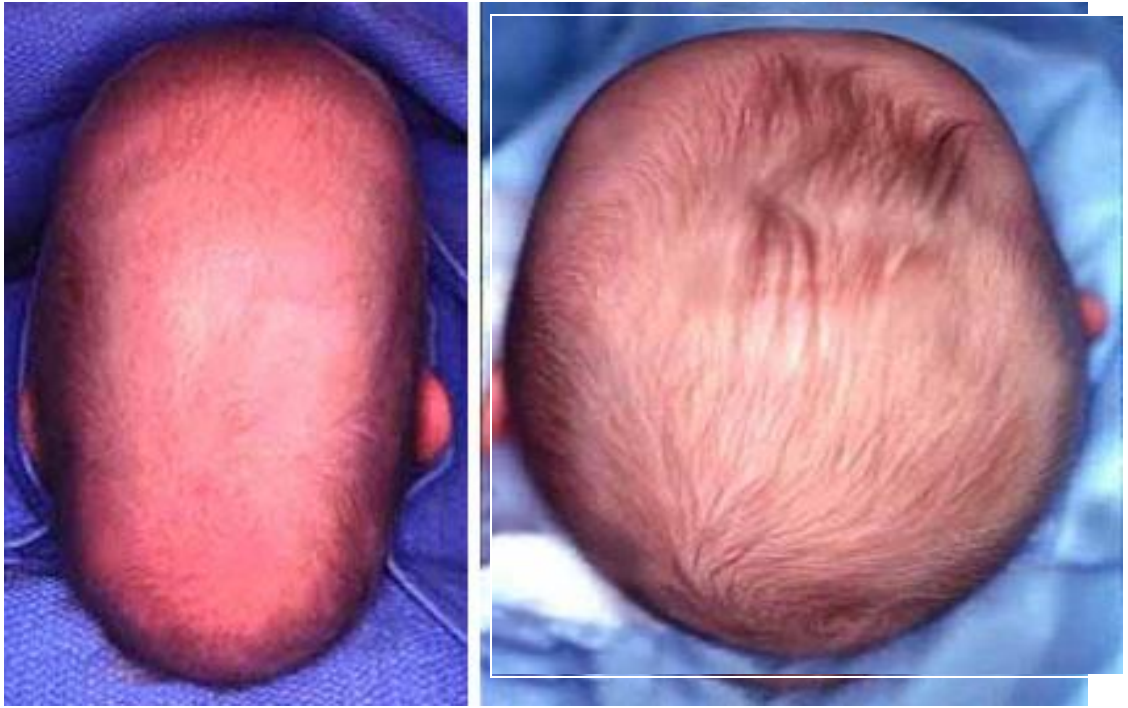
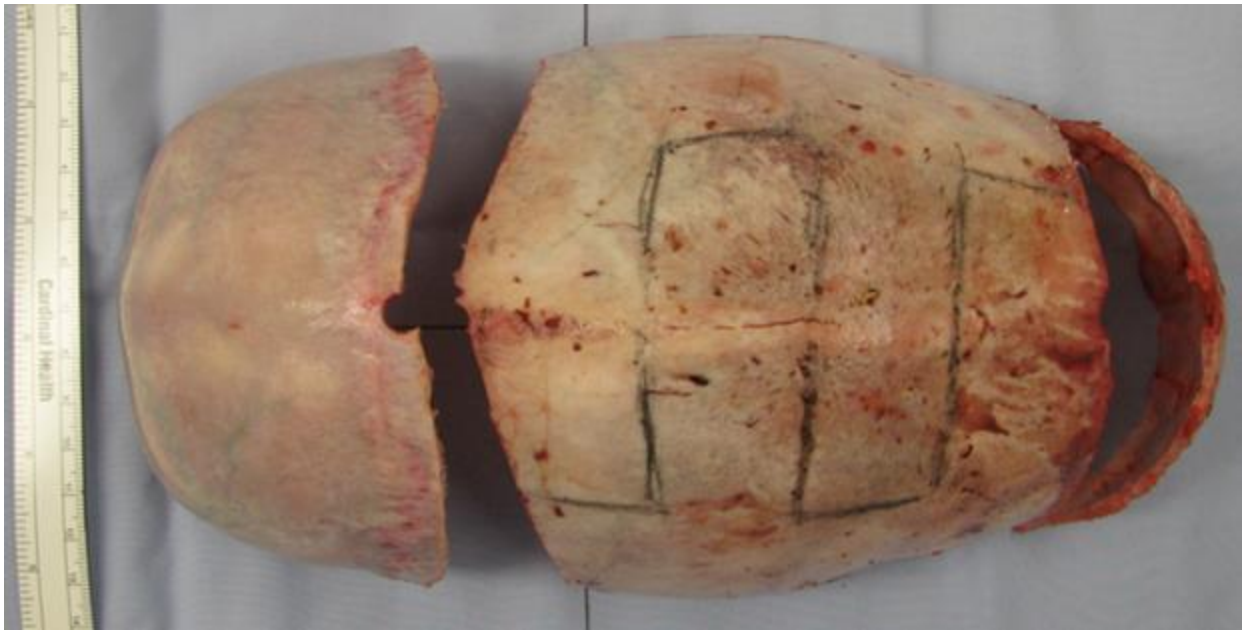
Sagittal Synostosis

- Treatment *earlier* than other synostoses
  - 6-12 weeks
  - More limited posterior calvarial reconstruction
  - 1.5-2 hours, ~50% transfusion rate
  - Endoscopic surgery (must be combined with a molding helmet for ~1 year)
- Delayed treatment (> 6 months) more extensive, involves total vault reconstruction

Posterior Vault Reconstruction



Sagittal Synostosis



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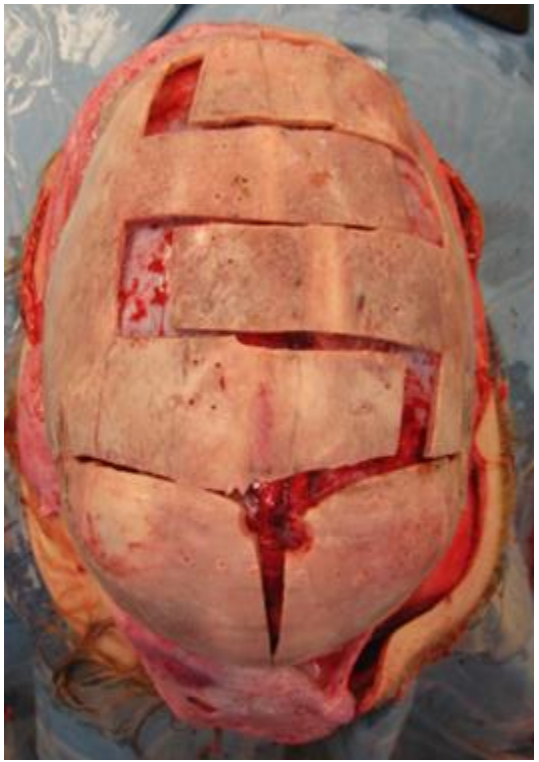
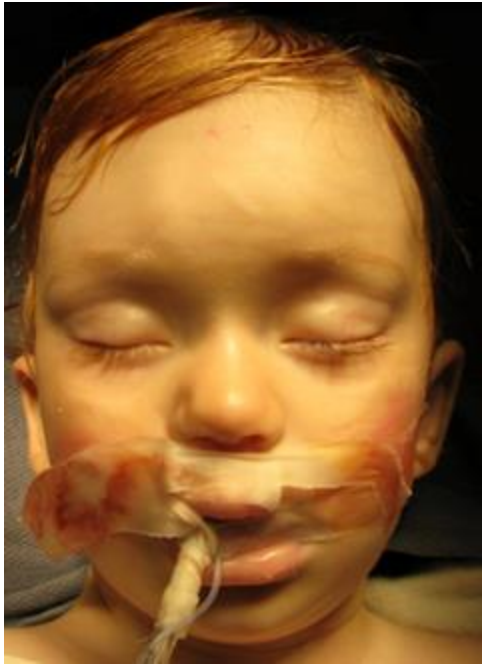
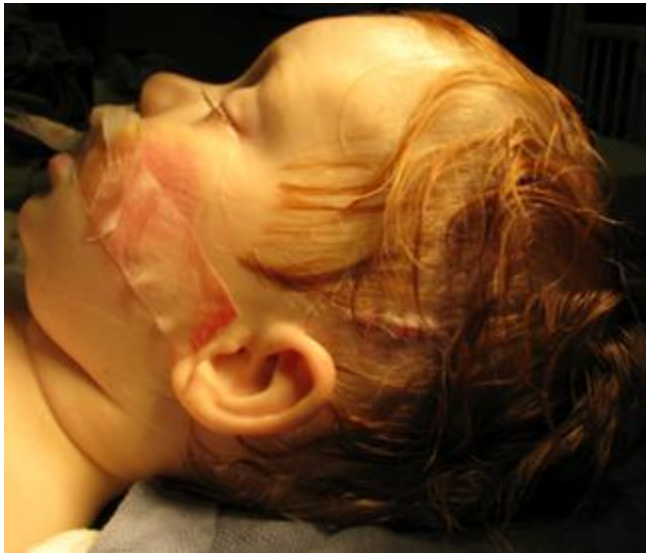


