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The Sydney children's Hospitals Network

Pediatric Craniofacial Disorders: Diagnosis

2023 AUSTRALASIAN SET REGISTRARS CONFERENCE

Christopher R. Forrest, MD, MSc, FRCS(C), FACS, FAAPS

Staff Surgeon

Division of Plastic Surgery

Sydney Children's Hospital Network

Land Acknowledgment

"I would like to acknowledge the Gadigal of the Eora Nation, the traditional custodians of this land and pay my respects to the Elders both past and present."

Program

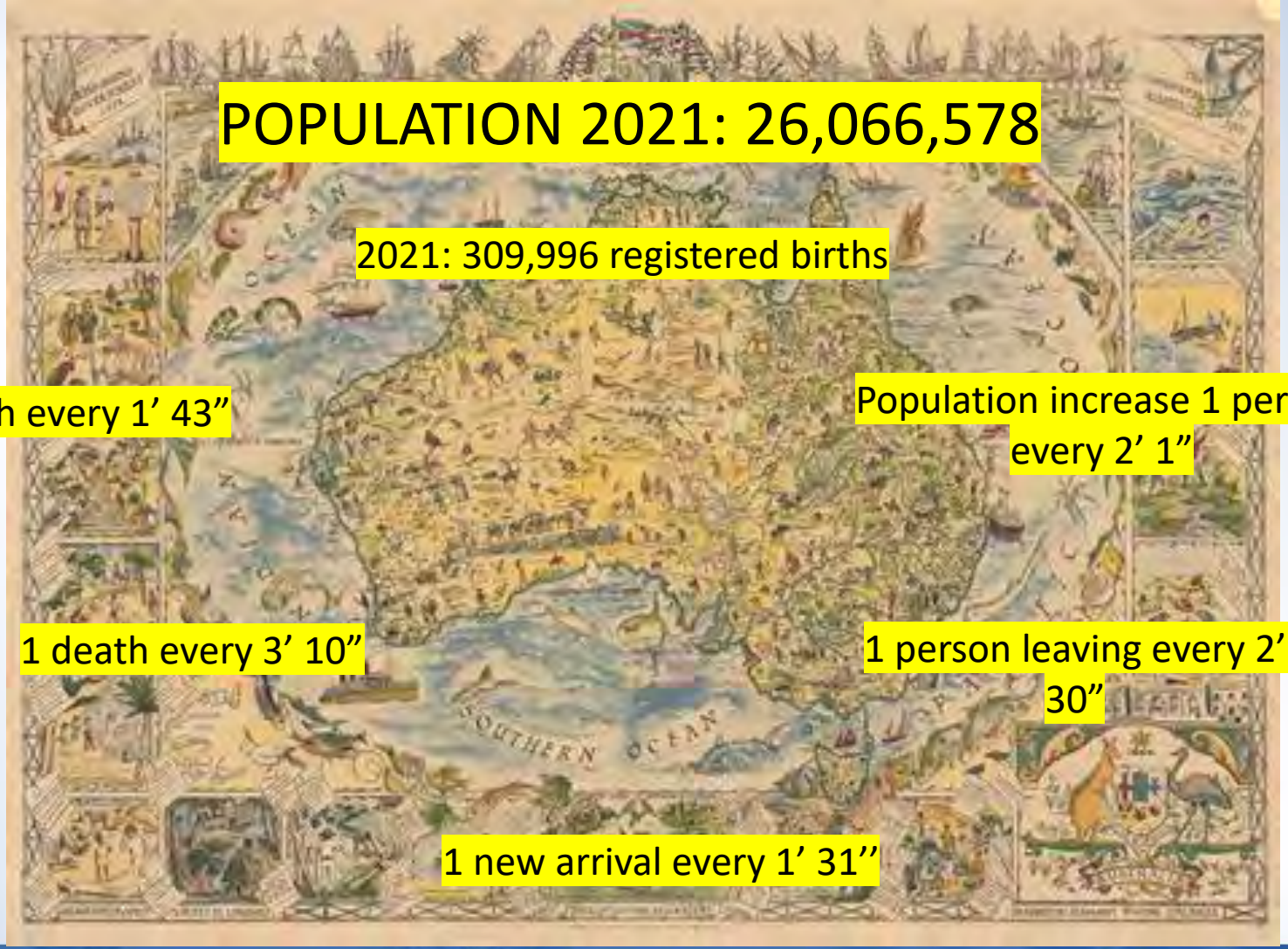
2023 Australasian SET Registrars Conference

4-9 MARCH

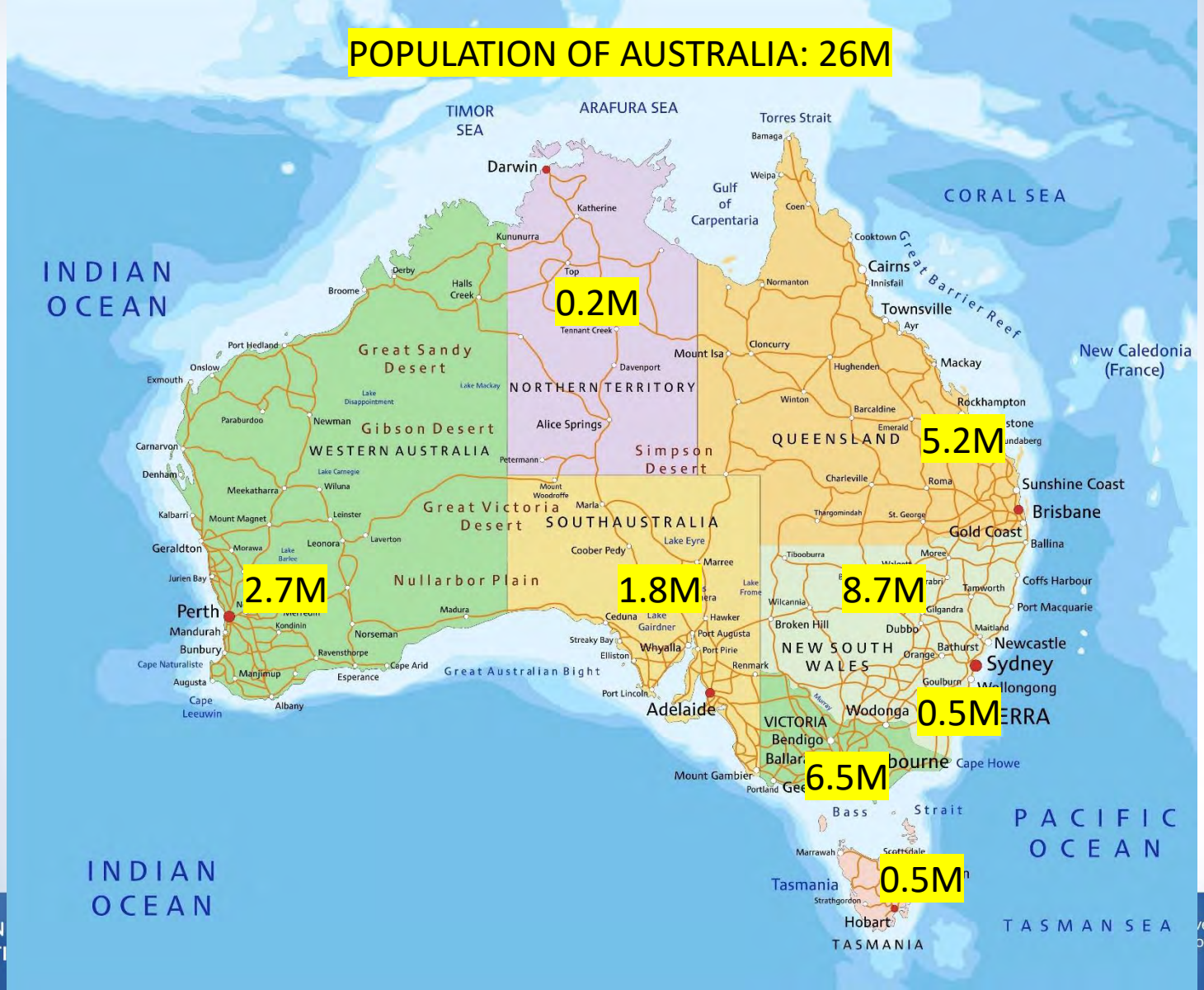
10:30-12:30	Formal lectures on **Craniofacial** *continued*	*Chris Forrest*
10:30	- Non syndromic craniosynostosis and positional plagiocephaly	
11:00	- Syndromic craniosynostosis	
11:30	- Time line management of craniosynostosis	
12:00	- Management of craniosynostosis	

Australian Society *of* Plastic Surgeons

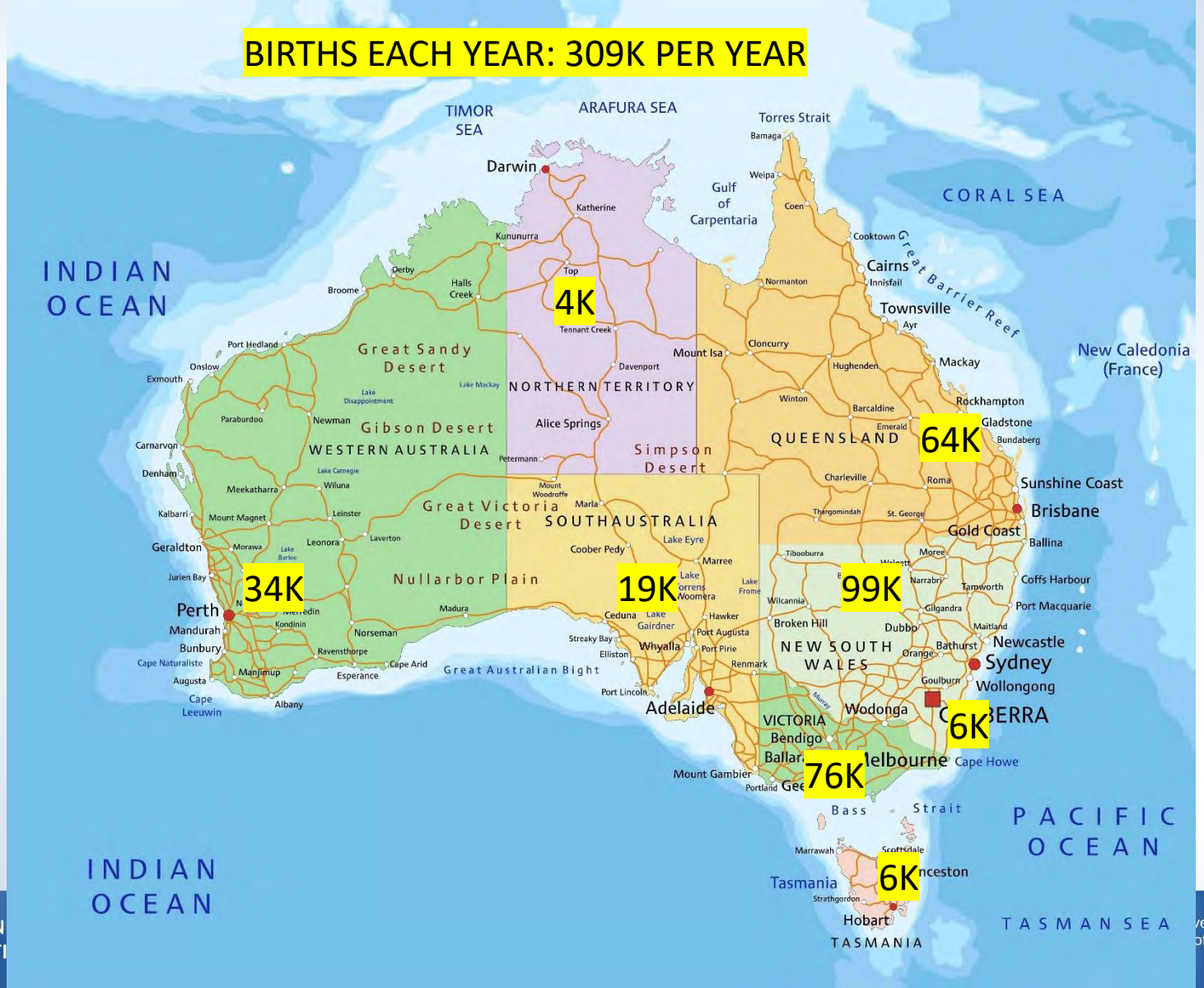
NEW ZEALAND ASSOCIATION *of* Plastic Surgeons
Te Kāhui Whakamōhou Kiri


POPULATION 2021: 26,066,578
2021: 309,996 registered births
I birth every 1’ 43”
Population increase 1 person every 2’ 1”
1 death every 3’ 10”
1 person leaving every 2’ 30”
1 new arrival every 1’ 31’’
SOUTHERN OCEAN

POPULATION OF AUSTRALIA: 26M



BIRTHS EACH YEAR: 309K PER YEAR



Epidemiology: Australia: 2021

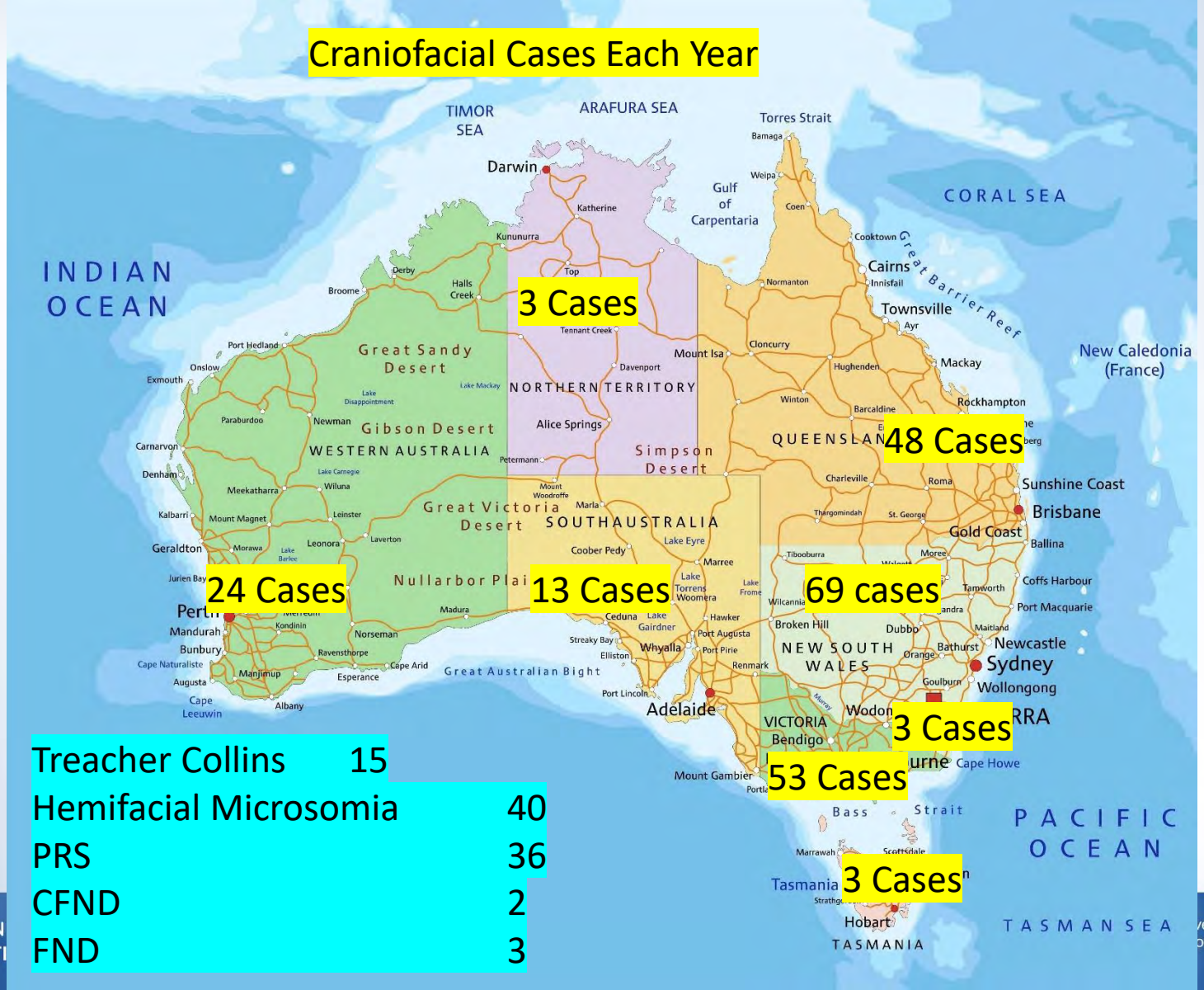
- 26 M people; 309K births

Syndrome	Prevalence	Expected Number per year
Saggital synostosis	1 :2,500	124
Metopic s		
Unicorona		
Bicoronal		
Lambdoid synostosis	1:60,000	5
Crouzon syndrome	1:60,000	5
Apert syndrome	1:80,000	4
Pfeiffer syndrome	1:100,000	3
Saethre Chotzen syndrome	1:50,000	7
Muenke syndrome	1:30,000	10