Sagittal Synostosis



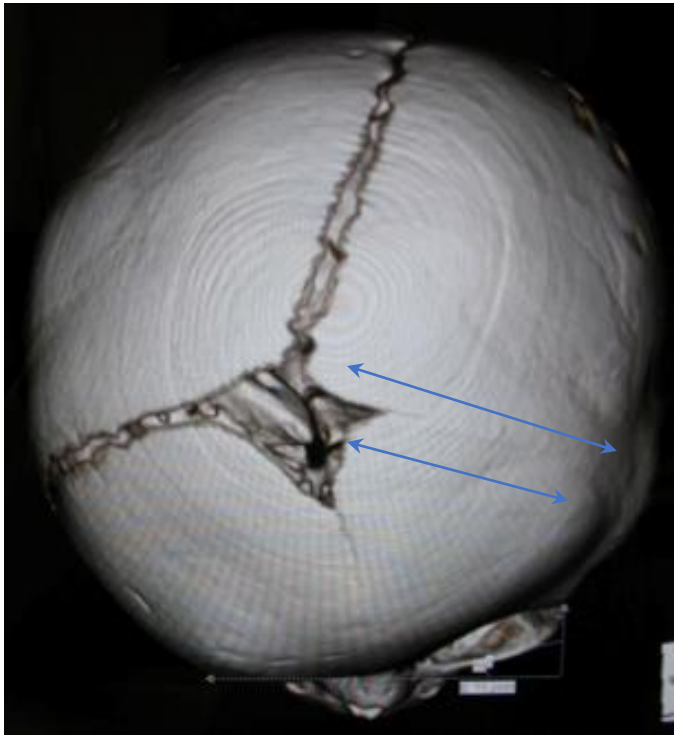


Sagittal Synostosis Result

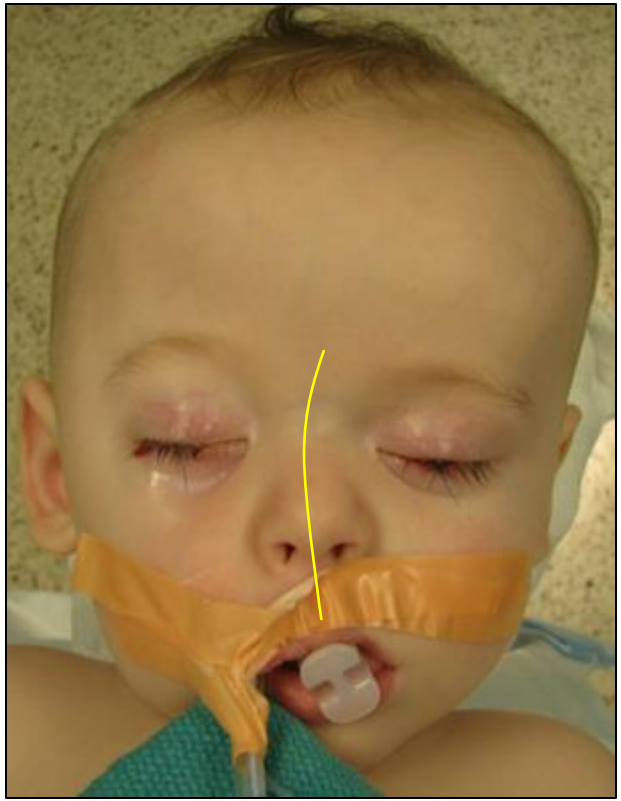
Sagittal Synostosis Result

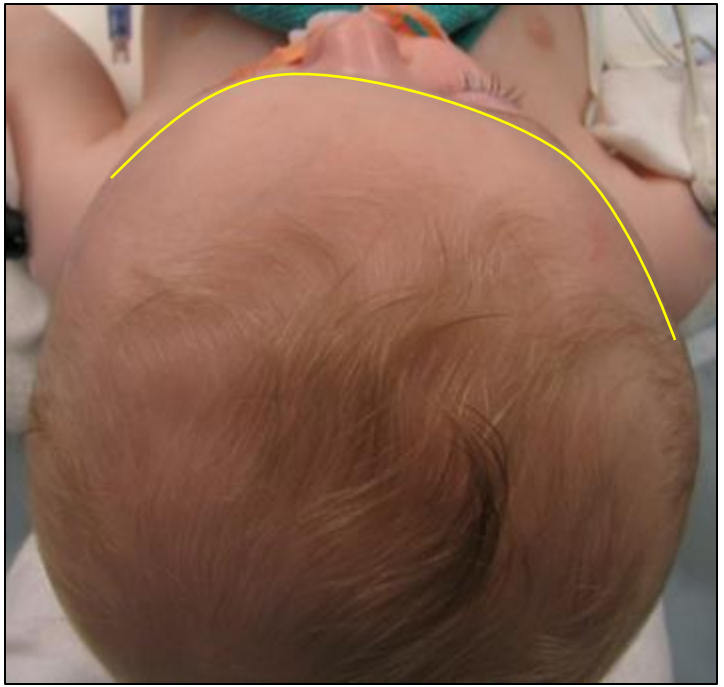
Unicoronal Synostosis

- Typical findings
  - Ipsilateral forehead retrusion
  - Contralateral forehead bossing
  - Facial scoliosis
  - Ipsilateral ear posteriorly displaced
- Most challenging reconstructive case

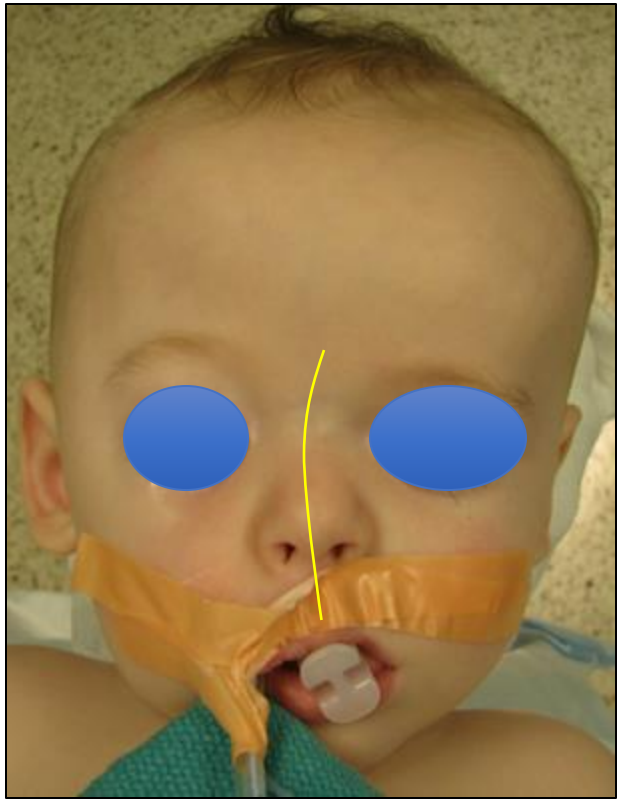


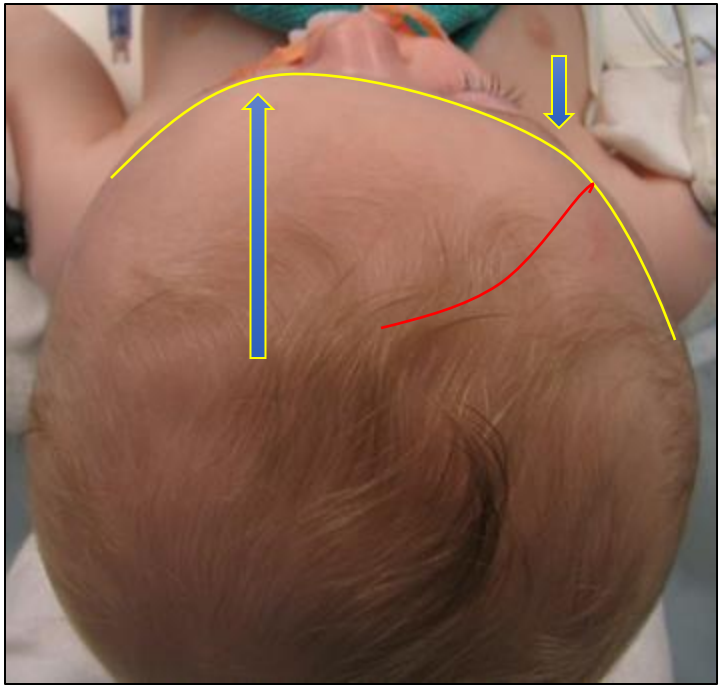
Unicoronal Synostosis



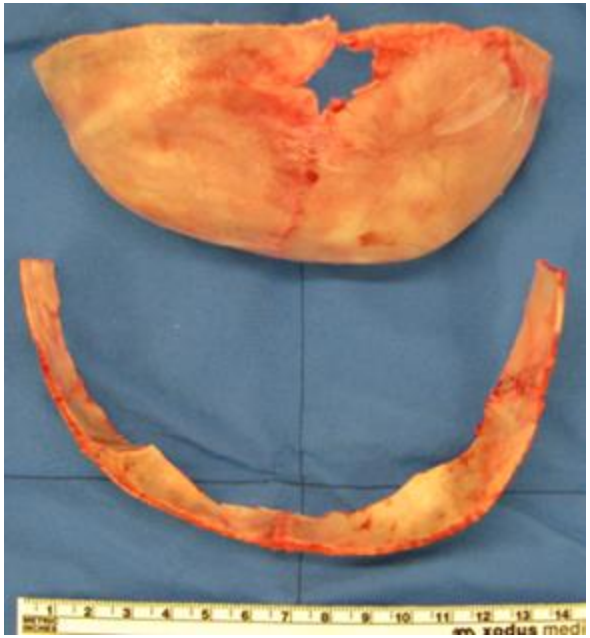


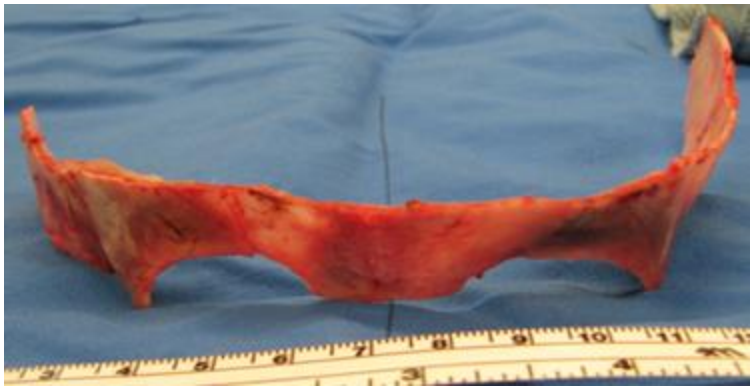
Unicoronal Synostosis



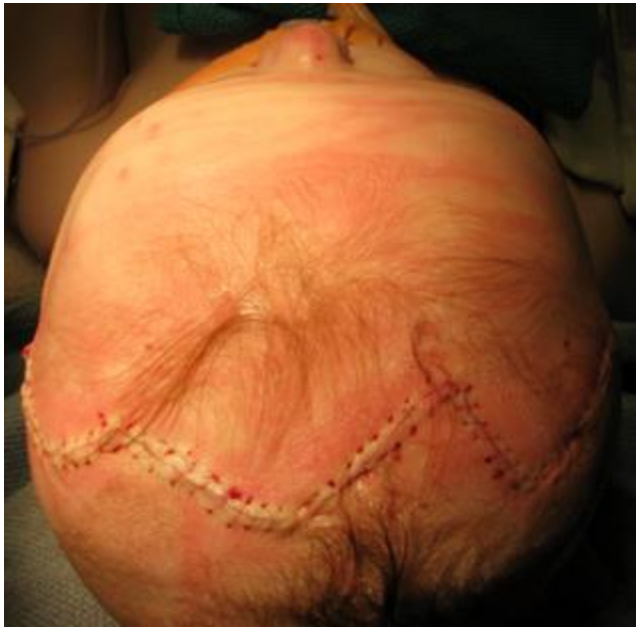


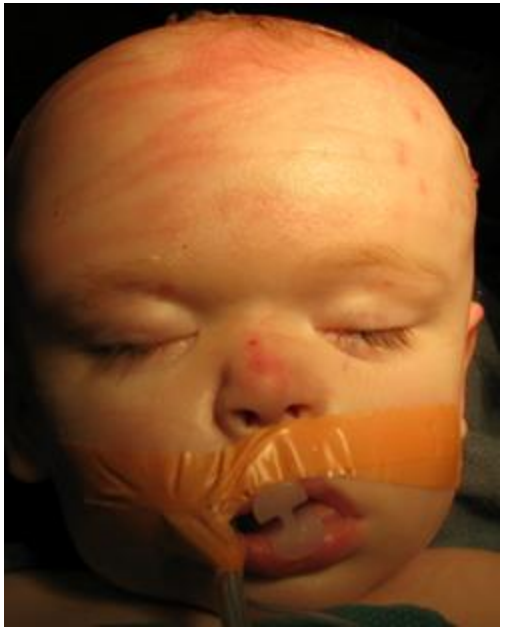
Unicoronal Synostosis



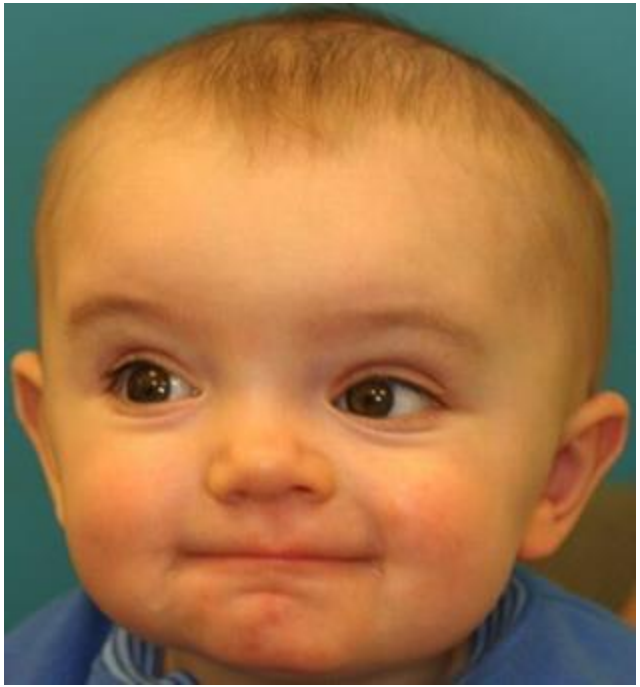


Unicoronal Synostosis

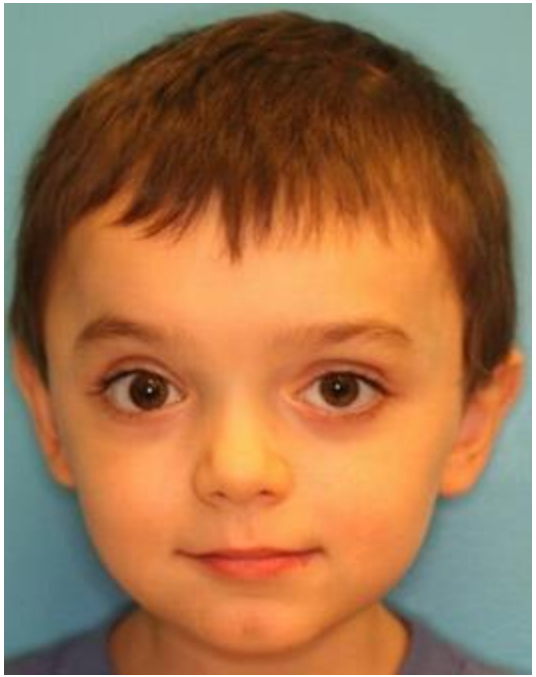