187 cases non-syndromic CS/year

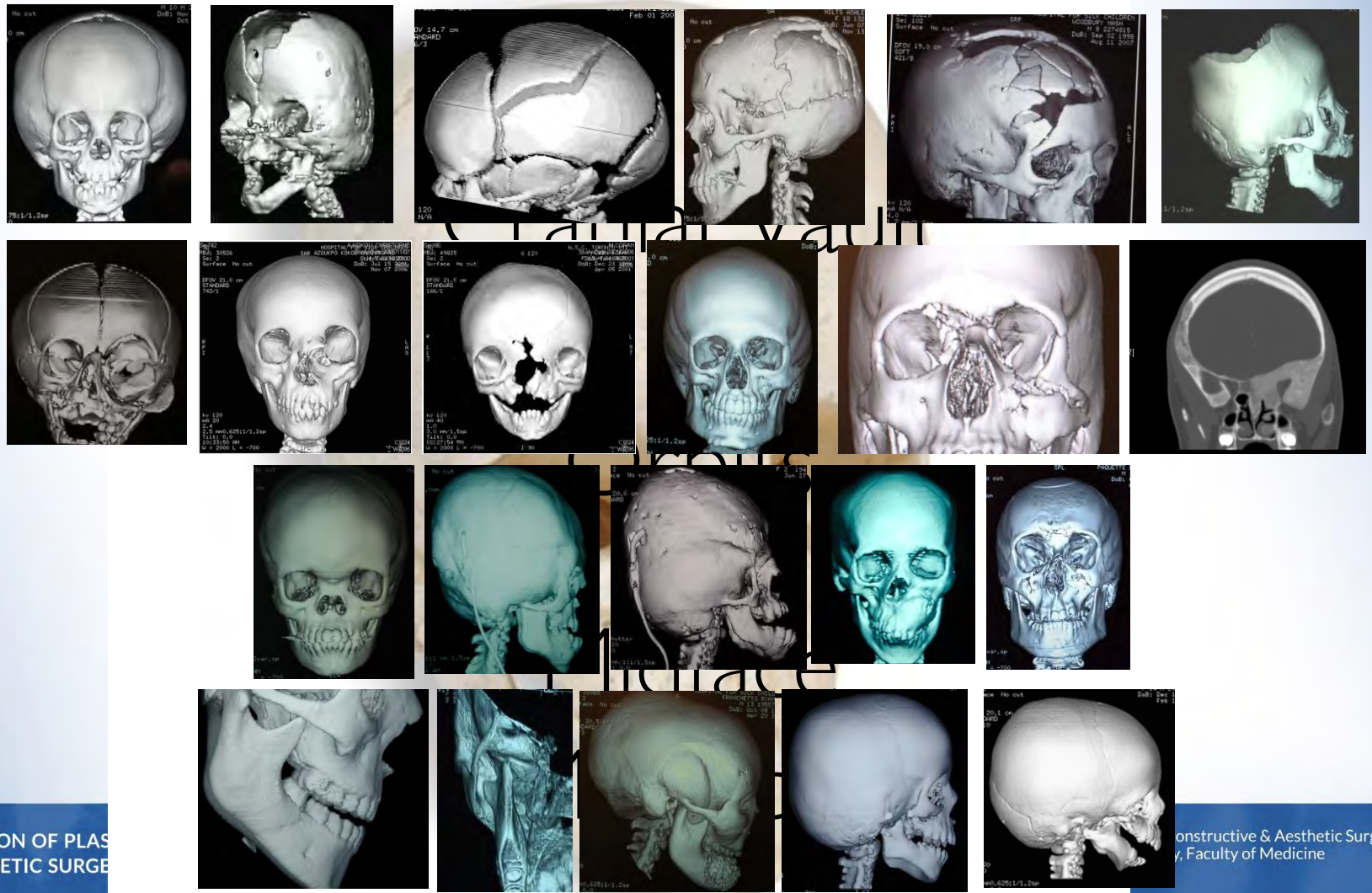
29 cases syndromic CS/year

Craniofacial Cases Each Year

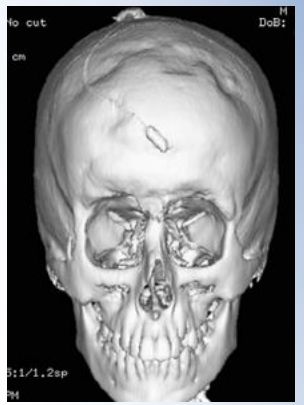


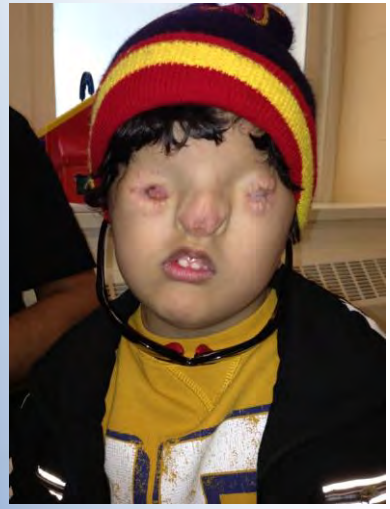
Treacher Collins	15
Hemifacial Microsomia	40
PRS	36
CFND	2
FND	3

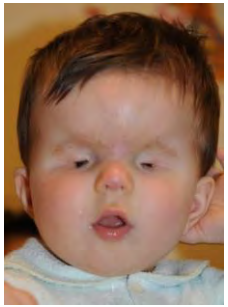
Pediatric Craniofacial Surgery: Scope

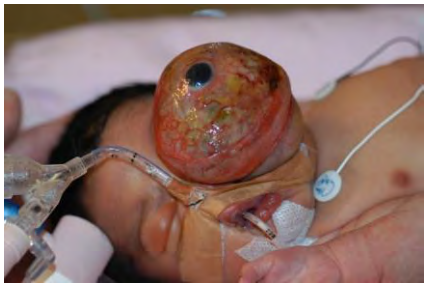


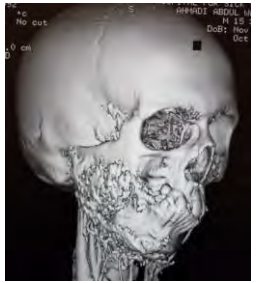
Pediatric Craniofacial Reconstruction: Miscellany and Unusual Conditions

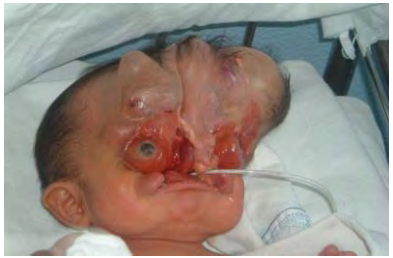


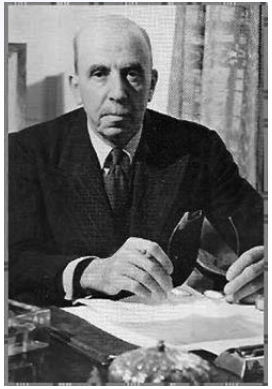












Sir Harold D. Gilles: 1882-1960

OPERATIVE CORRECTION BY OSTEOTOMY OF RECESSED MALAR MAXILLARY COMPOUND IN A CASE OF OXYCEPHALY

By Sir Harold Gillies, C.B.E., F.R.C.S., and
Stewart H. Harrison, F.R.C.S., L.D.S., R.C.S.
From the Plastic and Jaw Unit, Rookisdown House, Park Prewett Hospital, Basingstoke

Ætiology

Oxycephaly is a condition of cranial dysostosis produced by premature synostosis of the coronal suture. The effect of early closure of the coronal suture is to produce a "turret-shaped skull," thus the condition is sometimes called turricephaly.

This deformity is often associated with some degree of hydrocephalus. The raised intracranial pressure may give rise to headaches which are a common feature of the disease. Cerebral symptoms may be present. The prominence of the eyes is due to the forward projection of the great wing of the sphenoid which accompanies expansion of the middle cranial fossa. The deformity of the sphenoid may also give rise to a narrowing of the optic canal and defective vision (Thoma, 1946). Recession of the maxilla gives the appearance of an apparent prognathism. It is often associated with a high-vaulted palate due to premature synostosis of the vomer and palate. Separation of the eyes may occur secondarily.

Oxycephaly is said to be familial, although this is often difficult to elicit as minor degrees of this deformity are apt to pass unrecognised. A Mendelian dominant is said to be the causative factor (Ellis, 1948).

Diagnosis

Early diagnosis can be made radiologically. The synostosis can be seen, and digital markings, the result of raised intracranial pressure, can often be identified. A raised cerebrospinal fluid pressure may also be present.

Case Report

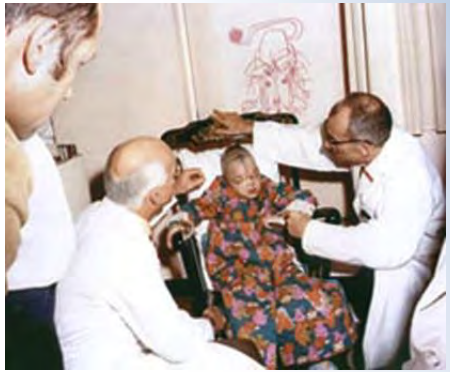
A female, aged 14, was referred to H. D. G. in 1942. She was suffering from the condition of oxycephaly, and presented the following features :—

The head was turret-shaped, and there was a marked retrusion of the upper jaw, giving rise to an apparent prognathism. The degree of retrusion was so marked that the tip of the nose nearly touched the upper lip. The nose itself was asymmetrically developed. The palate was very high and narrow. The eyes were so prominent that the lids were well below the iris, and the patient was inclined to walk with her head down.

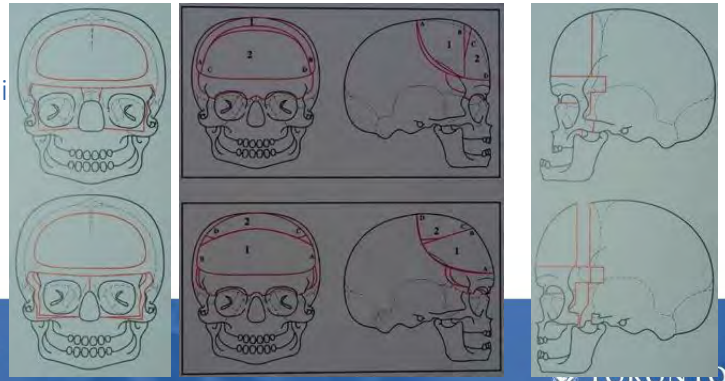
She did not complain of eye symptoms and there was no history of headaches. There was slight deafness, but no loss of the sense of smell. Mental development was within normal limits, and she was anxious to train as a nurse.

- "Grand Remedy" planned - 1942

Dr. Paul Tessier

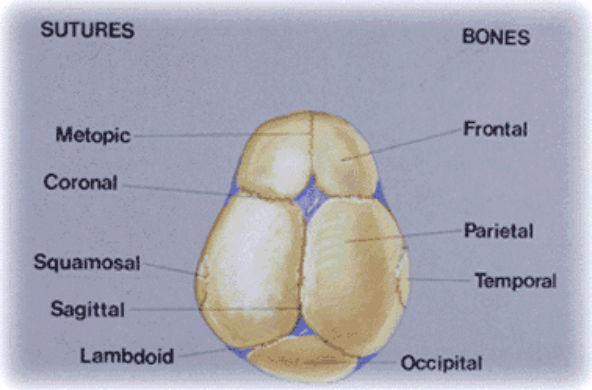


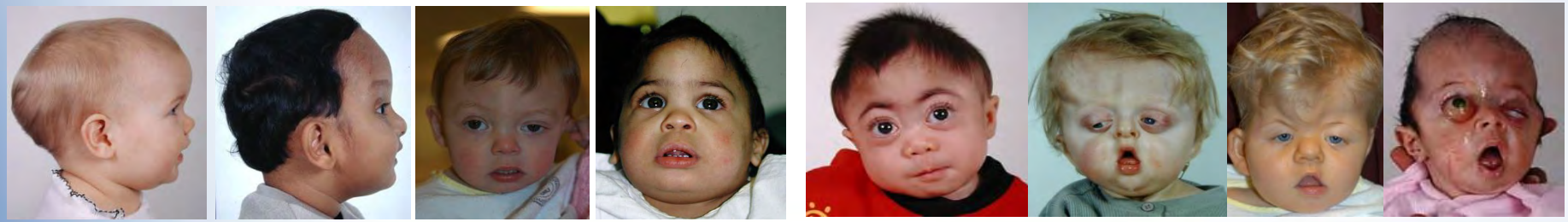
- Craniofacial Surgery
  - Cranial vault
    - ACVR
    - PCVR
    - TCVR
  - Fronto-orbital
    - FOA
  - Combined
    - Mono-bloc
    - Bipartition
  - Orbital
    - Intra-cranial/Extra-crani
    - Subcranial
  - Nasal
  - Midface
    - Lefort III
    - Lefort II



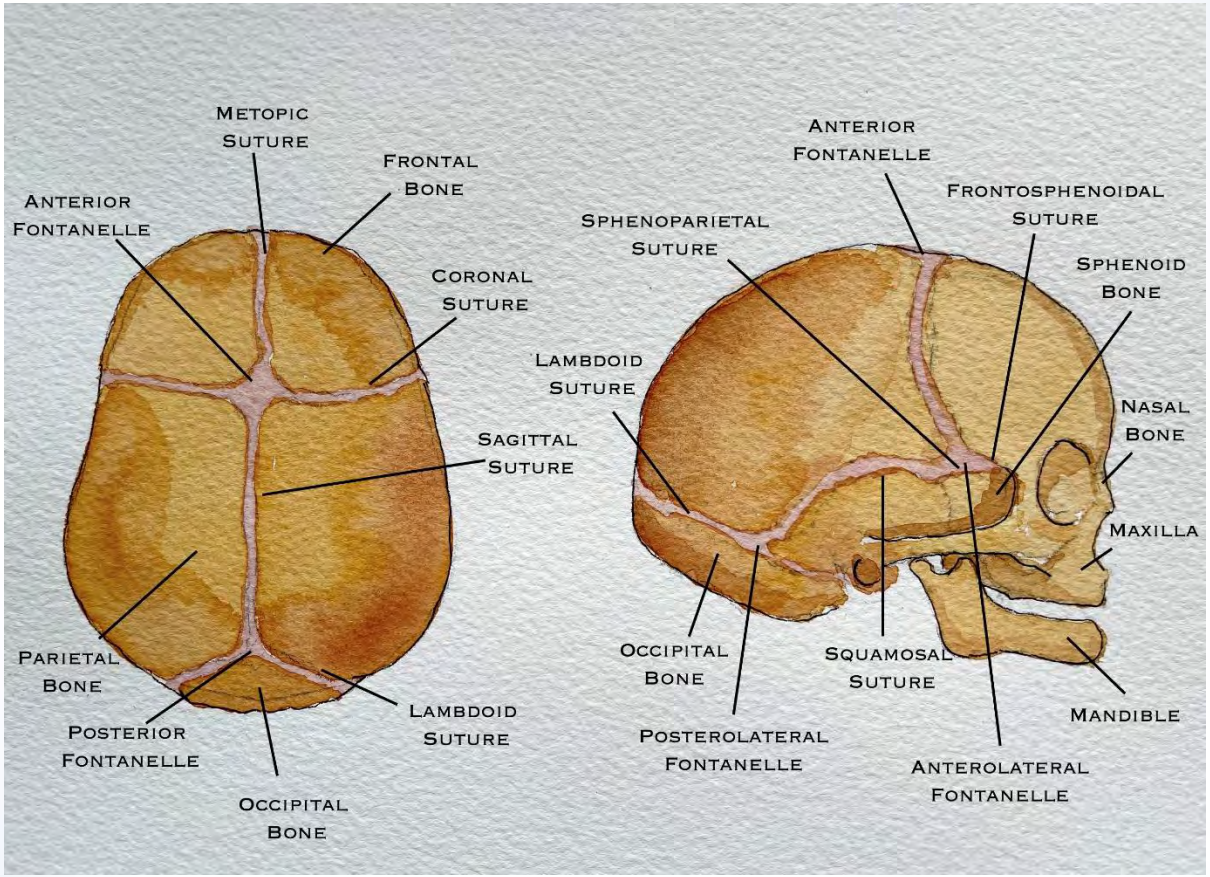
Craniosynostosis

Definition: Premature fusion of one or more sutures in cranial vault or base

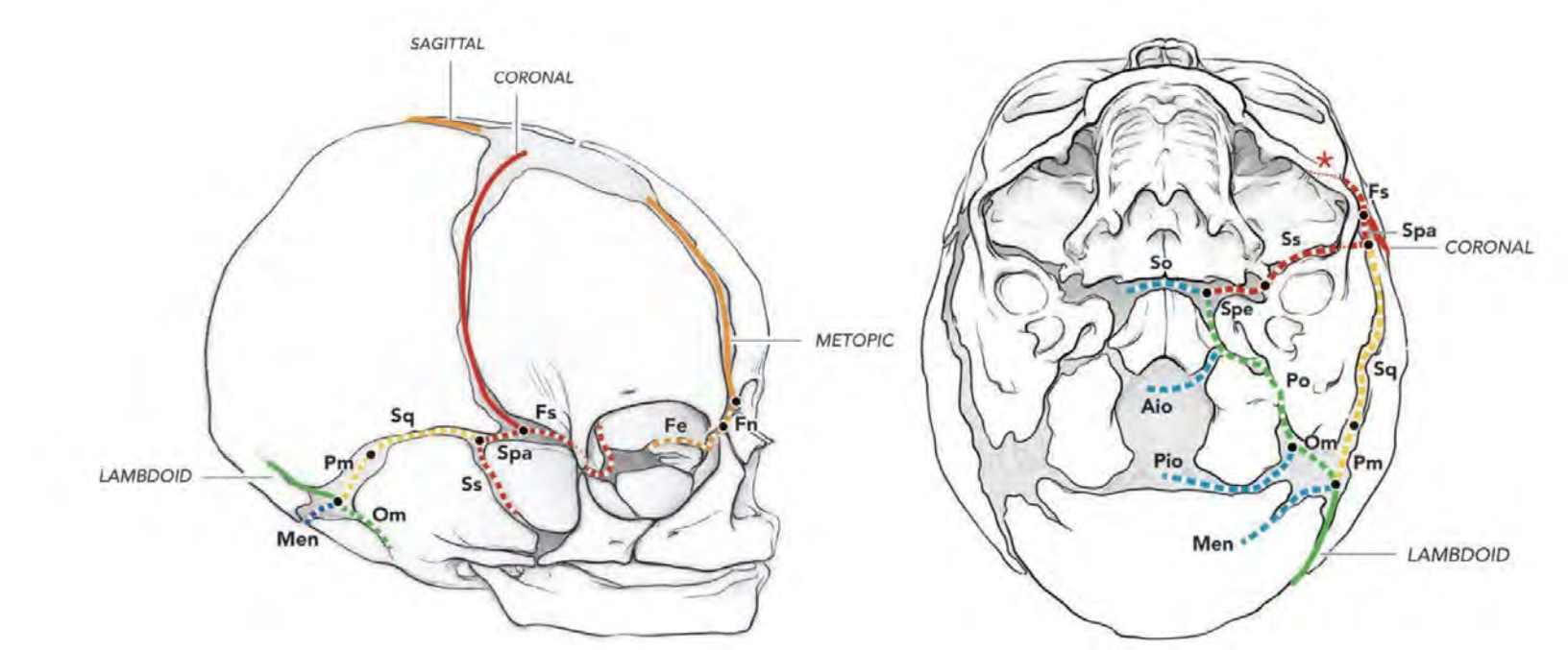




ANATOMY: MAJOR SUTURES

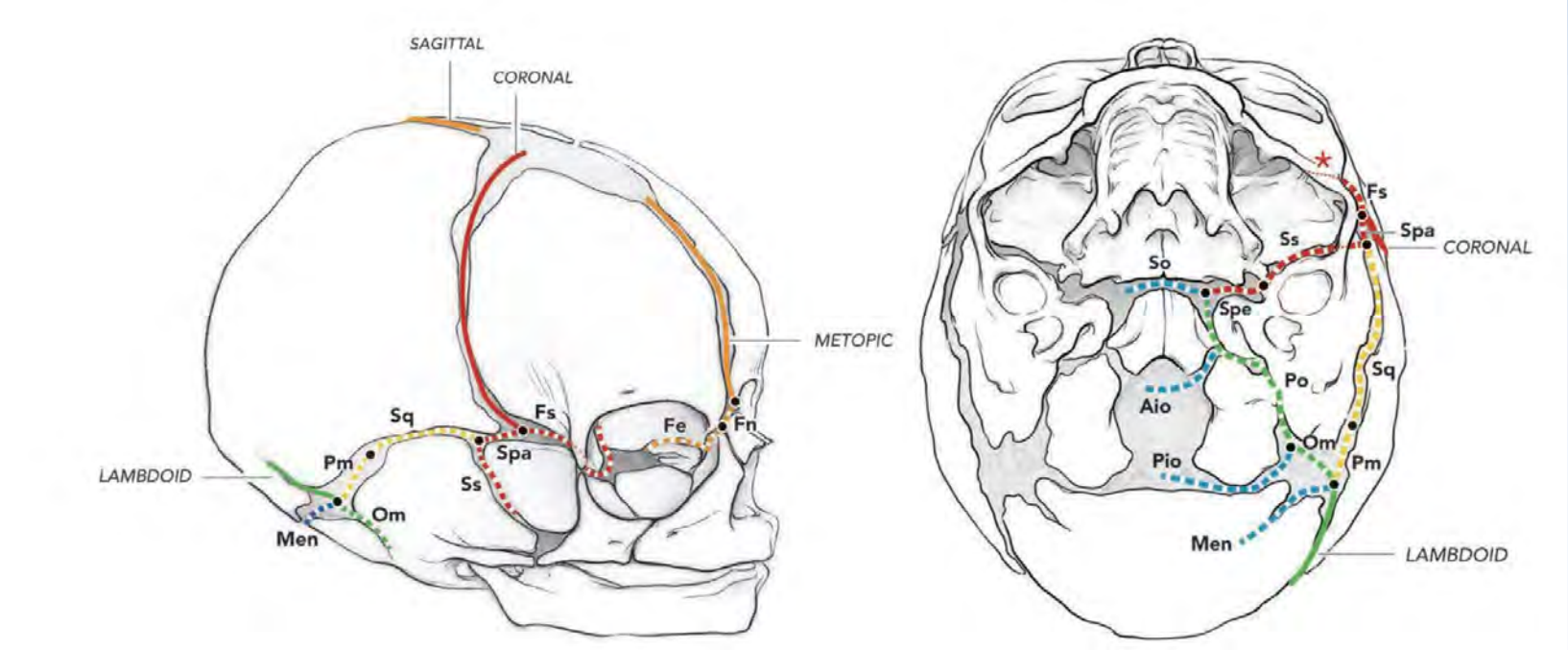


ANATOMY: MINOR SUTURES



SAGITTAL ARCH: ORANGE – SAGITTAL, METOPIC, FRONTONASAL (FN), FRONTOETHMOIDAL (FE)
CORONAL ARCH: RED – CORONAL, FRONTOSPHENOIDAL (FS), SPHENOPARIETAL (SPA), SPHENOSQUAMOUS (SS), SPHENOPETROSAL (SPE)
SQUAMOSAL ARCH: YELLOW – SQUAMOSAL, PARIETOMASTOID (PS)
LAMBDOID ARCH: GREEN – LAMBDOID, OCCIPITOMASTOID (OM), PETRO-OCCIPITAL (PO)

ANATOMY: SKULL BASE SUTURES = SYNCHONDROSES

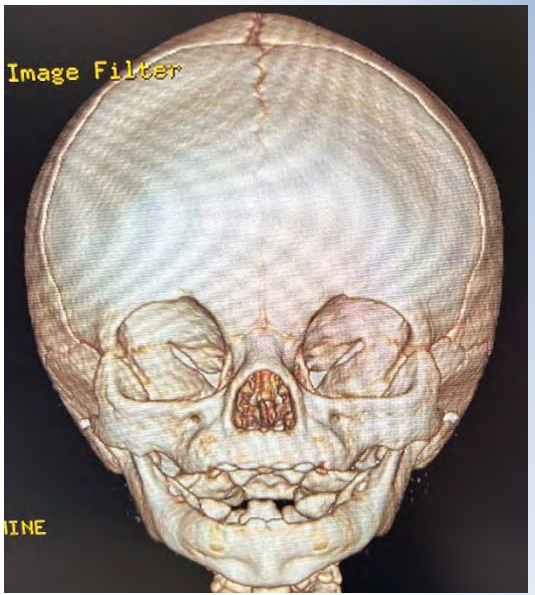


SPHENO-OCCIPITAL (S0)
POSTERIOR INTRAOCCIPITAL (PIO)
ANTERIOR INTRA-OCCIPITAL (AIO)
MENDOSAL (MEN)

ANATOMY: SUTURE/SYNCHONDROSIS CLOSURE

Normal Timing of fusion of calvarial sutures/synchondroses

Suture/Synchondrosis	Fusion Begins	Fusion Complete
Coronal	1–25 yr	43 yr
Ethmoidofrontal	Unknown	Unknown
Ethmoidosphenoidal	2 yr	15 yr
Frontosphenoidal	5 yr	15 yr
Lambdoid	6–18 yr	43 yr
Metopic	3 mo	12 mo
Occipitomastoid	5 yr	12–18 yr
Occipital synchondroses		
Anterior intraoccipital	1–2 yr	7–10 yr
Mendosal	1 mo	4–6 yr
Posterior intraoccipital	1-2 yr	4-7 yr
Parietomastoid	Unknown	Unknown
Parietosquamosal	Unknown	Adulthood
Petro-occipital	1 yr	17 yr
Sagittal	8-25 yr	41 yr
Squamosal	Unknown	Adulthood
Spheno-occipital	8 yr	18 yr
Sphenoparietal	Unknown	Unknown
Sphenopetrosal	4 yr	7-12 yr
Sphenosquamosal	4 yr	7-12 yr



Craniosynostosis: Classification

Anatomic

Single-suture: Major
- Sagittal
- Metopic
- Unicoronal
- Lambdoid

Single-suture: Minor
- Fronto-sphenoidal
- Squamosal

Multiple-suture
- Bicoronal
- Mercedes-Benz (Bilateral lambdoid and sagittal)
- Kleeblattschädel
- Metopic-sagittal
- Any combination possible

Craniosynostosis: Classification

Etiologic

- Primary
  - Non-syndromic – no genetic cause identified
    - Sagittal
    - Metopic
    - Unicoronal
    - Bicoronal
    - Lambdoid
  - Syndromic (>180 identifiable syndromes)
    - Muenke
    - Saethre-Chotzen
    - Apert
    - Crouzon
    - Pfeiffer
    - Craniofrontonasal dysplasia
  - Non-syndromic – genetic cause identified
    - TCF12-related craniosynostosis (craniosynostosis 3)
    - ERF-related craniosynostosis (craniosynostosis 4)
    - SMAD6 mutations (craniosynostosis 7)

Craniosynostosis: Classification

Etiologic

SECONDARY
- BIOMECHANICAL
- BONE METABOLIC DISORDERS
- MUCOPOLYSACCHARIDOSES
- NUTRITIONAL
- ENVIRONMENTAL

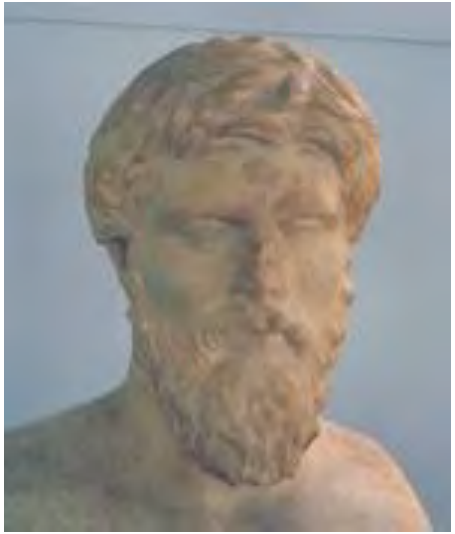
Nomenclature: What's in a name?

Nomenclature of Craniosynostosis

Name	Shape	Etiologic Factors	Incidence
Scaphocephaly Dolicocephaly	Keel or boat-shaped Long and narrow	Sagittal synostosis	1 in 2,500
Trigonocephaly	Triangular	Metopic synostosis	1 in 10,000
Plagiocephaly Anterior Posterior	Asymmetrical	Unicoronal synostosis Lambdoid synostosis Deformational	1 in 15,000 1 in 30,000 to 1 in 100,000 1 in 3 to 1 in 7
Turricephaly Brachycephaly Turribrachycephaly	Tower or tall Short, broad Tall and short/broad	Bilateral coronal	1 in 10,000 to 1 in 100,000
Oxycephaly	Conical	Bilateral coronal synostosis (Delayed onset)	
Kleeblattschädel	Clover-Leaf	Bicoronal and bilateral lambdoid	Rare

Craniosynostosis: Historical

Pericles (495 - 429 BC):
Athenian statesman, general, orator



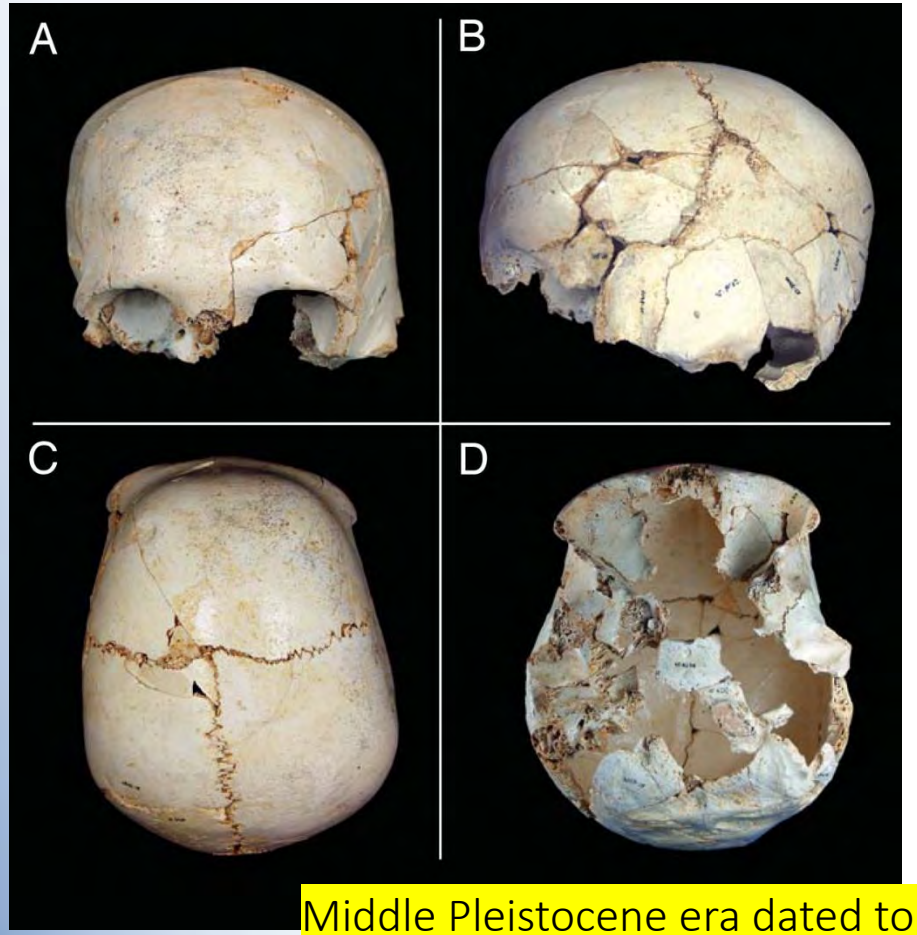
Mestrius Plutarchus (Plutarch)
(46-120 AD): Greek Historian

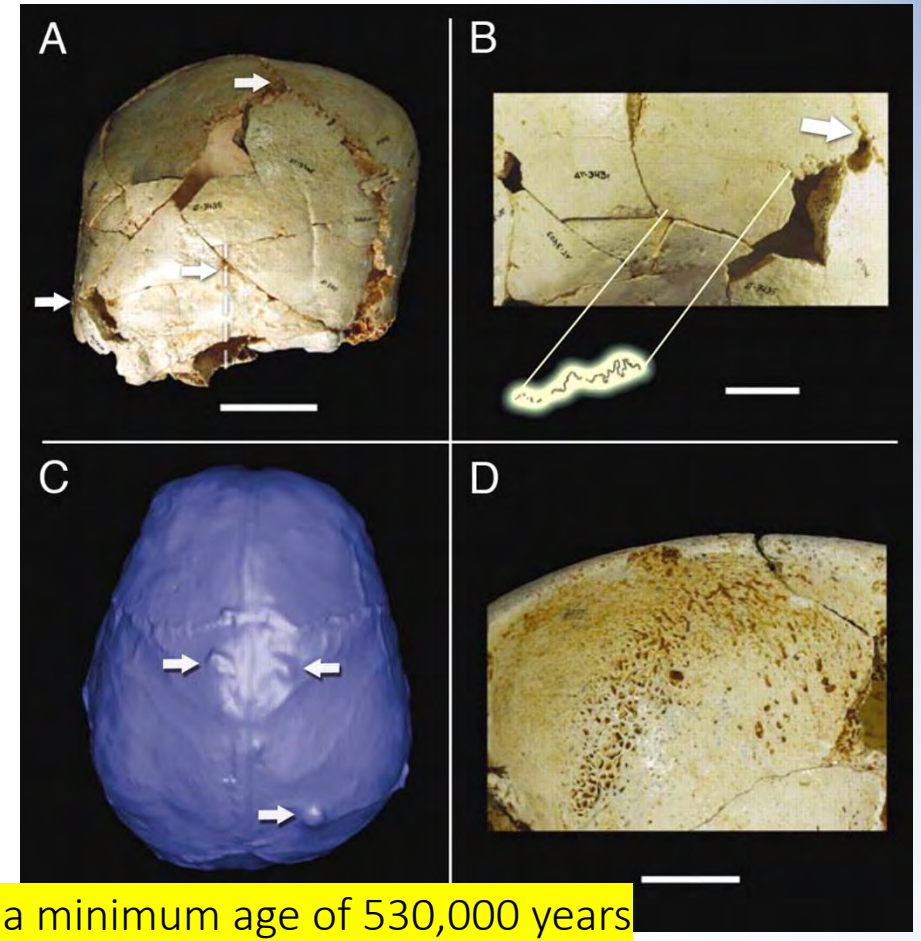
“Squill-headed”

Craniosynostosis: Historical

•Andreas Vesalius -
•De Humani Corporis Fabrica, 1543

Craniosynostosis: Historical





Middle Pleistocene era dated to a minimum age of 530,000 years

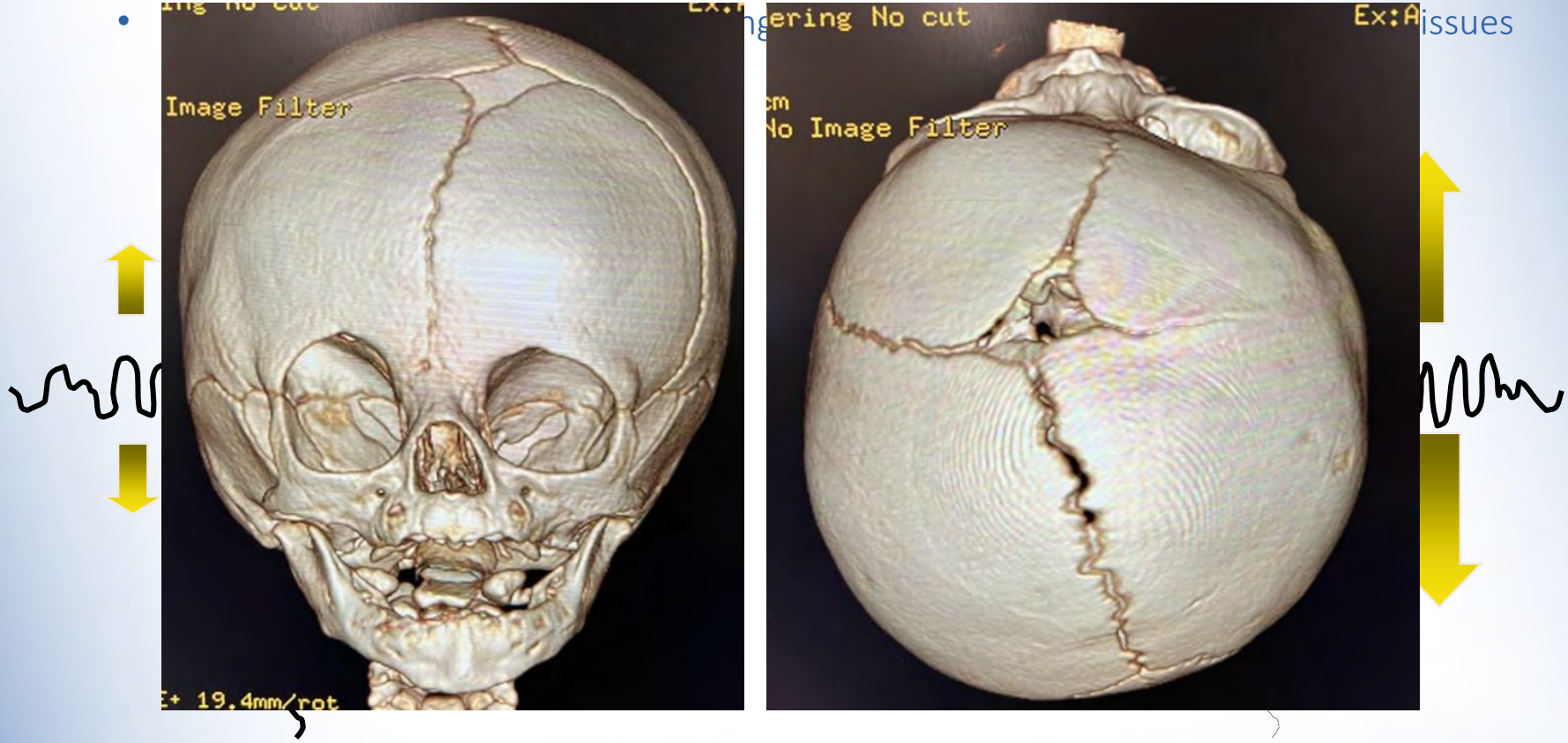
Rudolf Ludwig Karl Virchow (1821-1902)

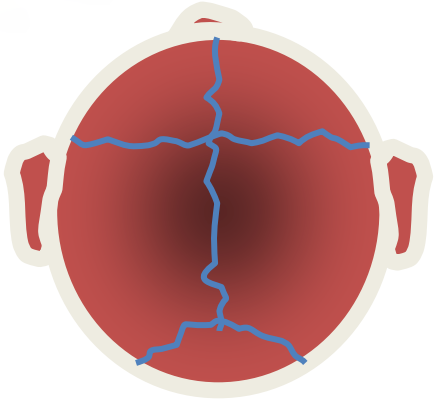
- Prototype “German anatomist at the turn of the century”
- Pathologist, politician and statesman
- Modern concept of pathological process - diseases arising from cells
- Discovered leukemia
- Elucidated mechanism of pulmonary embolism
- Virchow's law - *Omnis cellula e cellula* - every cell from a cell.

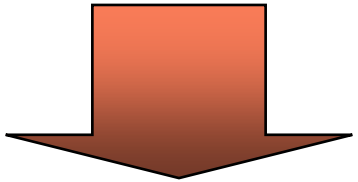
- Virchow's node - Enlargement of one of the supraclavicular lymph nodes.

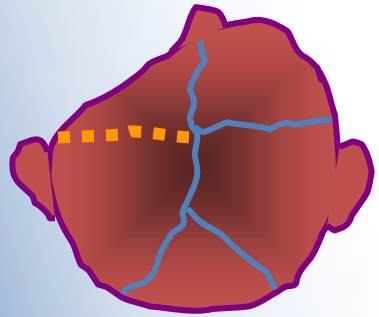
- Virchow's triad - The functional triad concerned in the pathogenesis of thrombosis.