Unicoronal Synostosis Result

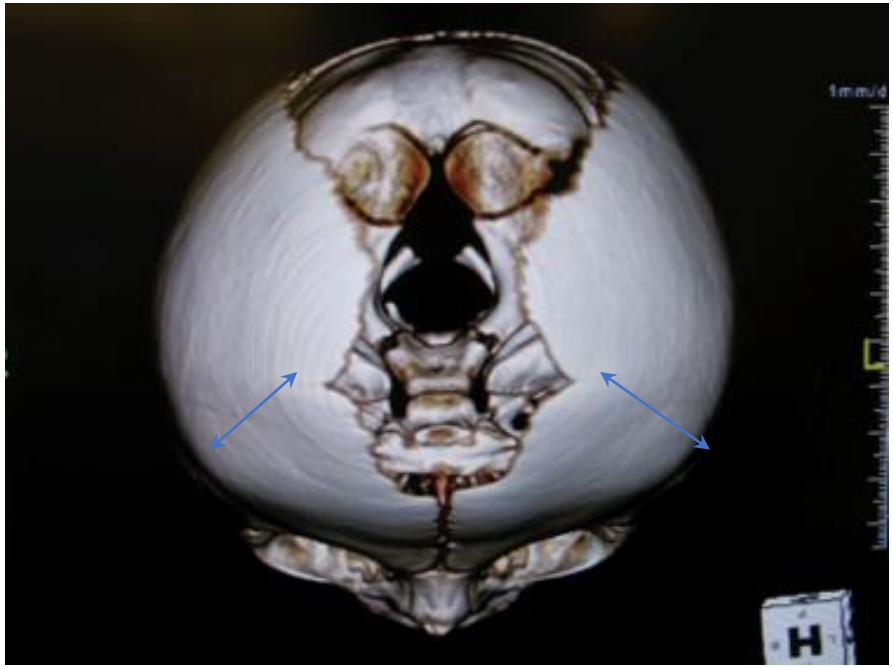


Unicoronal Synostosis Result

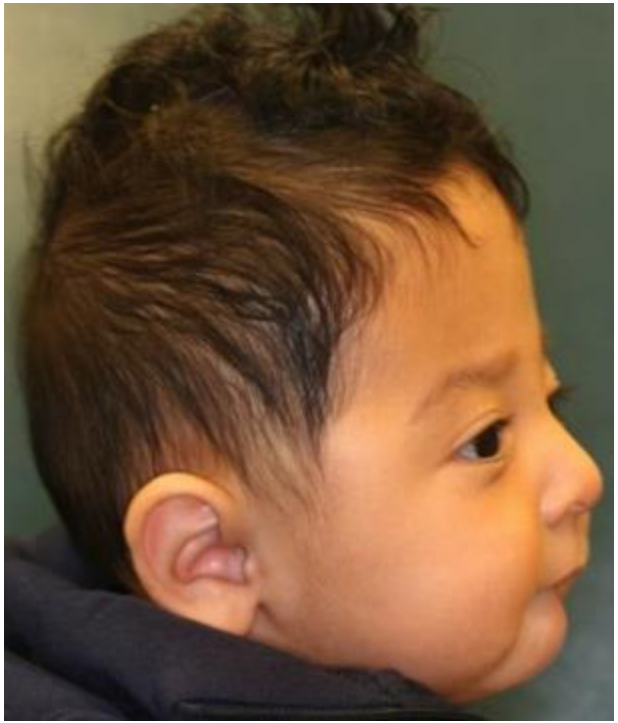


Bicoronal Synostosis

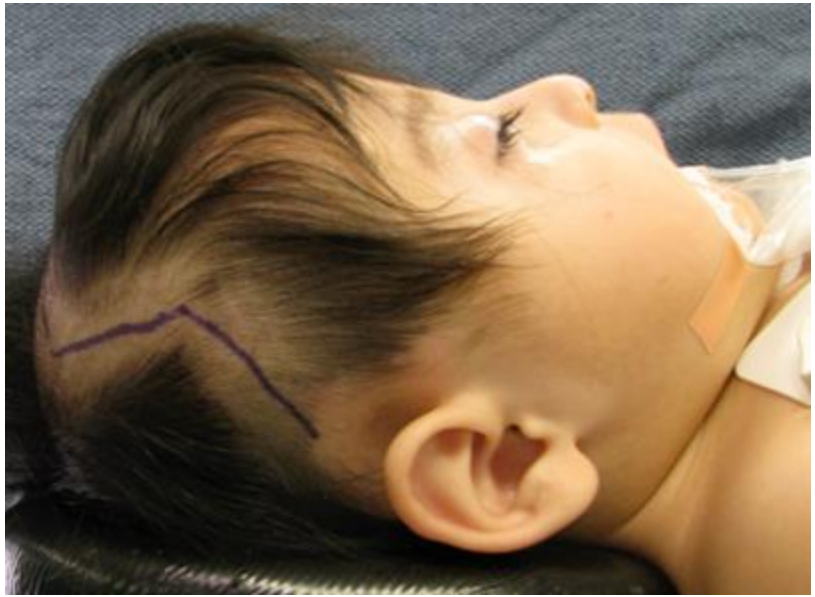
- Typical findings
  - Coronal sutural ridging
  - Shortened A-P distance
  - Increased transverse width
  - Turricephaly (tall skull)
- Most common synostosis associated with a syndrome