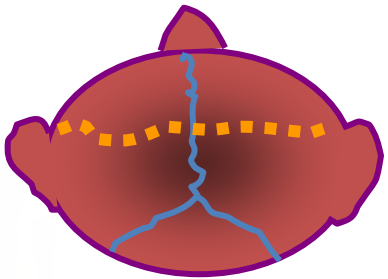
- Virchow's law: Growth of cranial sutures:
  - Bone growth at right angles to line of sutures
  - issues

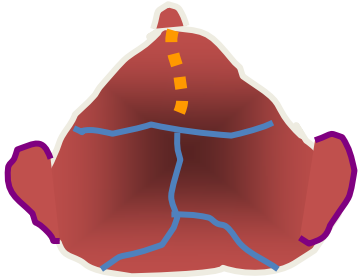


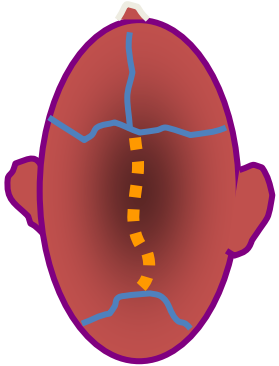


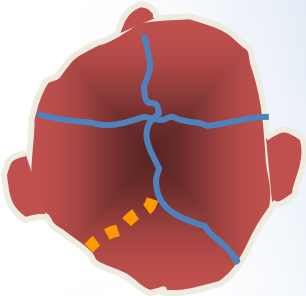










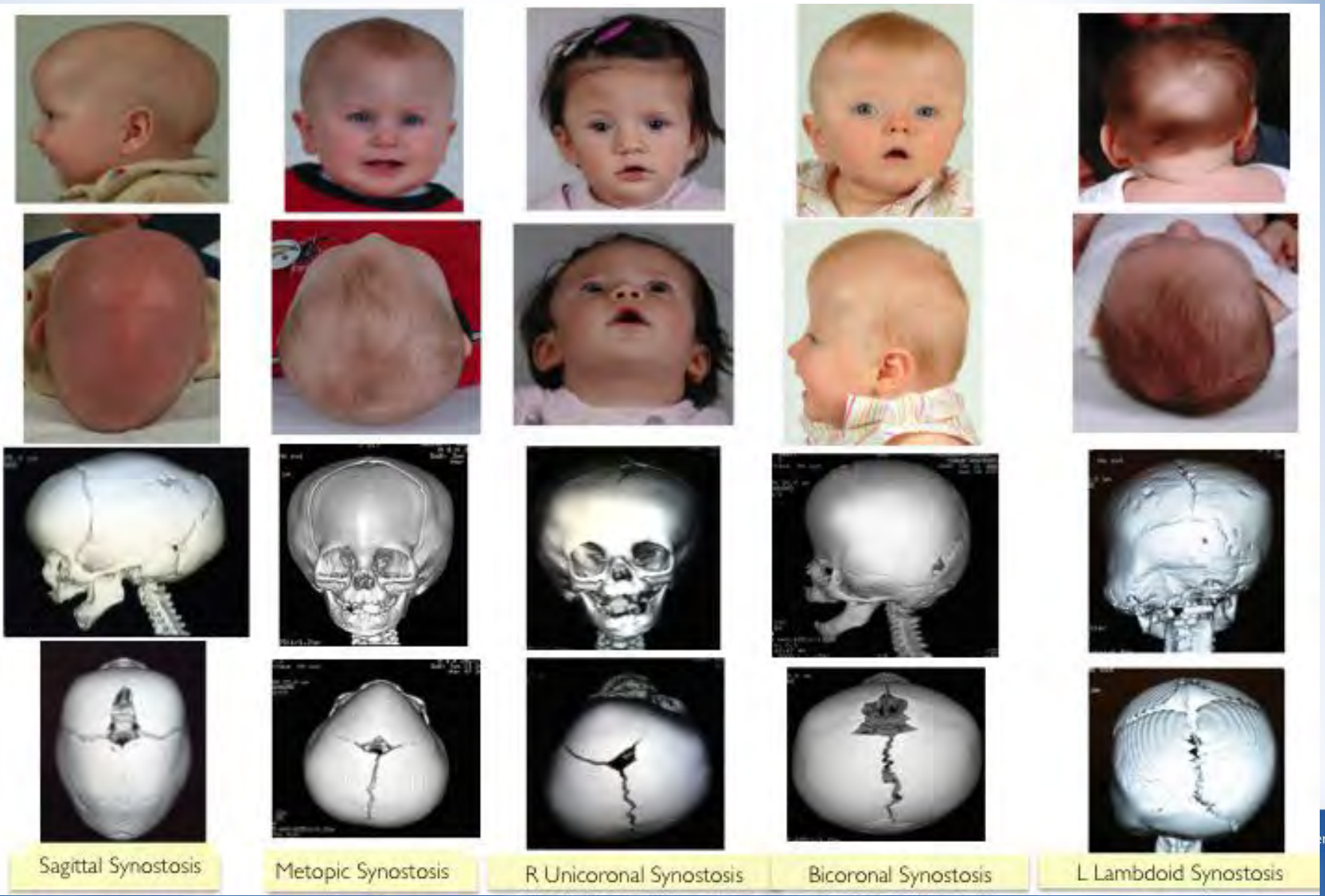


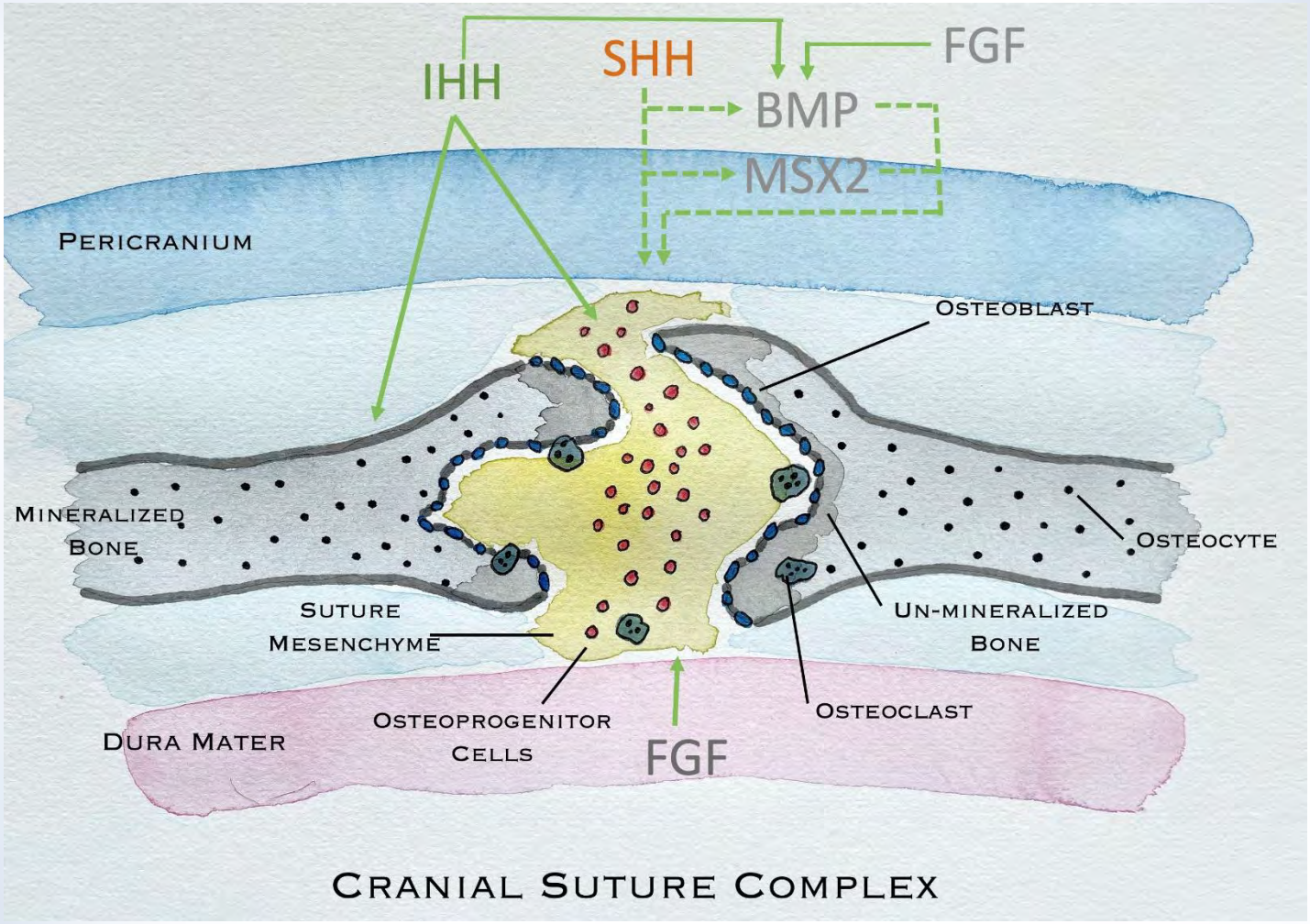
Uni-coronal
Lambdoid

Bicoronal

Metopic

Sagittal


Sagittal Synostosis
Metopic Synostosis
R Unicoronal Synostosis
Bicoronal Synostosis
L Lambdoid Synostosis



CRANIAL SUTURE COMPLEX

Etiology of Craniosynostosis

No simple answer

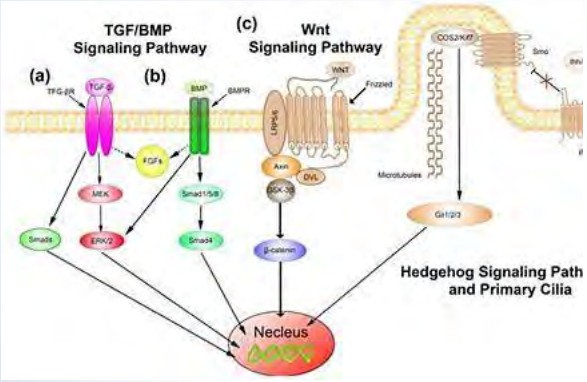
Multifactorial and heterogeneous: monogenic and polygenic, chromosomal, environmental/teratogenic and epigenetic factors

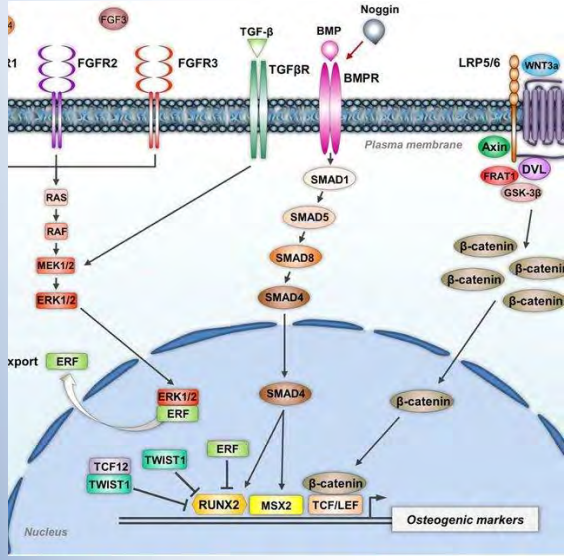
Complex interactions between genetics, epigenetics and the molecular signalling pathways important in embryogenesis and bone formation

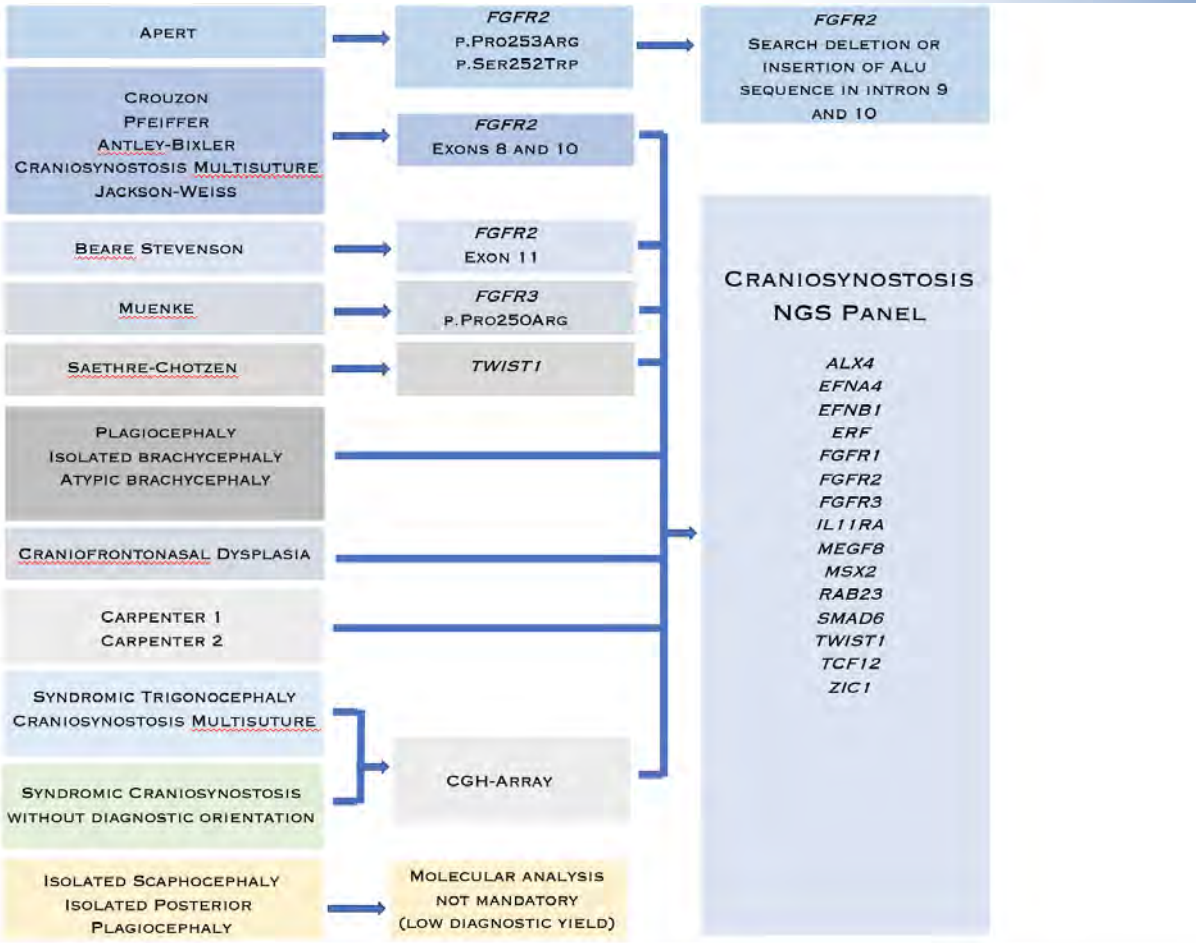
Balance between pro-osteogenic and anti-osteogenic forces at play during suture embryogenesis

Environmental factors

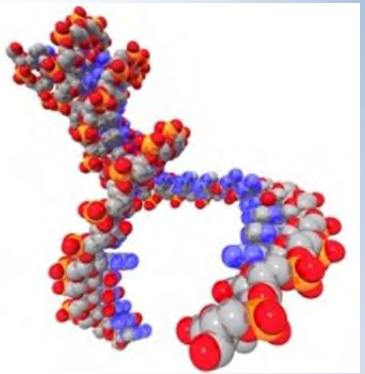
- advanced maternal and paternal age (greater than 35 years),
- maternal smoking
- prenatal exposure to nitrosable drugs, valproic acid and phenytoin, hyperthyroidism
- multiple births
- birth weight > 4 kg
- paternal occupation (more common in agriculture, forestry, repairmen and mechanics)


TGF/BMP
Signaling Pathway
(c)
Wnt
Signaling Pathway
(a)
(b)
Hedgehog Signaling Path
and Primary Cilia
Necleus


FGF3
Noggin
TGF-β
BMP
FGFR2
FGFR3
TGFβR
BMPR
LRP5/6
WNT3a
Plasma membrane
Axin
FRAT1
DVL
GSK-3β
RAS
RAF
MEK1/2
ERK1/2
SMAD1
SMAD5
SMAD8
SMAD4
β-catenin
ERF
TCF12
TWIST1
RUNX2
MSX2
TCF/LEF
Osteogenic markers
Nucleus


APERT
FGFR2
P.PRO253ARG
P.SER252TRP
CROUZON
PFEIFFER
ANTLEY-BIXLER
CRANIOSYNOSTOSIS MULTISUTURE
JACKSON-WEISS
FGFR2
EXONS 8 AND 10
BEARE STEVENSON
FGFR2
EXON 11
MUENKE
FGFR3
P.PRO250ARG
SAETHRE-CHOTZEN
TWIST1
PLAGIOCEPHALY
ISOLATED BRACHYCEPHALY
ATYPIC BRACHYCEPHALY
CRANIOFRONTONASAL DYSPLASIA
CARPENTER 1
CARPENTER 2
SYNDROMIC TRIGONOCEPHALY
CRANIOSYNOSTOSIS MULTISUTURE
SYNDROMIC CRANIOSYNOSTOSIS
WITHOUT DIAGNOSTIC ORIENTATION
CGH-ARRAY
ISOLATED SCAPHOCEPHALY
ISOLATED POSTERIOR
PLAGIOCEPHALY
MOLECULAR ANALYSIS
NOT MANDATORY
(LOW DIAGNOSTIC YIELD)
FGFR2
SEARCH DELETION OR
INSERTION OF ALU
SEQUENCE IN INTRON 9
AND 10
CRANIOSYNOSTOSIS
NGS PANEL
ALX4
EFNA4
EFNB1
ERF
FGFR1
FGFR2
FGFR3
IL11RA
MEGF8
MSX2
RAB23
SMAD6
TWIST1
TCF12
ZIC1

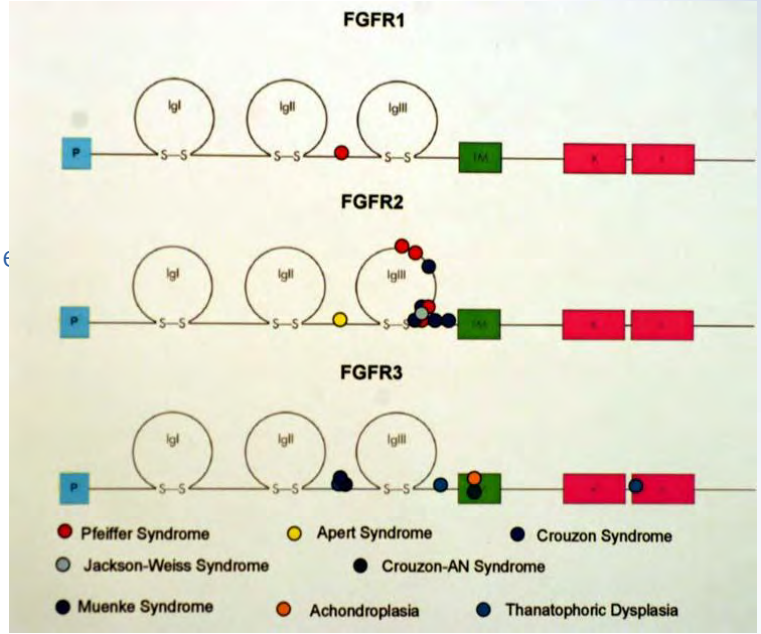
Molecular genetics of craniosynostosis



- >60 different genes felt to be causative
- Majority de novo mutations
- 14+ syndromes
  - 3 categories
    - Transcription factors
      - MSX1 gain-of-function mutation in muscle segment homeobox 2 (MSX2) gene
      - TWIST – basic helix-loop-helix transcription factor
    - FGFR - gain of function
      - FGF family of 22 highly conserved proteins
      - regulate cellular proliferation, differentiation and migration
      - important in wound healing, angiogenesis, limb development, mesoderm induction, malignant
    - Structural protein - EFBN1
      - Craniofrontonasal dysplasia may be caused by mutations or defects in the ephrin-B1 gene (EFNB1)
      - ephrin-B1 protein - important for normal development of the frontonasal neural crest

Molecular genetics of craniosynostosis

- MSX1 – Boston-type craniosynostosis
- FGFR-mediated craniosynostoses: 8
  - Apert syndrome (FGFR2)
  - Crouzon syndrome (FGFR2)
  - Jackson-Weiss syndrome (FGFR2)
  - Muenke syndrome (FGFR3)
  - Crouzon syndrome with acanthosis nigricans (Crouzonode syndrome) (FGFR3)
  - Pfeiffer syndrome (FGFR1 and FGFR2)
  - Beare-Stevenson syndrome (FGFR2)
  - FGFR2-related isolated coronal synostosis.
- TWIST – Saethre-Chotzen
- EFNB1 – Craniofrontonasal dysplasia



Molecular Testing of recurrent mutations (Adapted with permission from [5]).

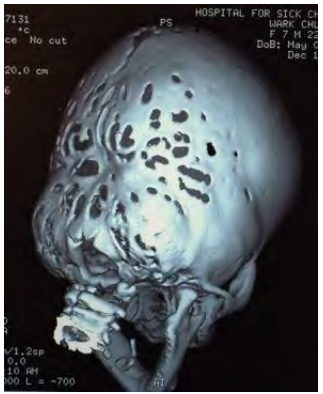
Disorder	Gene (% responsible)	Gene (% responsible) Mutations	Mutation Detection Rate
Pfeiffer	FGFR2 (>95%) FGFR1 (<5%)	Several	67%
Apert	FGFR2 (100%)	Ser252Trp, Pro253Arg	>98%
Crouzon	FGFR2 (100%)	Several	>50%
Crouzon with acanthosis	FGFR3 (100%)	Ala391Glu	100%
Muenke	FGFR3 (100%)	Pro250Arg	100%
Saerthre Chotzen	TWIST1	Several mutations & deletions	46% to 80%

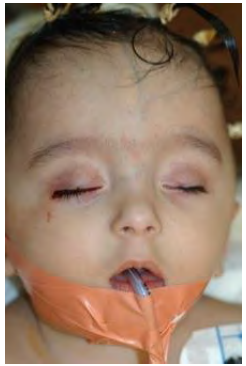
Craniosynostosis:

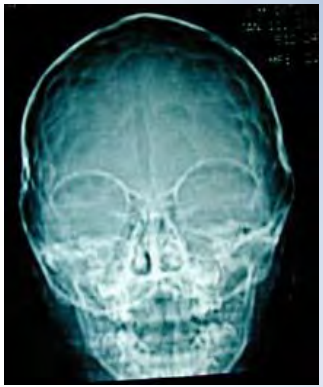
- Functional concerns
  - 1. Increased ICP
  - 2. Ocular
  - 3. Airway
  - 4. Neurodevelopmental
  - 5. Psychosocial

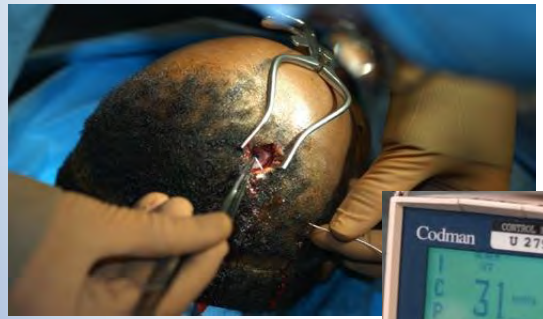
Craniosynostosis:

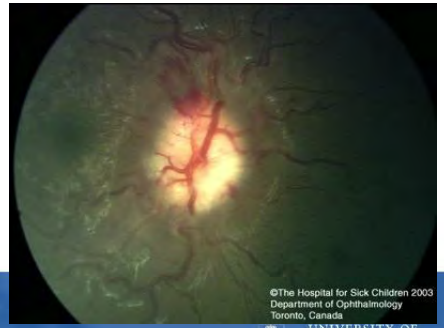
- Functional concerns
  - ICP
    - Incidence:*
      - NS: 14 - 17%
      - S: 40 – 51%
    - Diagnostic dilemmas
    - Significance

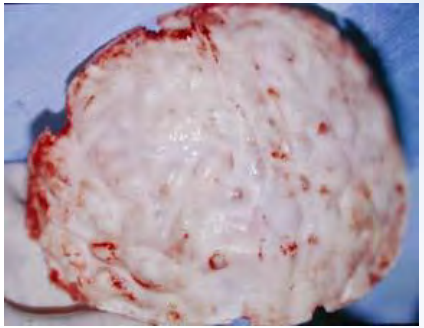








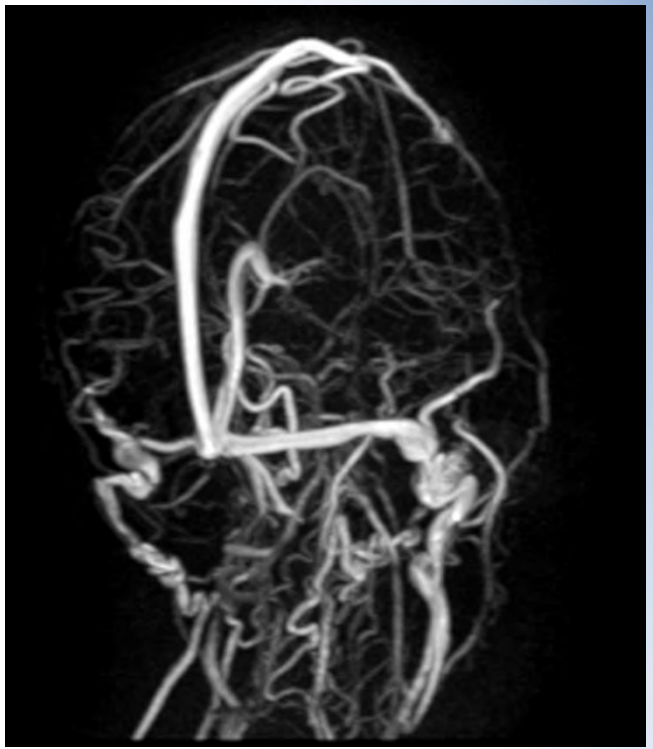


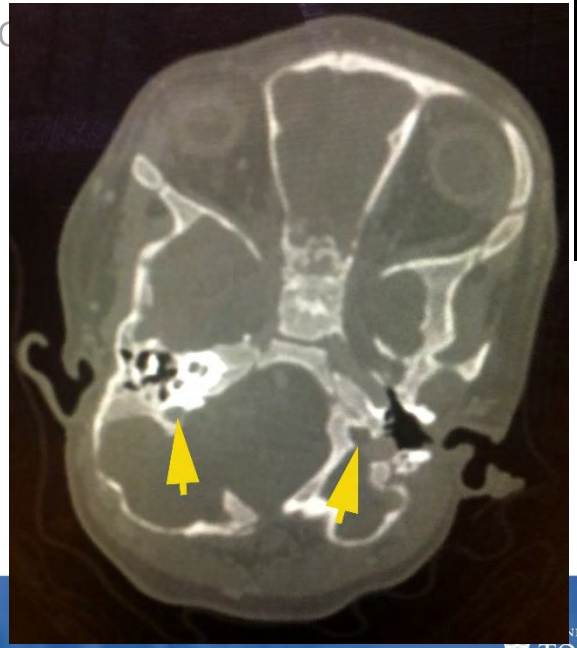


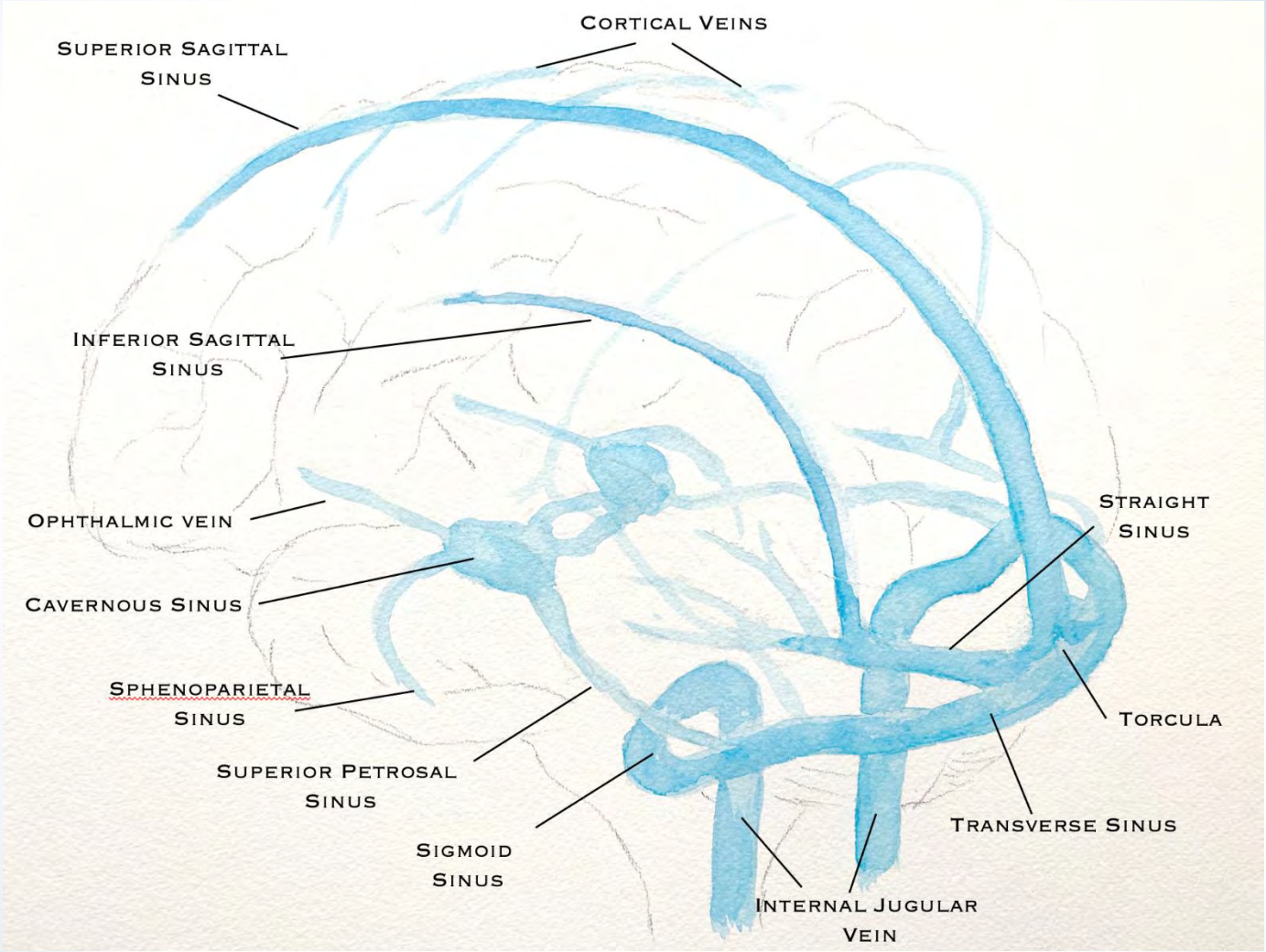
•Marchac and Renier, 1985
•Thomson et al. 1995
•Bannink et al. 2008

Craniosynostosis:

- ICP elevation: why?
  - Venous hypertension
  - Hydrocephalus
  - Cranial-cerebral disproportion
  - Airway obstructi

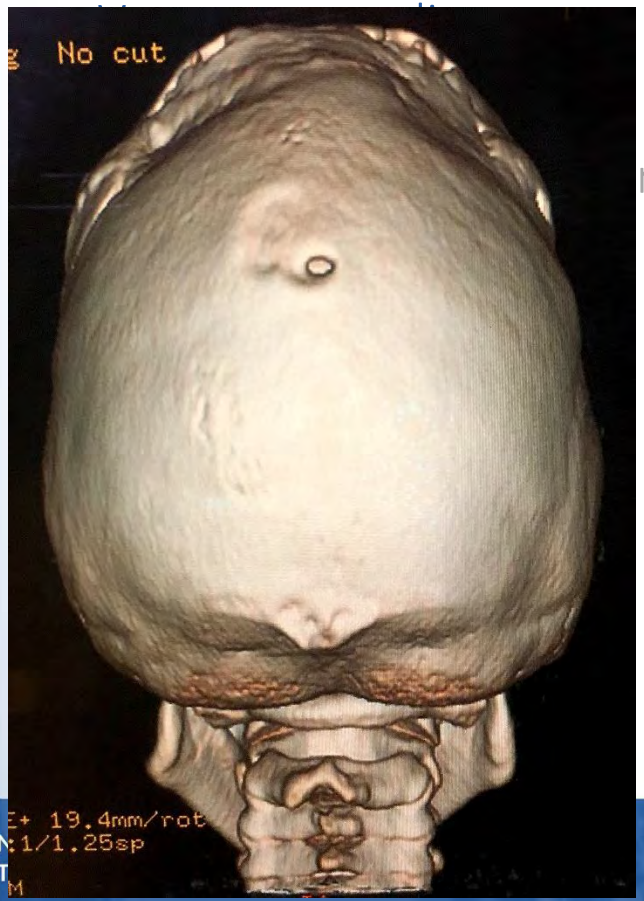


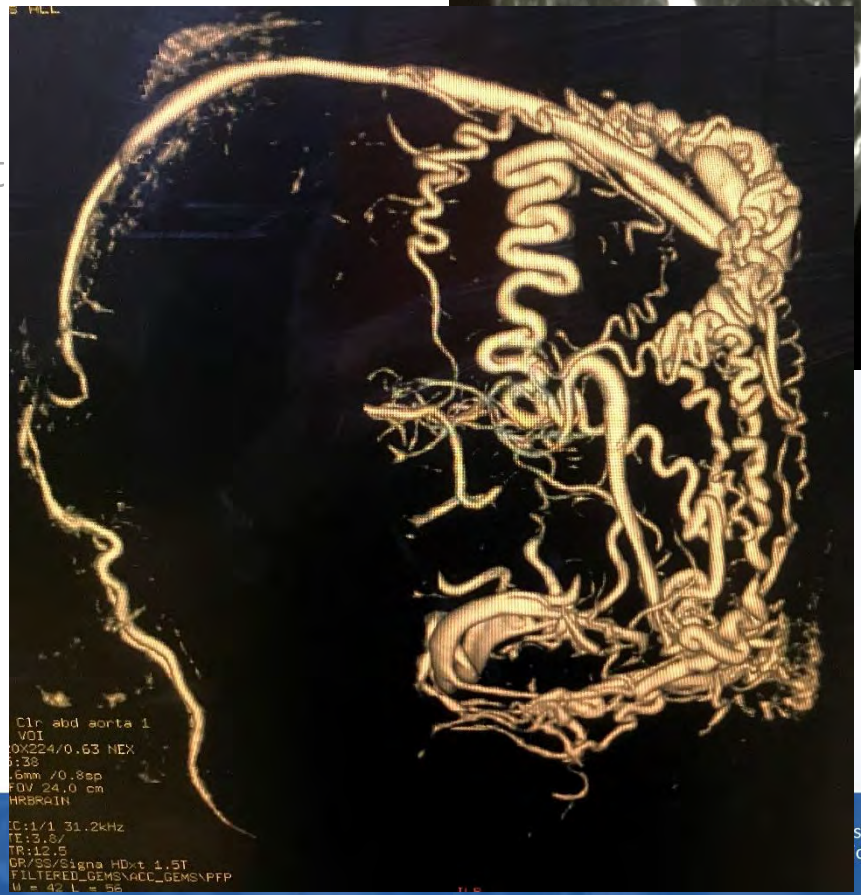



Cortical Veins
Superior Sagittal Sinus
Inferior Sagittal Sinus
Ophthalmic vein
Straight Sinus
Cavernous Sinus
Sphenoparietal Sinus
Torcula
Superior Petrosal Sinus
Transverse Sinus
Sigmoid Sinus
Internal Jugular Vein

Craniosynostosis:

- ICP elevation: why?





OPEN

PRS GLOBAL OPEN ORIGINAL ARTICLE

Pediatric/Craniofacial

Clinical Significance of Venous Anomalies in Syndromic Craniosynostosis

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Caitlin E. Hoffman, MD†
Vassilios Tsitouras, MD†
Dhruve S. Jeevan, MD†
Emily S. Ho, BSc, OT, MEd*
James M. Drake, MD, MB, BCh, MSc, FRCSC†
Christopher R. Forrest, MD, MSc, FRCSC, FACS*

Background: The pattern of cranial venous drainage in syndromic craniosynostosis is unpredictable and not adequately understood. Collateral channels substitute for stenotic venous sinuses and pose potential risk for surgical intervention. The purpose of this study was to analyze the patterns of venous drainage in patients with syndromic craniosynostosis and their influence on operative planning and morbidity.

Methods: A retrospective study of patients with syndromic craniosynostosis from 2000 to 2013 was performed. Demographic data were collected including phenotype and associated pathologies. Pre- and/or postoperative venous imaging was reviewed for venous sinus stenosis, collateral emissaries, and persistent fetal sinuses. Categorization of anomalous venous drainage was performed, and the relationship with surgical morbidity was assessed.

Results: Forty-one patients were identified. Anomalies were present in 31 patients (76%) consisting of dural sinus stenosis in 28 (68%), dilated emissaries in 26 (63%), and fetal sinuses in 7 (17%). Pfeiffer syndrome was most commonly associated with anomalous drainage (100%). Venous anomalies were associated with elevated ICP, shunted hydrocephalus, Chiari malformations, and sleep apnea. In 5 cases, the surgical plan was adjusted based on anomalous anatomy. No mortalities occurred. Intraoperative complication rate was 7.3%, all with anomalous drainage. Median estimated blood loss was 1,100 cc for patients with anomalies versus 400 cc without anomalies ($P = 0.181$).