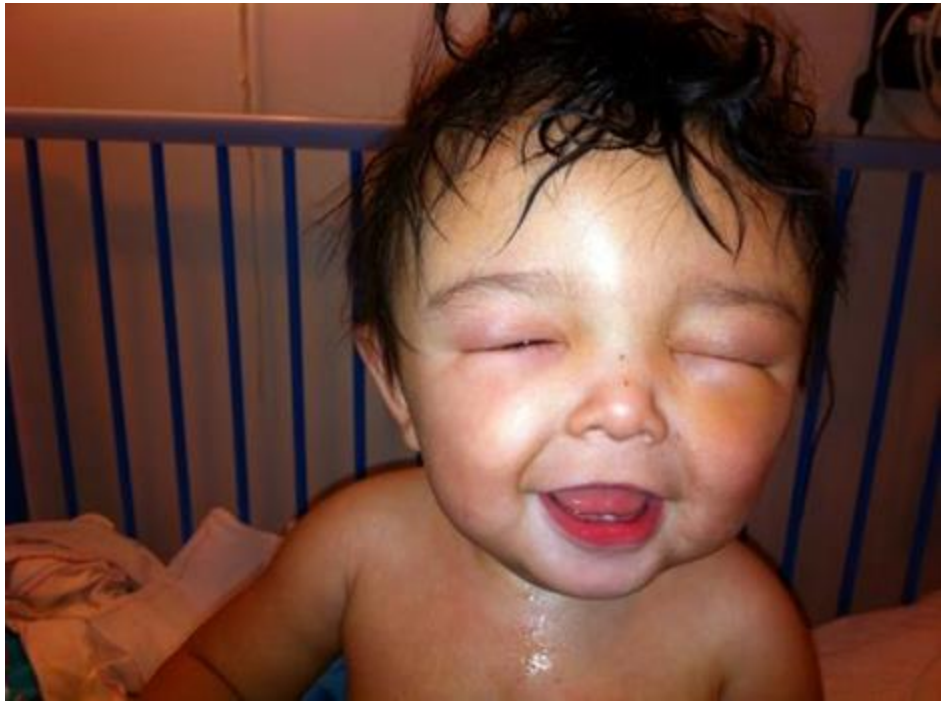
Bicoronal Synostosis



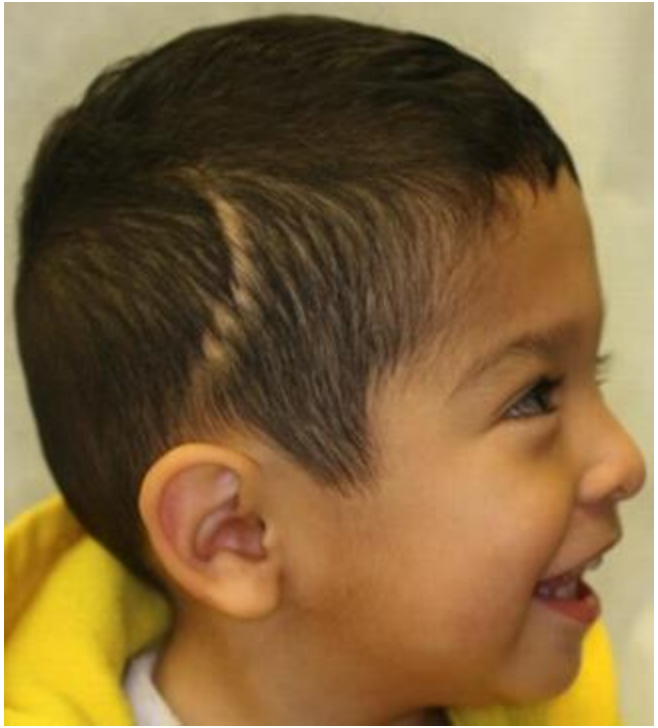
Bicoronal Synostosis



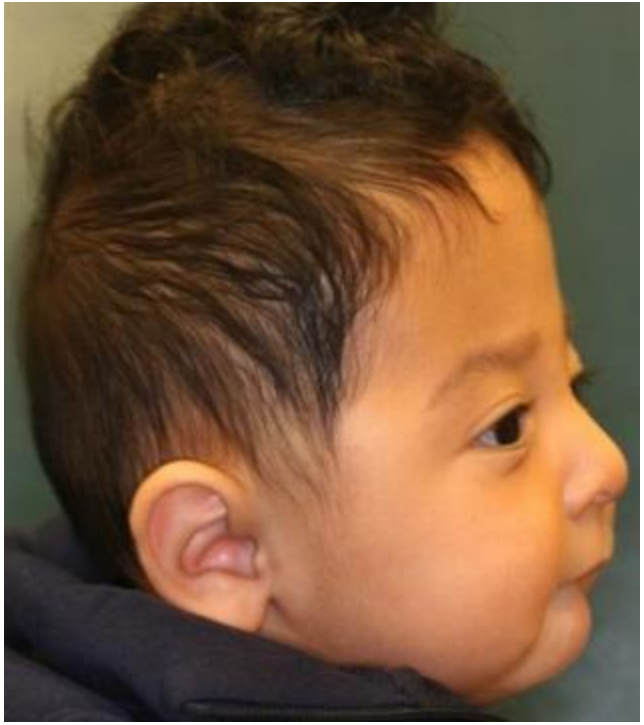
Two Days Postoperative

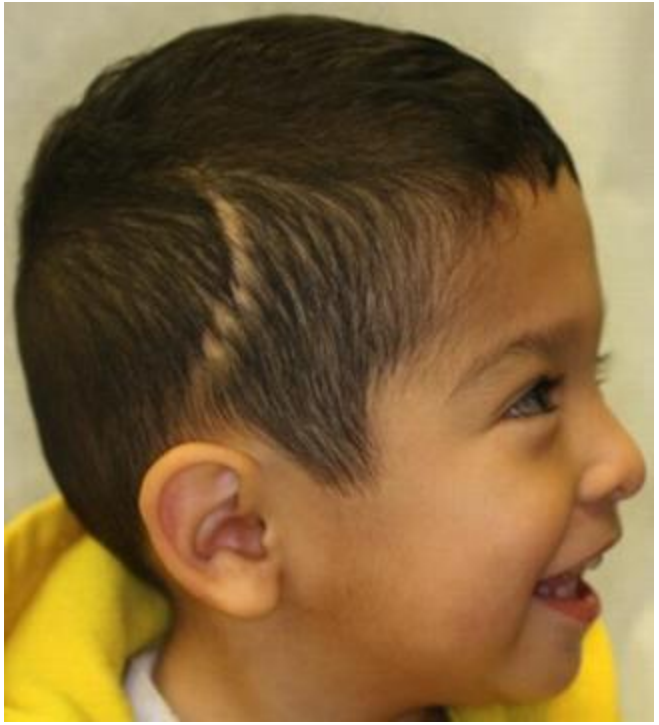


Bicoronal Synostosis Result



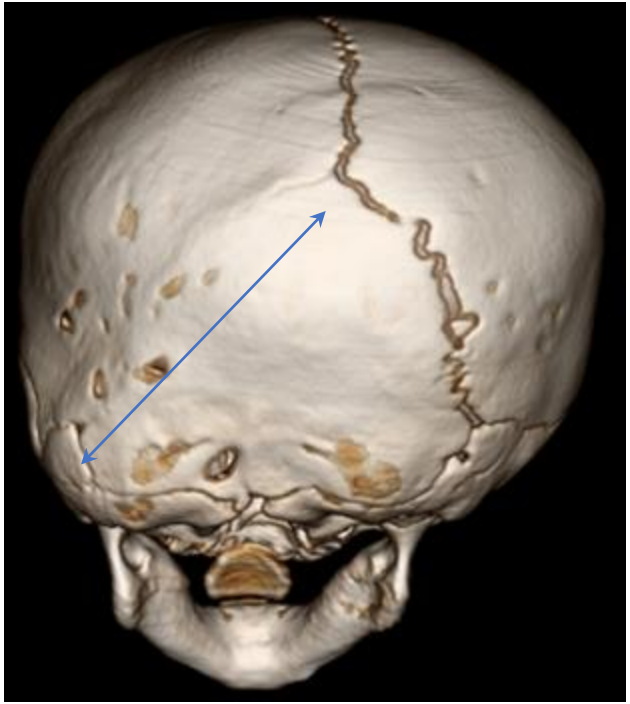
Bicoronal Synostosis Result





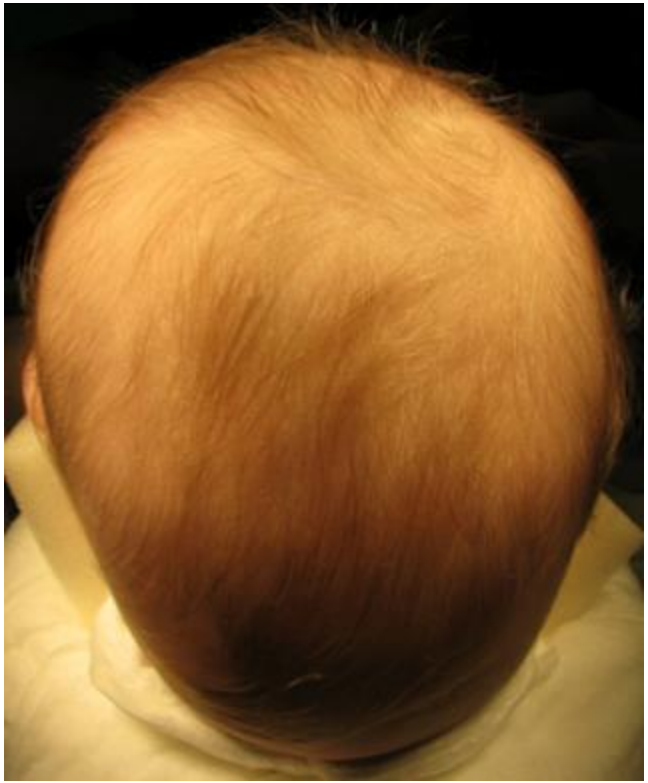
Lambdoid Synostosis

- Typical findings
  - Ipsilateral occipital flattening
  - Contralateral occipital bossing
  - *Posterior* displacement of *ipsilateral* ear
  - Possible frontal bone bossing
- Least common synostosis

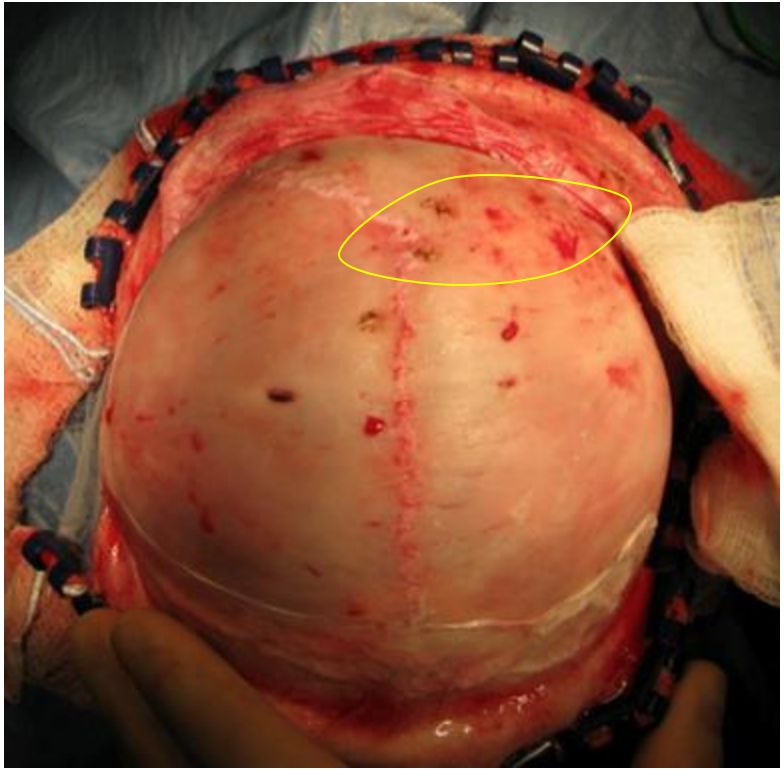


Lambdoid Synostosis

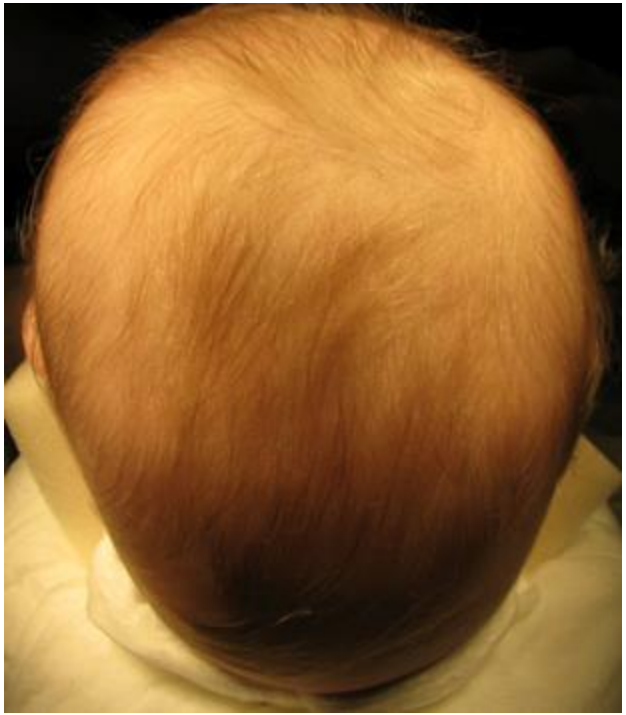
- Patient must be positioned prone
  - Padding key
  - Does not allow for forehead correction at the same time/same position