Conclusion: Cranial venous anomalies are commonly detected in patients with syndromic craniosynostosis and may affect surgical morbidity and outcome with a higher estimated blood loss, alteration of procedure, and postoperative morbidity. Detailed preoperative imaging of the venous drainage is therefore recommended in cases of syndromic synostosis. *(Plast Reconstr Surg Glob Open XXX;6:e1613; doi: 10.1097/GOX.0000000000001613; Published online xxx xxx 2017.)*

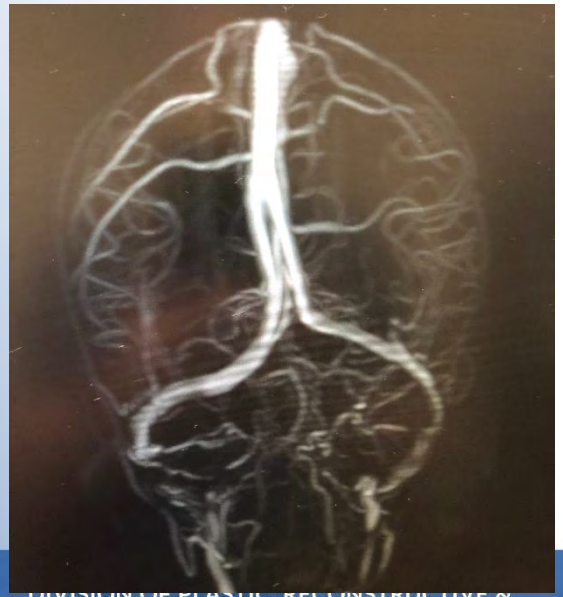
AQ5

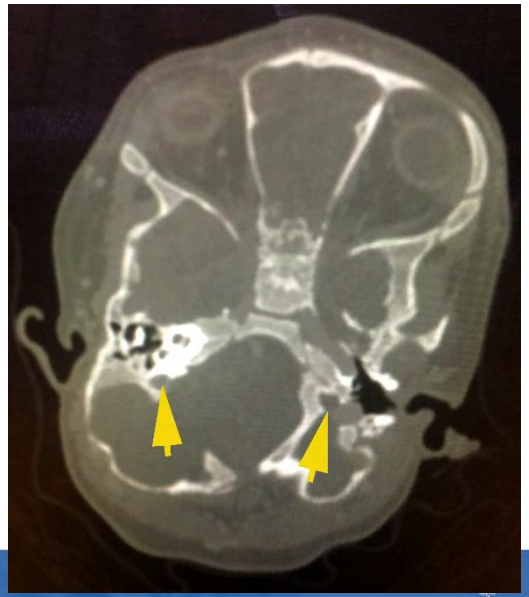
Table 2. Venous Anomaly by Syndrome

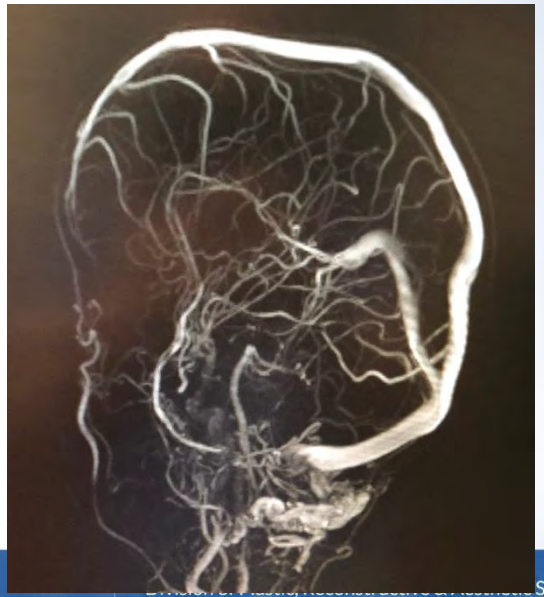
	Apert	Crouzon	Pfeiffer	SC	Muenke	Total
Total no. patients	11	19	7	3	1	41
Anomalous venous anatomy (%)	7 (64)	16 (84)	7 (100)	1(33)	0 (0)	31 (76)

Table 5. Frequency of Venous Anomalies by Syndrome

	Pfeiffer (n = 7)	Crouzon (n = 19)	Apert (n = 11)	SC (n = 3)	Muenke (n = 1)
Venous anomalies, n (%)	7 (100)	16 (84)	7 (64)	1 (33)	1 (100)
DVS stenosis, n (%)	7 (100)	14 (74)	6 (55)	1 (33)	0
JF stenosis	4 (57)	9 (47)	6 (55)	1 (33)	0
Emissary veins	7 (100)	14 (74)	4 (36)	1 (33)	0
Persistent fetal sinuses	3 (43)	5 (26)	0	0	0

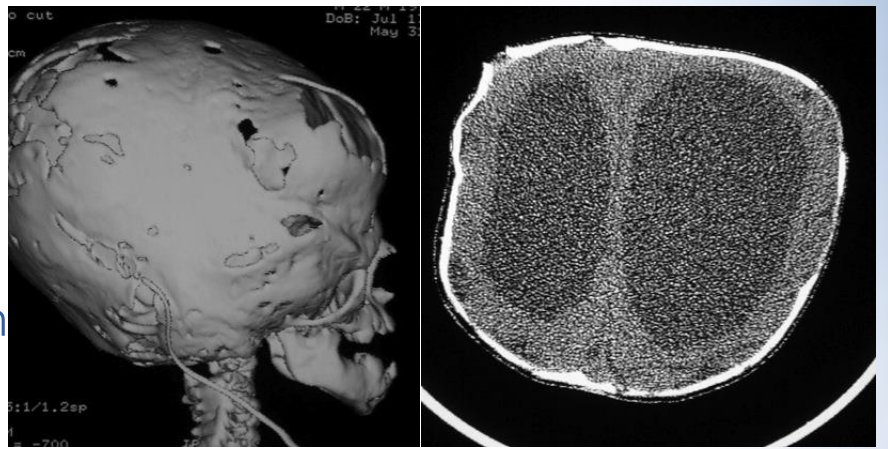


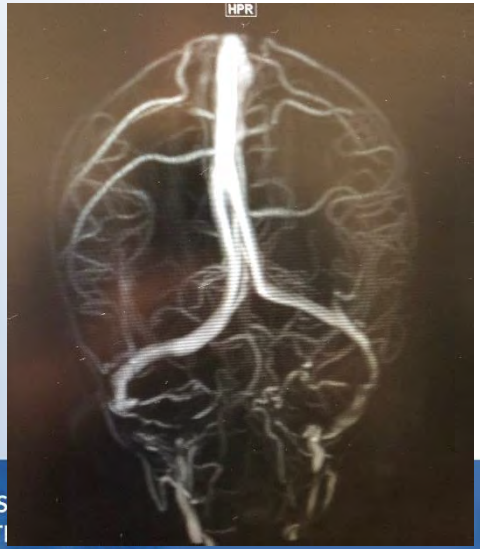


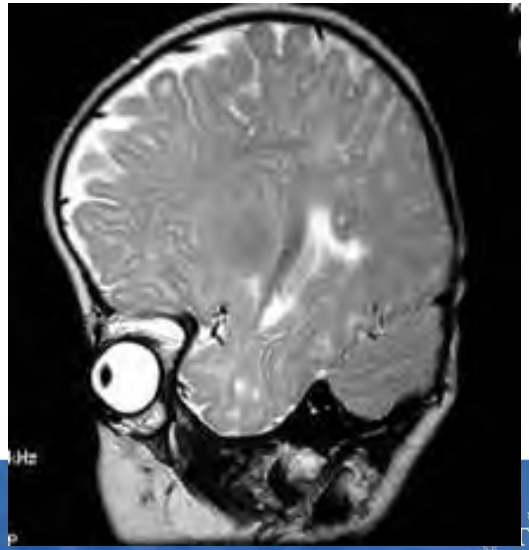


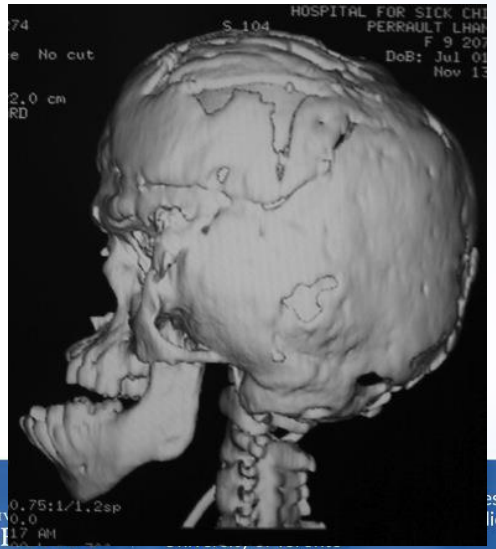
Craniosynostosis:

- ICP elevation: why?
  - Venous anomalies
  - Hydrocephalus
  - Cranial-cerebral disproportion
  - Airway obstruction-OSA









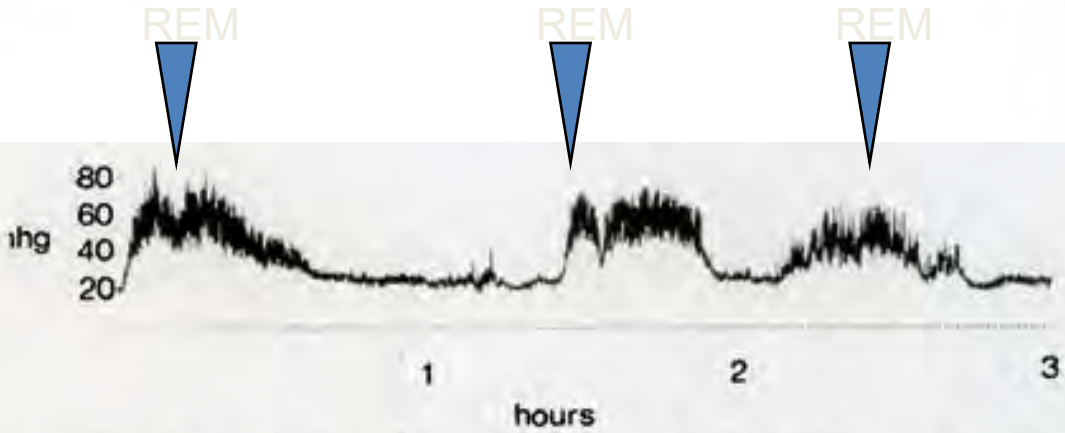


Fig. 3 Intracranial pressure monitoring trace (sleeping) of a child with Crouzon's syndrome and intermittent airway obstruction. Baseline pressures were around 20–25 mmHg, rising to plateaux (Lundberg A waves) of 45–60 mmHg that coincided with episodes of snoring (airway compromise)

- Hayward, R. Venous hypertension and craniosynostosis. Childs Nerv Syst (2005) 21:800-6

Sleep Medicine

J Craniomaxillofac Surg. 2011 Apr;39(3):153-7. doi: 10.1016/j.jcms.2010.04.011. Epub 2010 Jun 2.

Sleep-related disordered breathing in children with syndromic craniosynostosis.

Al-Saleh S[1], Riekstins A, Forrest CR, Philips JH, Gibbons J, Narang I.

Author information

Abstract

BACKGROUND: Syndromic craniosynostosis patients are at risk for sleep-related disordered breathing (SRDB) but the role of polysomnography (PSG) in assessing these patients has not been fully explored. Our aim was to evaluate the prevalence or severity of SRDB in children with syndromic craniosynostosis or the impact of treatments on their SRDB.

METHODS: We conducted a retrospective review of all patients with syndromic craniosynostosis referred between 1996 or 2008 for an initial PSG to rule out SRDB. For those with SRDB, we reviewed the interventions post PSG.

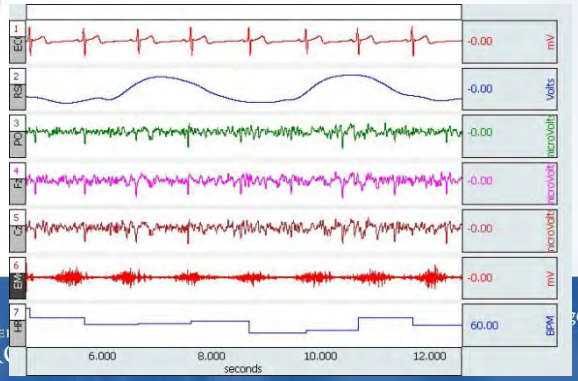
RESULTS: 35 patients (18 females) were included. Specific diagnoses were Crouzon's (n=18), Apert's (n=14), Pfeiffer (n=2) or Saethre-Chotzen (n=1) syndromes. Their mean age was 4.5 years or their median body mass index (BMI) was 16.9 kg/m(2). Of these patients, 26/35 (74%) had evidence of SRDB. The median obstructive apnoea index was 6.6/h (range 0.5-36.4/h) or median central apnoea index was 1.0/h (range 0.0-66.4/h). A total of 16 children had interventions to treat SRDB, of which 14/16 had a follow up PSG or only 10/14 (x%) had a significant improvement of their SRDB.

CONCLUSION: This review confirms a high prevalence SRDB in this referred population. Despite various interventions, complete resolution of SRDB could not be achieved.

PMID: 20627744 DOI: 10.1016/j.jcms.2010.04.011

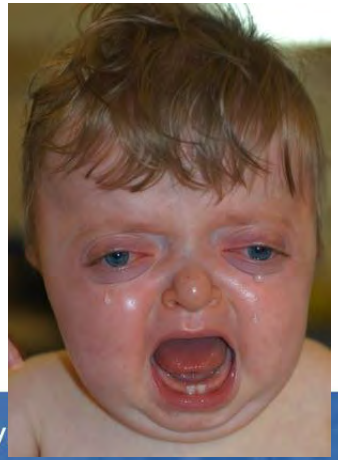
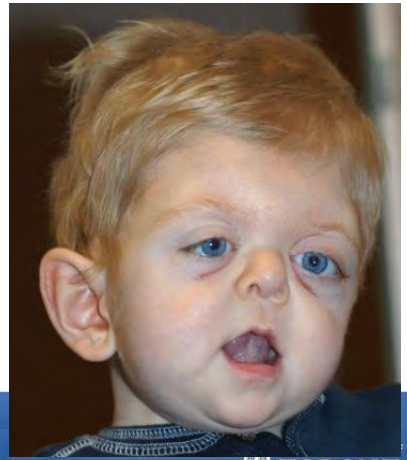
[Indexed for MEDLINE]

74% INCIDENCE OF SRDB



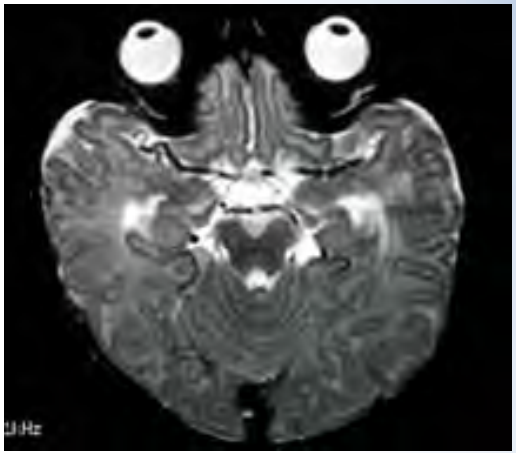
Pathophysiology of elevated ICP

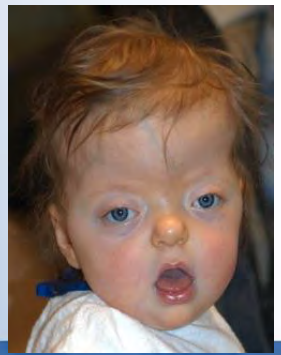
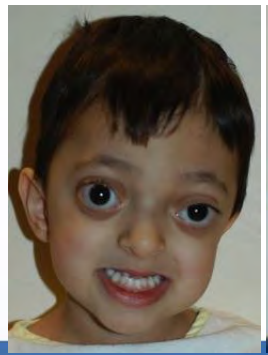
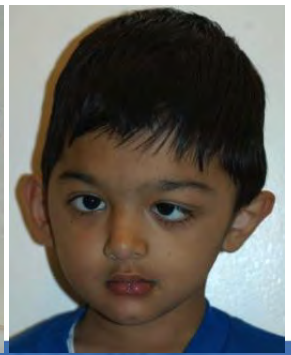
- ICP affects neurological function mainly through effect on cerebral blood flow
  - Brain dependent on flow for glucose, oxygen
  - Flow dependent on pressure gradient and inversely related to resistance
  - Elevation of ICP increases resistance to flow
  - Localized areas of hypoperfusion??

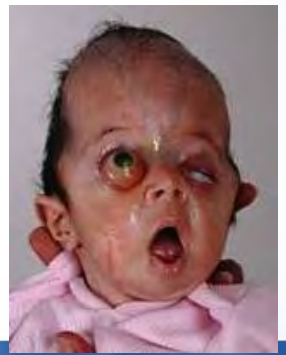





Craniosynostosis: Special Considerations

- Functional concerns
  - ICP
  - Visual - 30/58 patients syndromic craniosynostosis*
    - some degree of visual loss
      - Exorbitism
      - EOM anomalies
      - Optic nerve
      - Refractive errors/amblyopia >>> structural anomalies



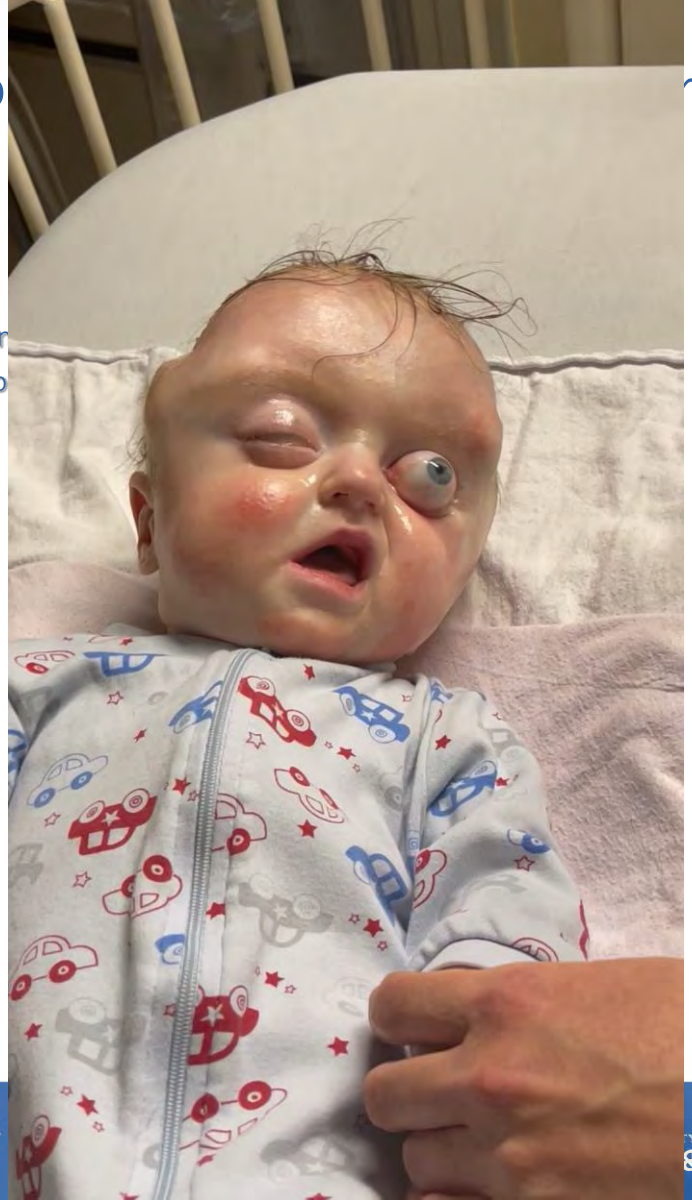






*Heckle et al., *J Ped Ophthal Strabis.* 28: 344, 199

Cranio rations

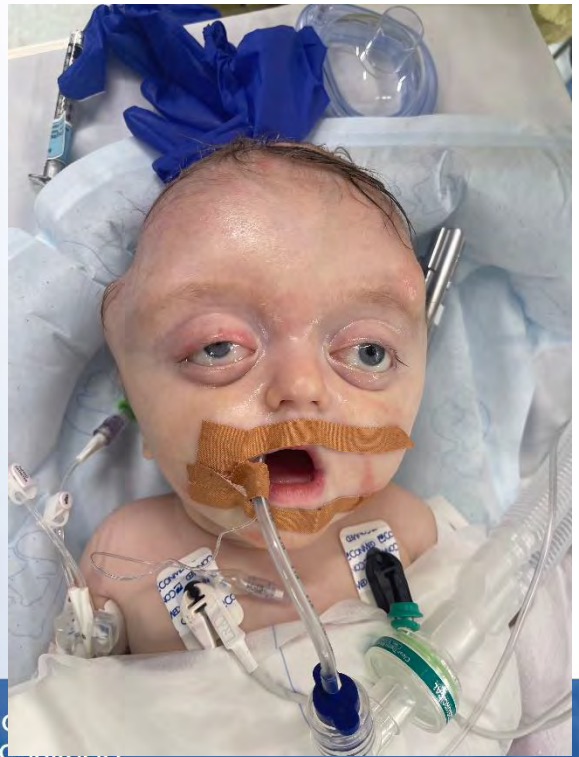
- Functional concerns
  - ICP
  - Visual - 30/58 patients syr
    - some degree o