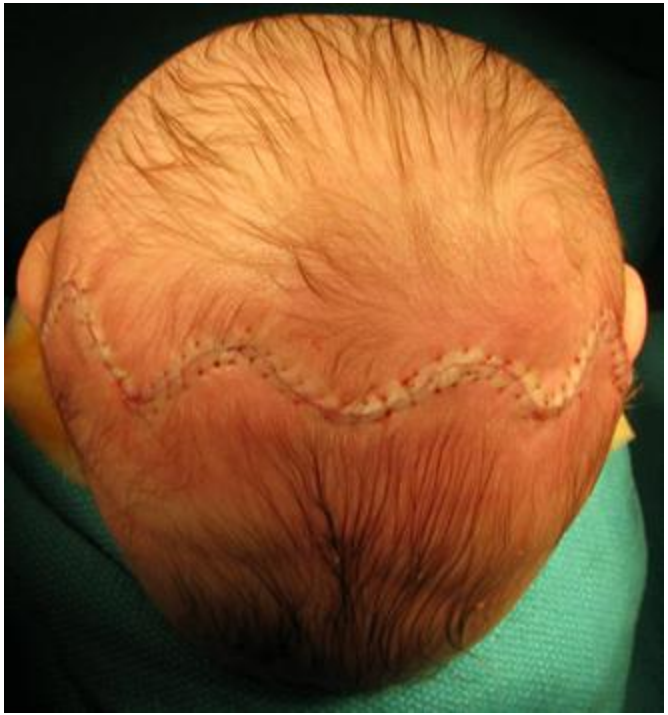


Lambdoid Synostosis



Lambdoid Synostosis

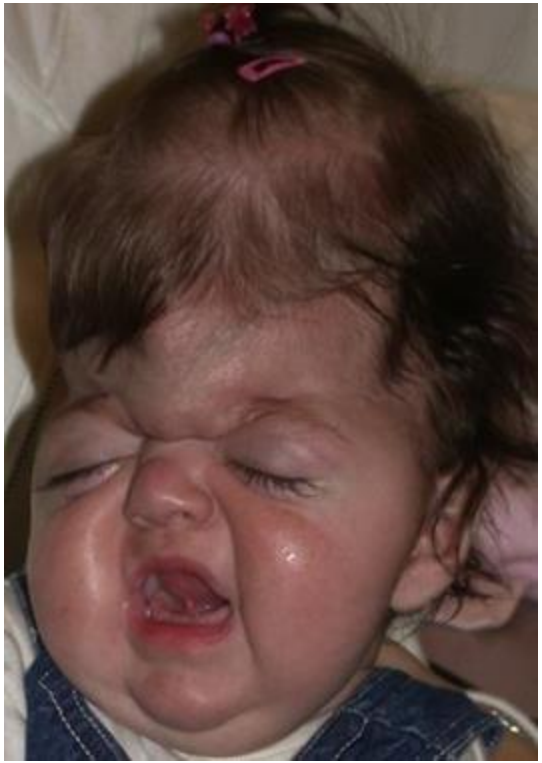




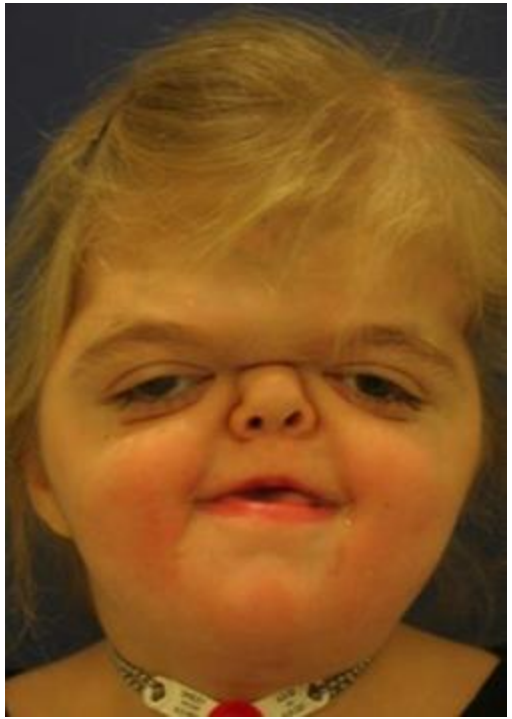
Syndromic Synostosis

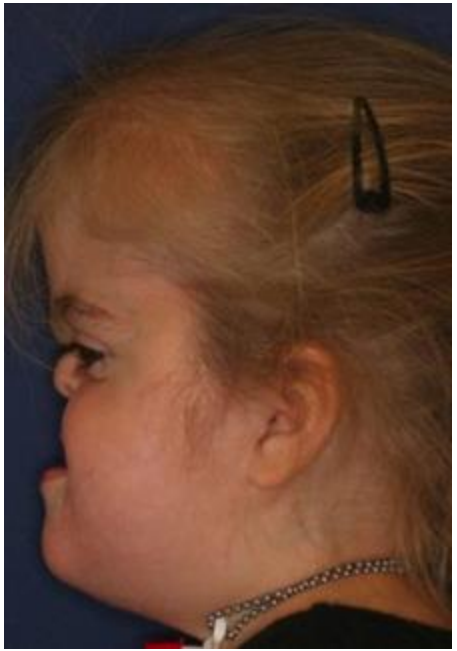
- Cranial dysostosis
  - Apert
  - Crouzon
  - Pfeiffer
  - Saethre-Chotzen
- Most commonly associated with bicoronal synostosis