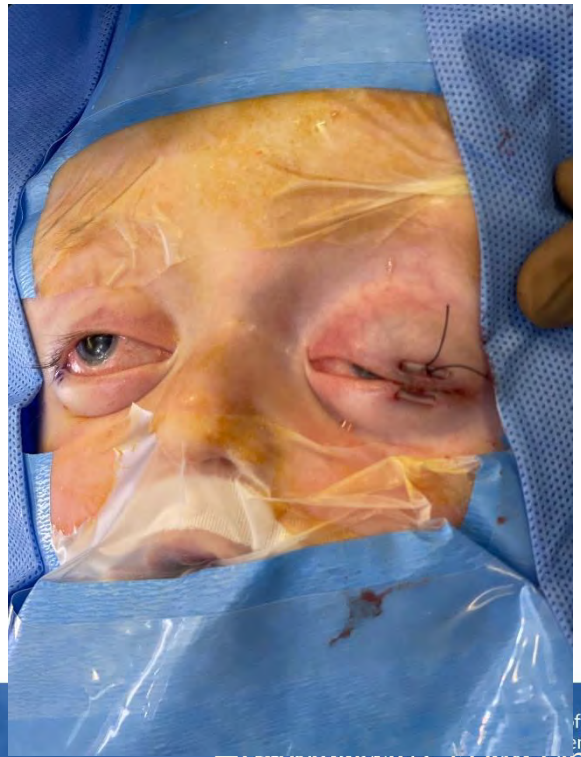


al., *J Ped Ophthal Strabis.* 28: 344, 199

Craniosynostosis: Special Considerations

- Functional concerns
  - ICP
  - Visual - 30/58 patients syndromic craniosynostosis*
    - some degree of visual loss

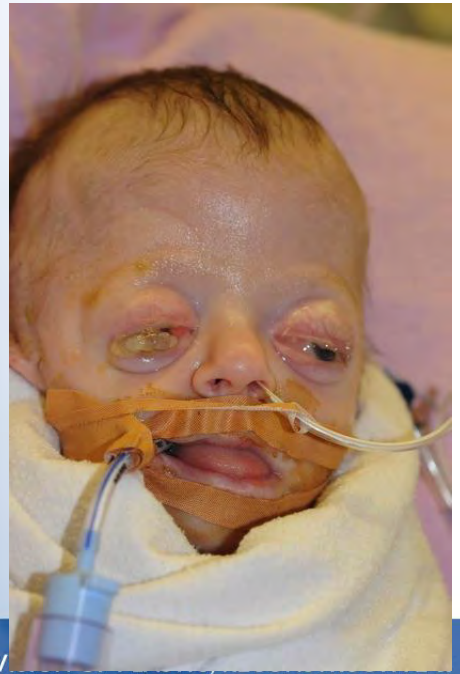


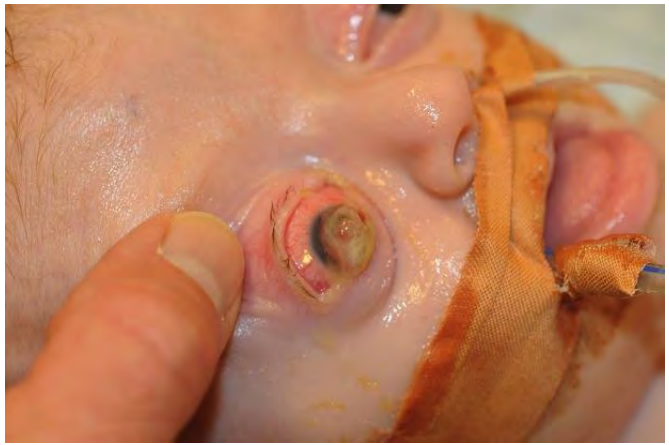


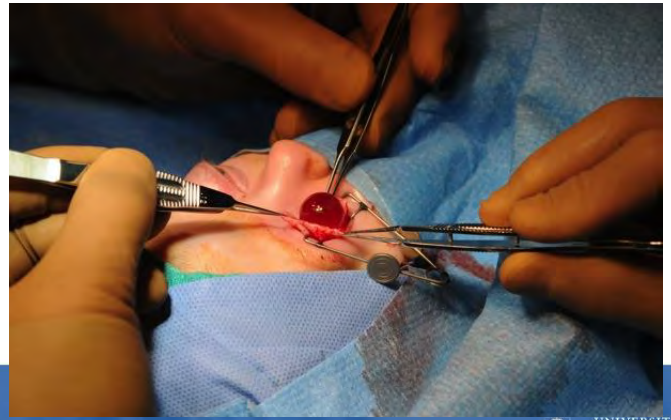
Heyne et al. J Pediatr Ophthal Strabis. 28: 344, 199

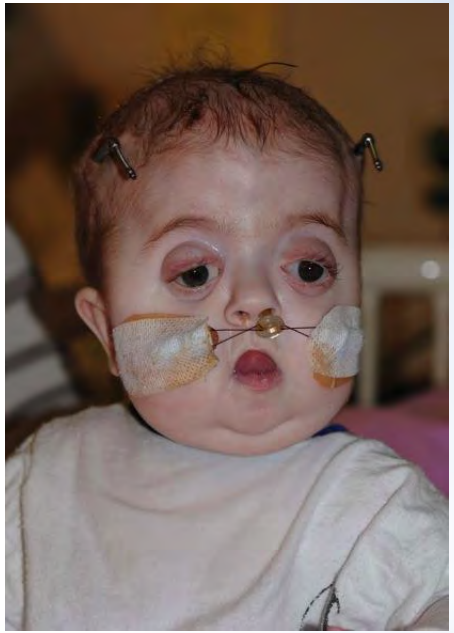
Craniosynostosis: Special Considerations

- Functional concerns
  - ICP
  - Visual
    - Exorbitism

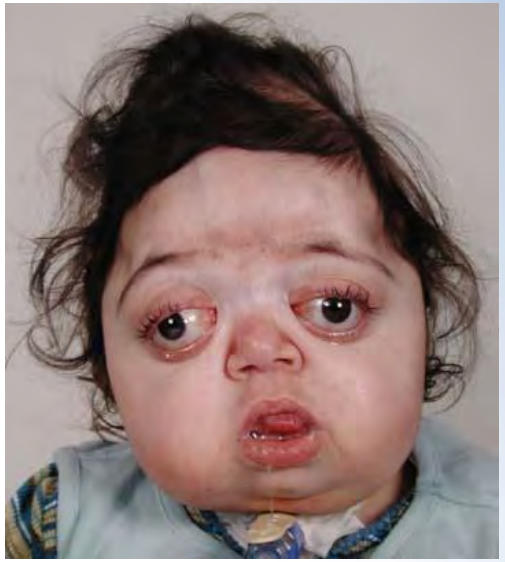


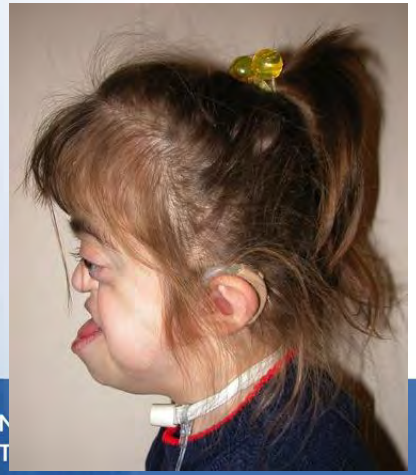


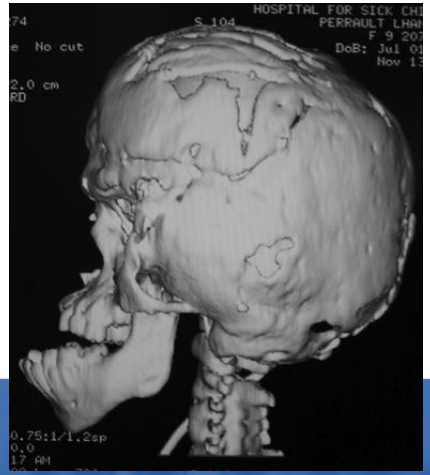


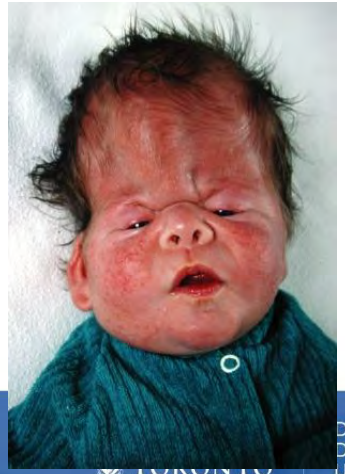


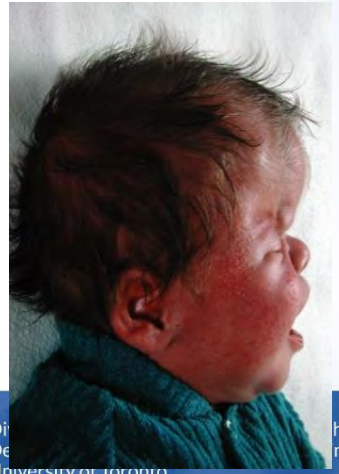
- Functional Concerns
  - ICP
  - Brain growth
  - Ocular
  - Airway
    - Midface retrusion
    - Choanal atresia
    - Tracheal abnormalities
    - GER





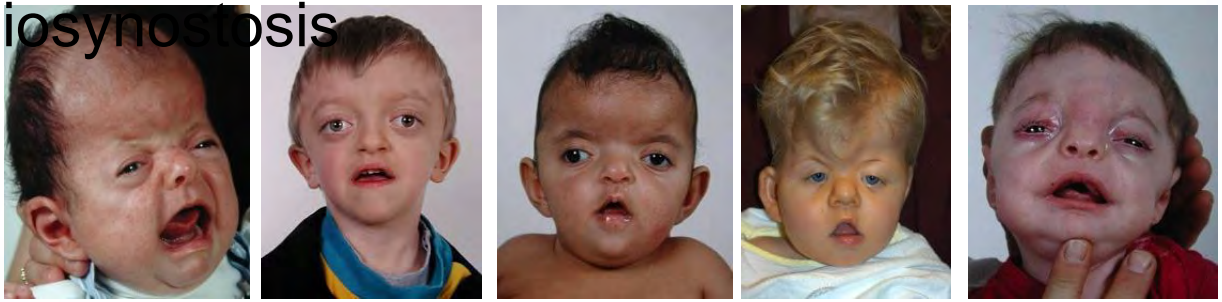




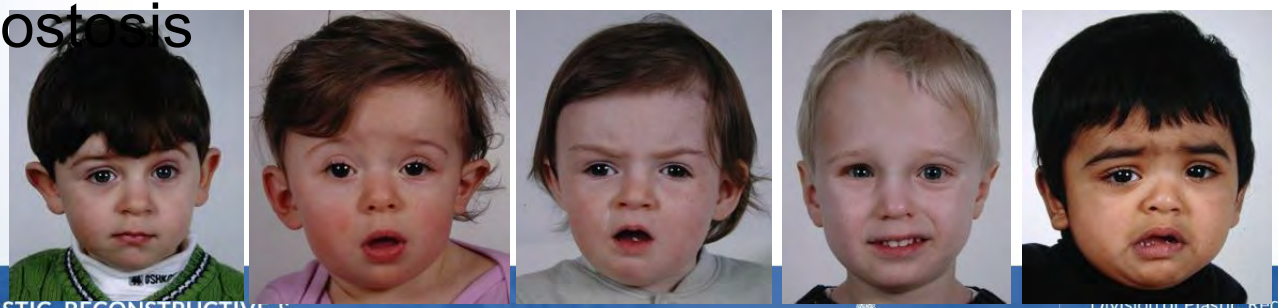


Neurobehavioural Issues

Syndromic/multiple suture craniosynostosis



Single suture synostosis



???

The Controversy

Synostosis --> brain deformation/ICP -->neuropsychological impairment

- NO! Children with non-syndromic synostosis do not differ from normalized sample in mental or physical development
  - Particularly within the first 2 years of life

Virtanen, R., et al. (1999). Neurocognitive sequelae of scaphocephaly. Pediatrics, 103(4), 791-5.

Kapp-Simon, K.A. (1998). Mental development and learning disorders in children with single suture craniosynostosis. *Cleft Palate-Craniofacial Journal, 35*(3), 197-203.

Speltz, M.L., et al. (1997). Presurgical and postsurgical mental and psychomotor development of infants with sagittal synostosis. *Cleft Palate-Craniofacial Journal, 34*(5), 374-379.

The Controversy

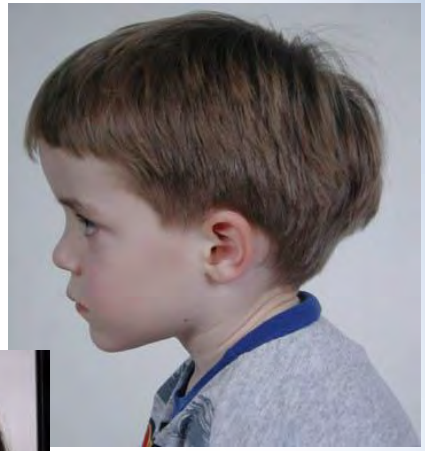
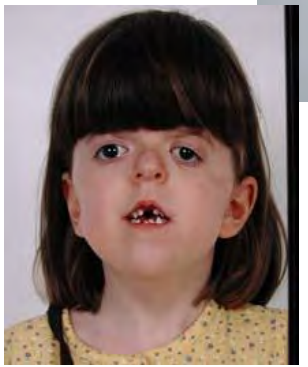
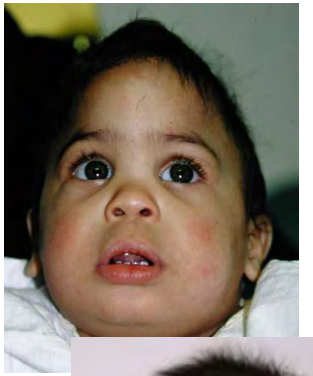
- YES! Children with non-syndromic synostosis have mild learning disability or motor development delay
  - Often detected around preschool years when a higher level of functioning is required

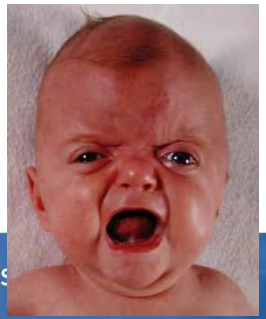
Cohen, S.R., et al. (2004). American Society of Maxillofacial Surgeons Outcome Study: Preoperative and postoperative neurodevelopmental findings in single-suture craniosynostosis. *Plastic and Reconstructive Surgery, 114*(4), 841-849.

Panchal, J., et al. (2001). Neurodevelopment in Children with Single-Suture Craniosynostosis and Plagiocephaly without Synostosis. *Plastic and Reconstructive Surgery, 108*(6), 1492-1498.

Kolar, J., et al. (1998). Long-term studies of craniosynostosis: Frequency of cognitive impairment and behavioral disturbances. *Plastic and Reconstructive Surgery, 9*[illegible] 276-281.

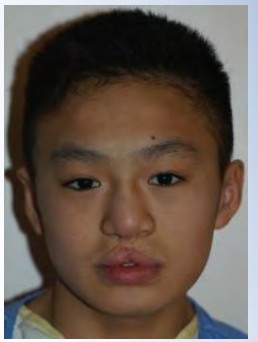
- Functional Concerns
  - ICP
  - Brain growth
  - Ocular
  - Airway
  - Aesthetics

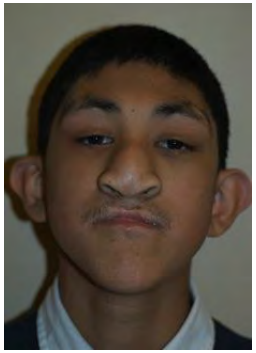





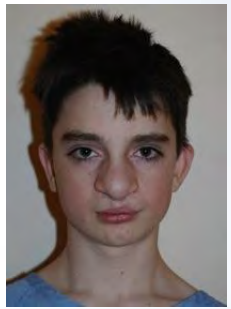


Children with facial differences

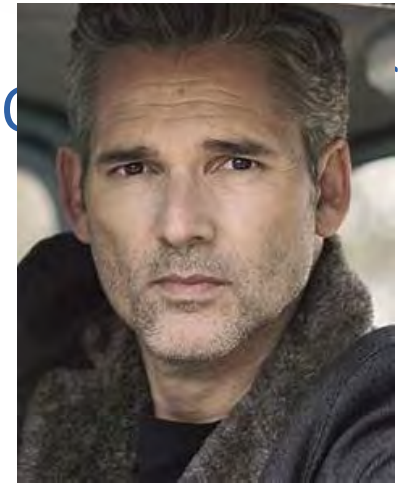
- Significant risk for social competence problems
  - Development of friendships
  - Progress in school
  - Participation in organizations
  - Social withdrawal
  - Negative effect upon development
  - Unhappiness and dissatisfaction
  - Lower problem solving ability
  - Poorer decision making
  - Less social competence
  - Low global self-esteem scores
  - More frequent negative social behaviour