Apert Syndrome

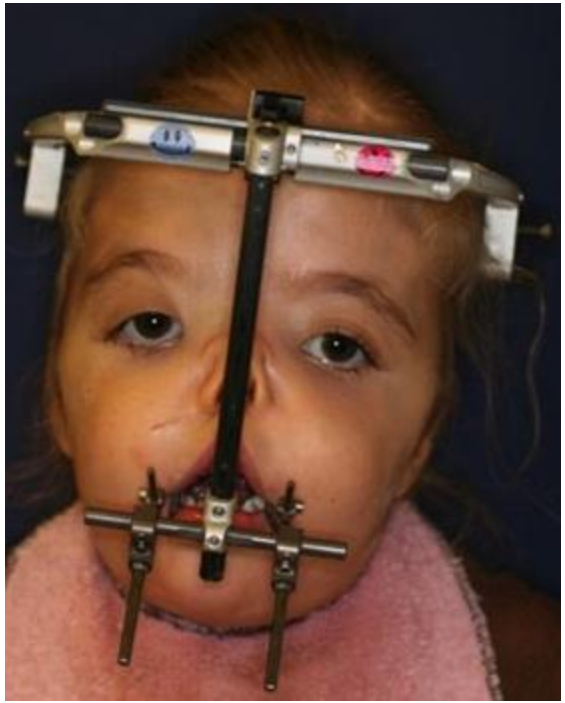


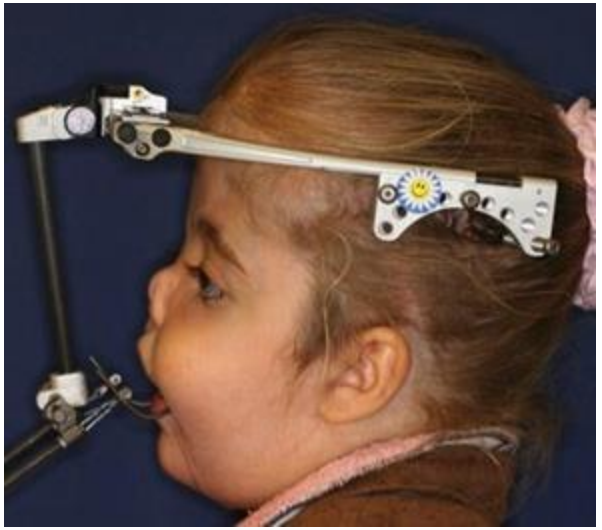
Apert Syndrome Result



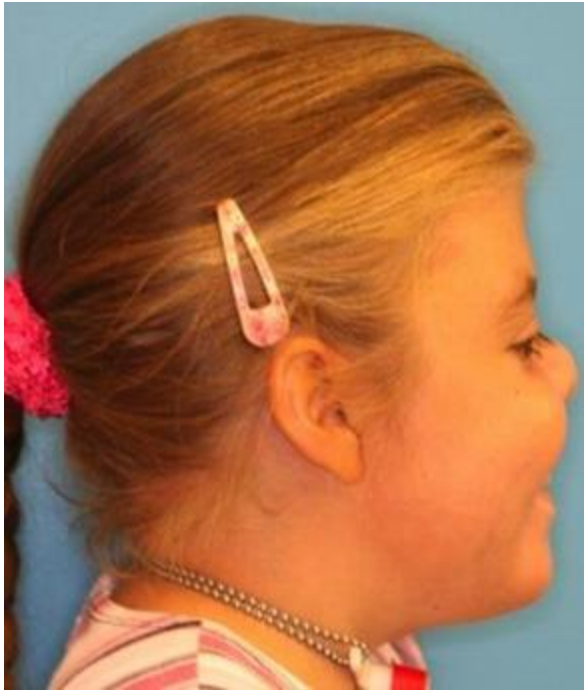


Apert Syndrome – Midface Distraction

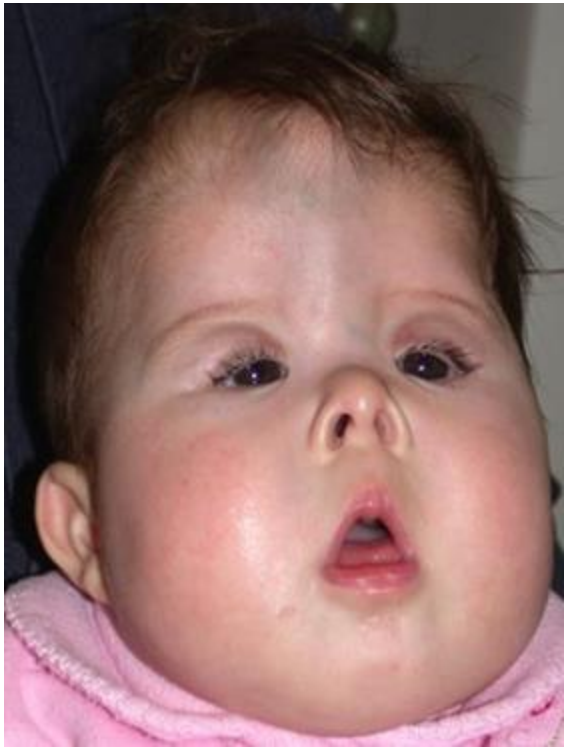




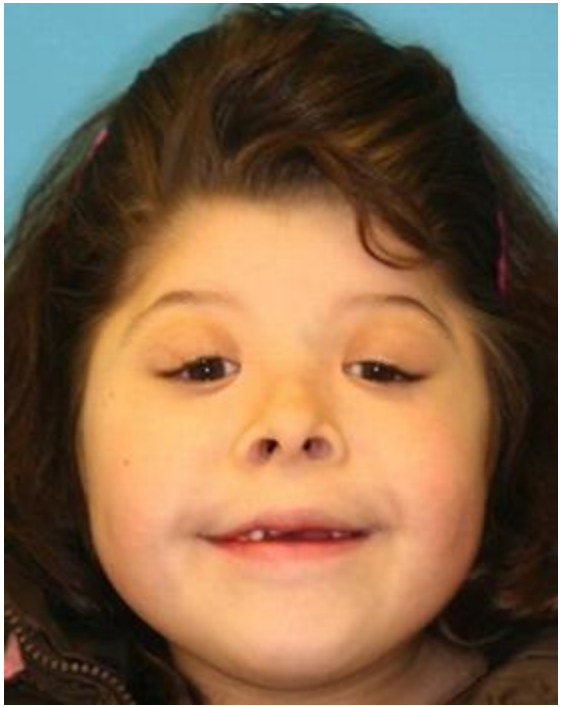
Apert Syndrome Result

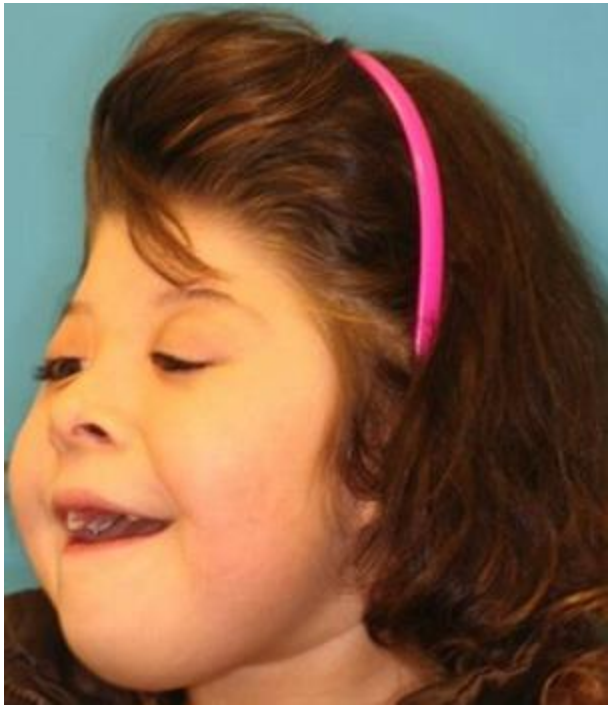


Saethre-Chotzen Syndrome



Saethre-Chotzen Syndrome Result

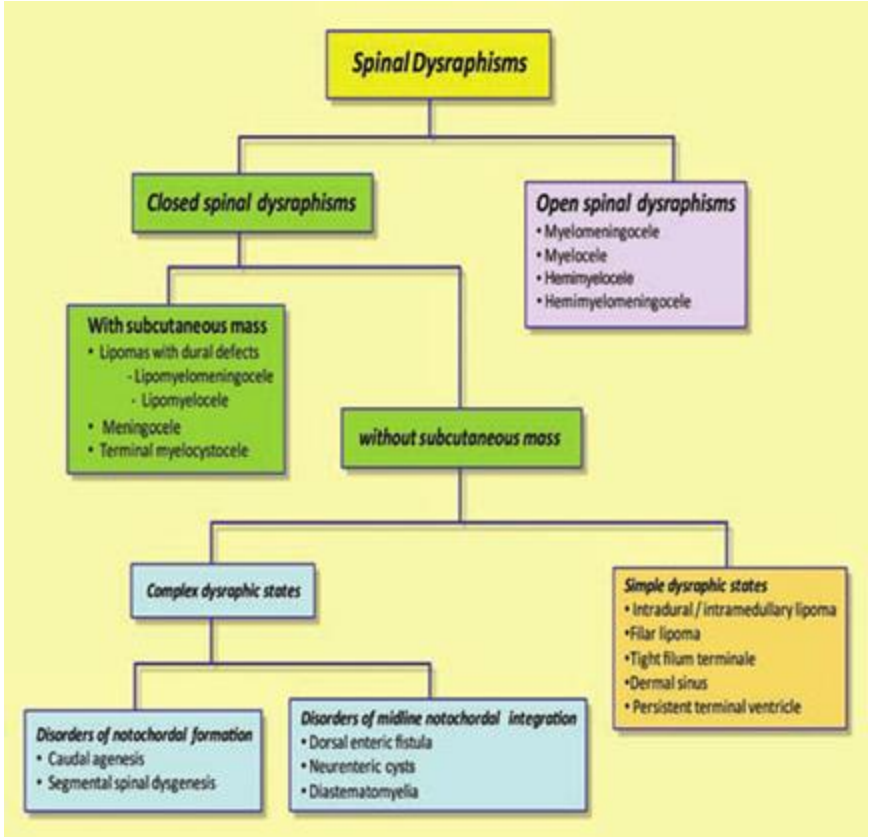




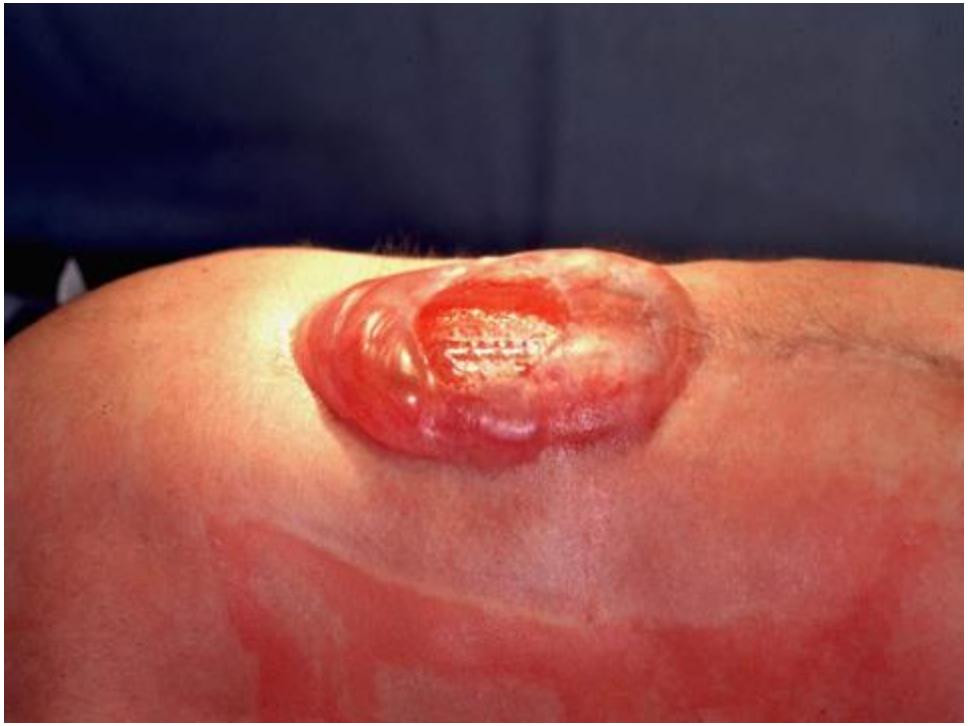
Craniospinal Dysraphism

- Classification: open vs occult spinal dysraphism
- Myelomeningocele
- Occult dysraphism: lipomyelomeningocele, dermal sinus tract, tethered cord syndrome

Craniospinal Dysraphism

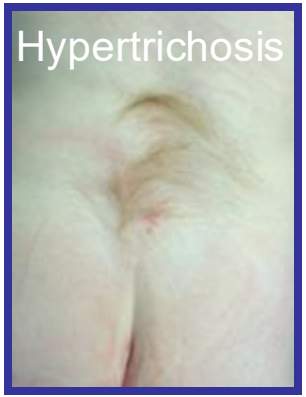


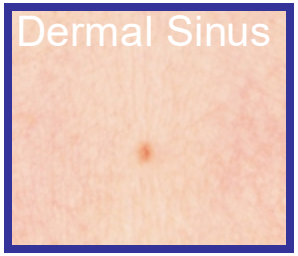
Open Spinal Dysraphism

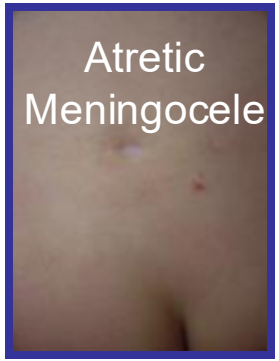


Spina Bifida Occulta

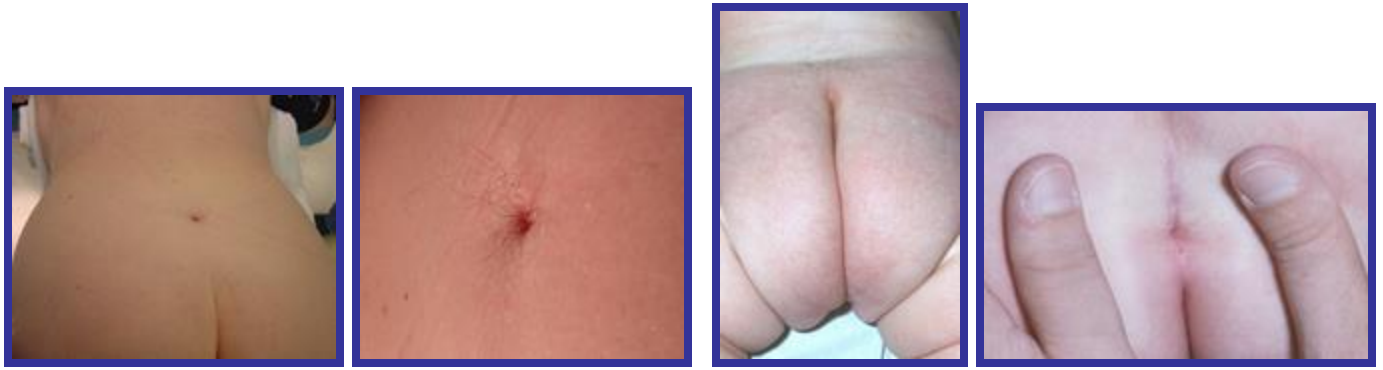
Skin Anomalies (60% of Patients)





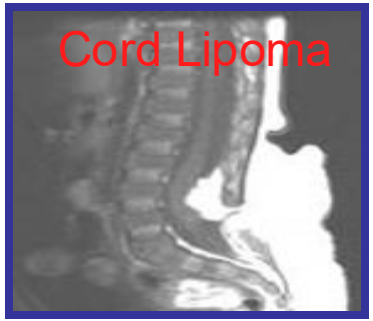


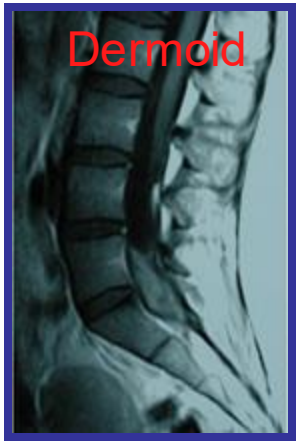
- Above intergluteal fold
- Other cutaneous stigmata

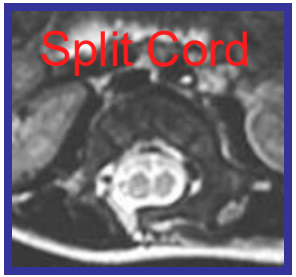


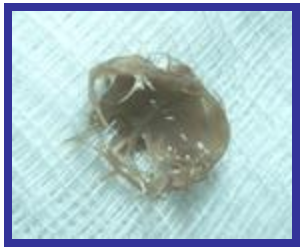
Located in the intergluteal fold
Tip of the coccyx
Pointing downward

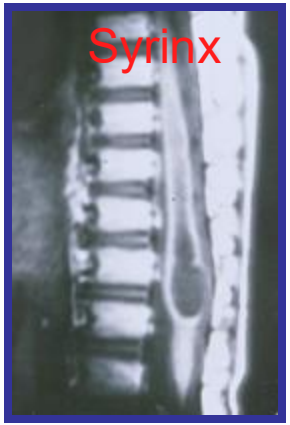
Spina Bifida Occulta
Spinal Anomalies











Conclusion

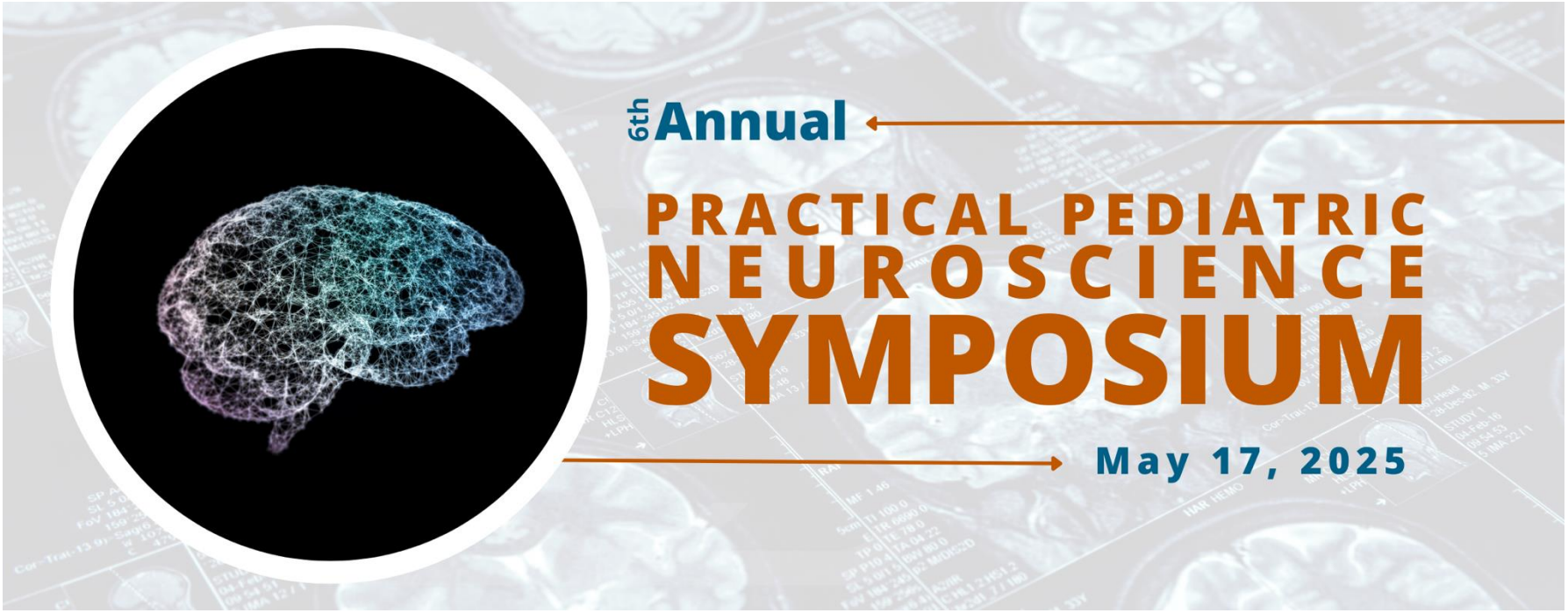
- Importance of early recognition and multidisciplinary care
- Advances in imaging and minimally invasive surgical techniques
- Ongoing research and emerging therapies


THANK YOU

The University of Texas at Austin
Dell Medical School

dell children's
Ascension

UT Health Austin Pediatric Neurosciences at Dell Children's



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