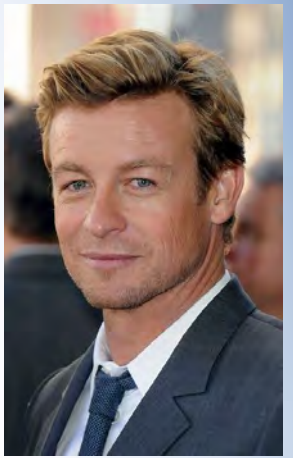




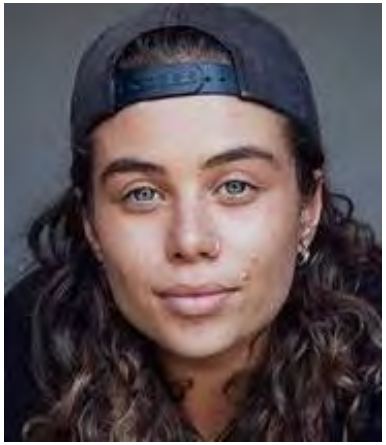




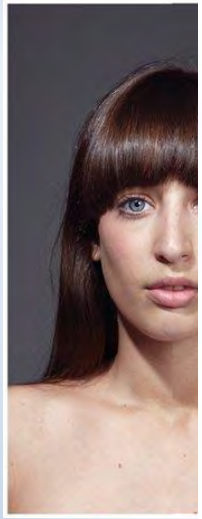









Symmetry



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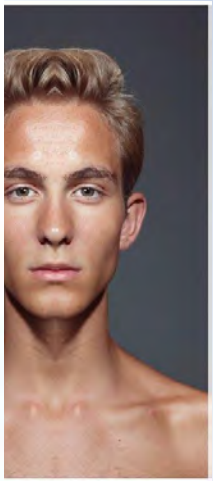
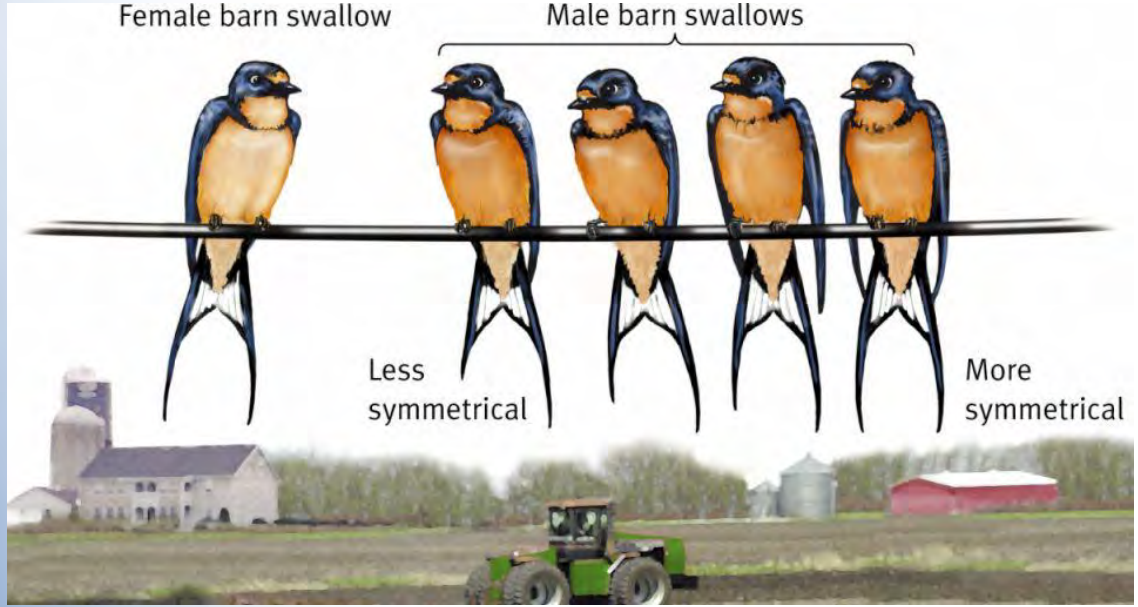
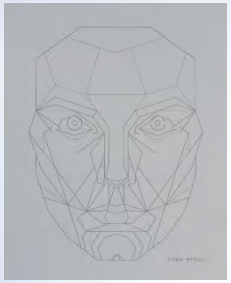


Attractiveness

- Symmetry:
  - Significant biological advantage (humans and animals)
  - Biological ad to having good genes
  - Facial symmetry linked to agreeableness, extraversion and conscientiousness
  - Male body symmetry associated with reproductive advantages (greater sperm counts, increased sperm speed)
  - Female breast symmetry associated with increased fertility

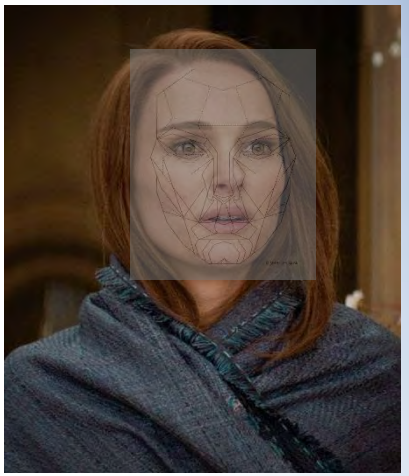
Attractiveness





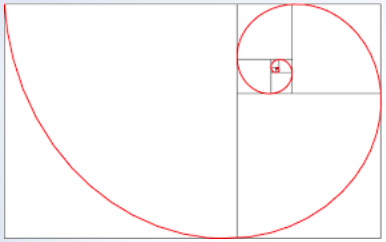
Attractiveness

- There are other factors besides symmetry....
  - Looking average (fitting in)
    - Mathematically average features
      - distance between your eyes should be 46 percent of your face's width
      - distance between your eyes and mouth to be about 36 percent of the length of your face

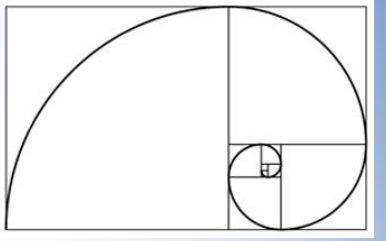


http://www.telegraph.co.uk/science/2016/03/12/the-science-of-sexiness-why-some-people-are-just-more-attractive/

Attractiveness



Attractiveness



- And it happens so fast!!!!
- Perception of the human face is unlike that of any other object
- “the most entertaining surface on earth”
- There is no other object that attracts our attention so rapidly
- 40 milliseconds to make a judgement
- 3 fundamental dimensions
  - Attractiveness
  - Trustworthiness
  - Dominance

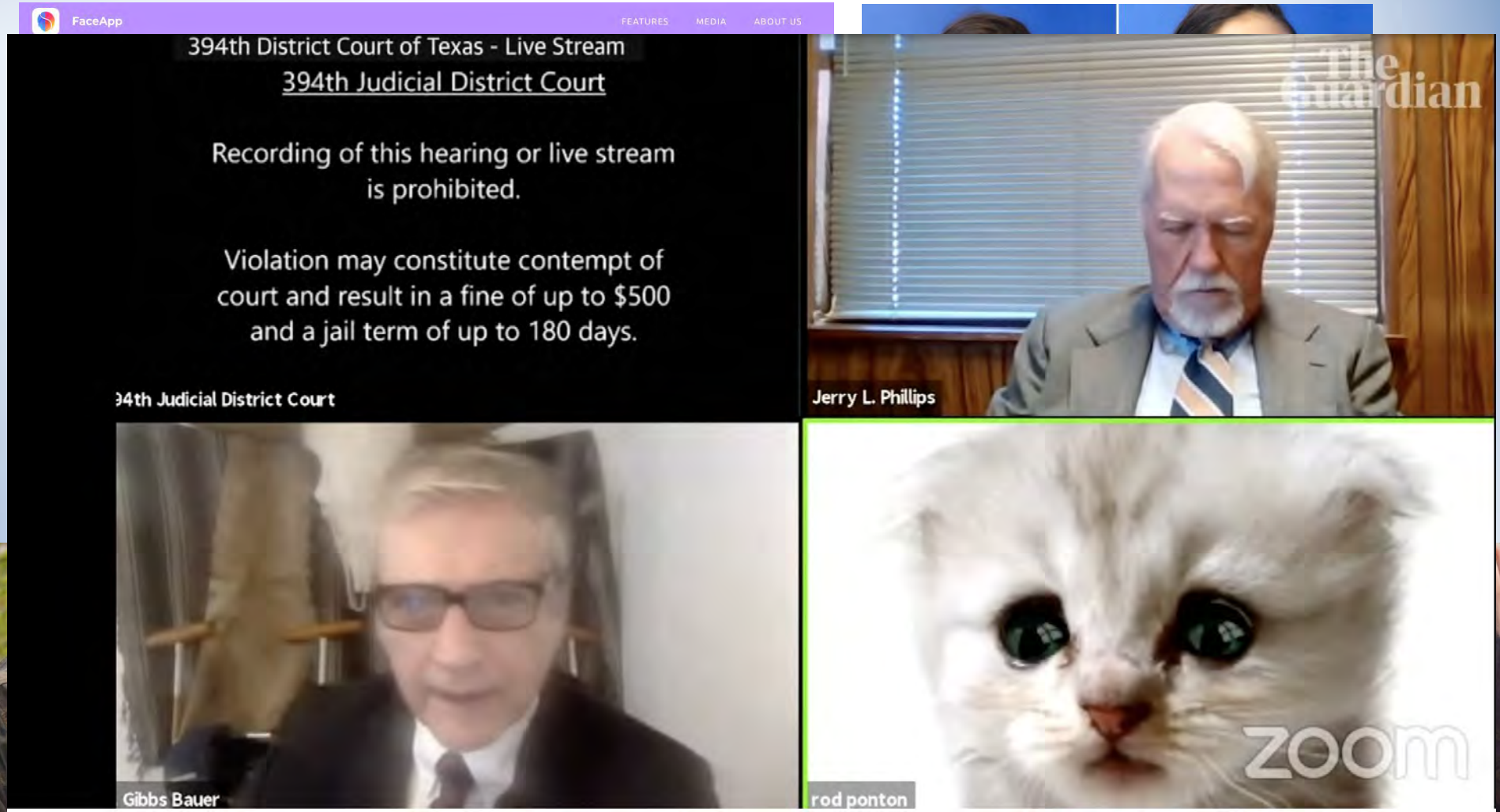
Face Value: Alexander Todorov, Princeton University Press, 2017

Attractiveness

- Associated with
  - Upward economic mobility
  - Greater likelihood of being hired
  - Enhanced social standing
  - Influence on perception and judgement
  - Lower bail payments

Little AC, Jones BC and Debruine LM. Facial attractiveness: evolutionary based research. Philos Trans F

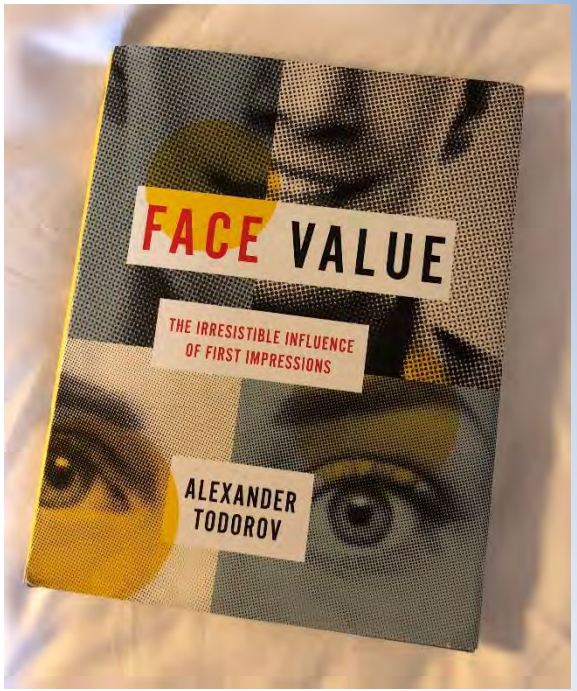
EB: 11 yo F: Apert syndrome: s/p ACVR/FOA, and Monobloc and CAD-CAM cranioplasty



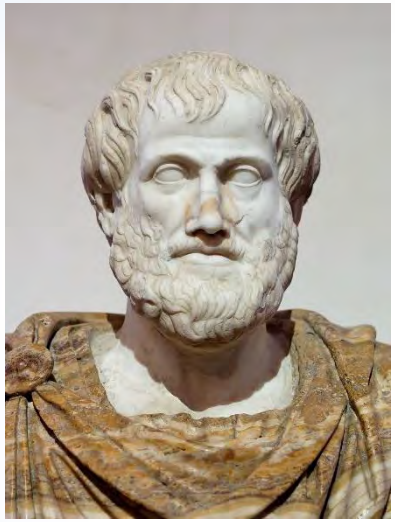
'I'm not a cat': lawyer gets stuck on Zoom kitten filter during court case

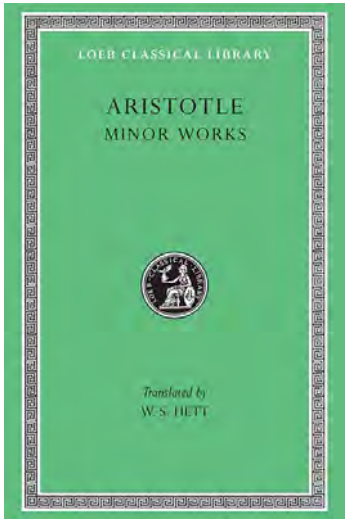
YouTube · 13,624,000+ views · 2021-02-09 · by Guardian News

Save



"impressions of the face are immediate and compelling and they are consequential"





The forehead that riseth in a round, signifies a man liberally merry, of a good understanding, and generally inclined to virtue.
He whose forehead is fleshy, and the bone of the brow jutting out, and without wrinkles, is a man much inclined to suits of law, contentious, vain, deceitful, and addicted to follow ill courses.
He whose forehead is very low and little is of a good understanding, magnanimous, but extremely bold and confident, and a great pretender to love and honour.

Aristotle: Greek philosopher and scientist: 384 BC – 322 BC

Angusta frons.

ARISTOTELES in suis Physiognomonic. Paruæ frontis mili suum fronte. Sed per paruam frontem angustam intelli enim fronte sues, quibus comparantur, præditi sunt, non paru mram Polemon, & Adamantius, qui inquiunt. Angusta frons non gumentum. Conciliator. Frons parua, angustaq; stolidum, indocilem tum hominem monstrant, ad sues relatum. Albertus eadem.

Planæ frontis simulacrum hic dextrorsum à dextris.

In hac figura media magnitudinis caput inter reliqua cum leo

Caput mediocre.

ARISTOTELES commendat Alexandro Mag sum constitutum, cum Polemon & Adamantius m Id videtur Leoni comparandum, ipse enim ad corpor gnitudine est: vt apud Aristotelem videre est, leonis formam Moderatum caput indicat ingenium, & sensum, aliquando sed ego non timidum, sed audacem, & magnanimum ad leon

Prima hac tabella in dextra sui parte non naturalem capitis formam refert, in immo similtra veri secundum capitis innaturalem formam, in qua pollica

Hos ego etiam puerarios iudicarem: abutuntur enim venere galli, gallinacei, perdices, & coturnices, quæ ferè similem nasum habent. Cum enim fœmina incubant, mares dimicant, pugnamque inter se conserunt, quos cælibes vocant, qui victus in pugna fuerit, victoris venerem patitur, nec nisi à suo victore subigitur, ex Aristotele: & multos amicos cognoui, eiusmodi naso præditos, huic enormi luxuriæ generi obnoxios. Fingunt Poetæ Iouem Aquilæ forma Ganimedem rapuisse, sub tali figmento id fortasse innuentes. Aelianus etiam ichneumonem huic turpitudini etiam obnoxium dixit. Tali naso Satyri, & Sileni ab antiquis effigiati sunt, & tali naso etiam Socrates ipse præditus fuit: nam Xenophon Socratem Silenis similem fuisse, & pressis naribus scribit.

Latus in medio nasus.

NASVS in medio latus, declinans ad summitatem, demonstrat mendacem, & verbosum. Aristoteles ad Alexandrum.

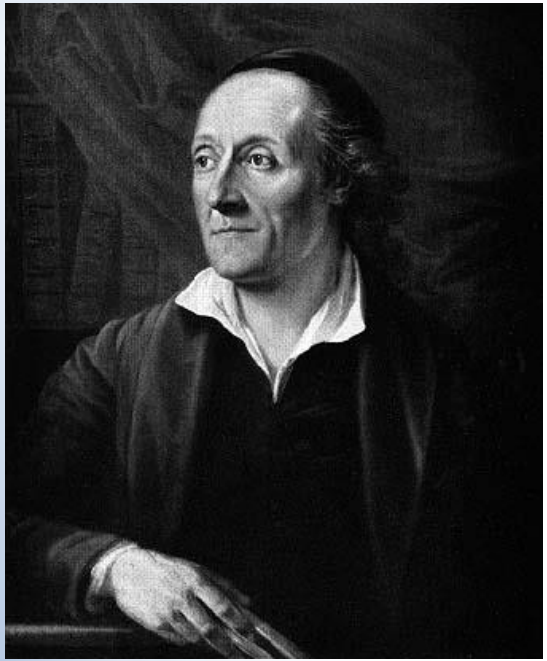
Si oculi nasum inspexerimus, & hominem similem effigiauerimus, vti ab hac, quæ hic cernitur figura, longe aberit, ita affabre in imo crassus deducitur.

Extremum nasi crassum.

QVI nasi extremum crassum habent, segnitiei obnoxij sunt, & ad boues referuntur, vt Aristoteles in Physiogn. est enim bouum segnities propria. Polemon, & Adamantius. Nasus in summo valde crassus, et depressus, iniquos homines ostendit. Interim Polemoni textus corrigatur, μεσῳ, non μικρῳ ex Adamantio. Idemq; in figura inuerecundi tribuunt ei nasum crassum.

Suis crassus à summo insigitur hic nasus, è cuius regione hominis similitudine petendus.

A sum-



J. C. Lavaters

Physiognomische

Fragmente

zur Beförderung der Menschenkenntniß und Menschenliebe.

Verkürzt herausgegeben von

Johann Michael Armbruster.

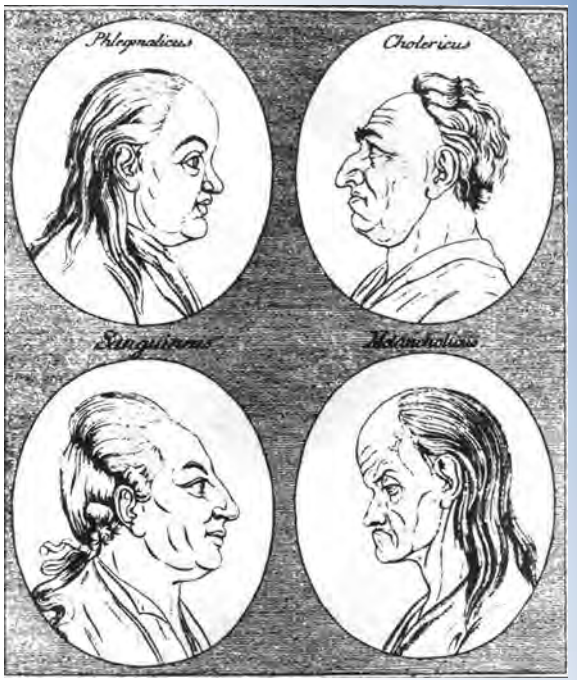
Erster Band.

Mit vielen Kupfern.

Winterthur.

In Verlag Heinrich Steiners und Compagnie

1783.

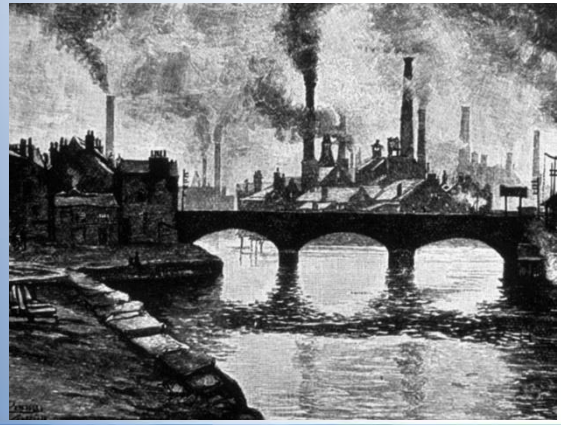


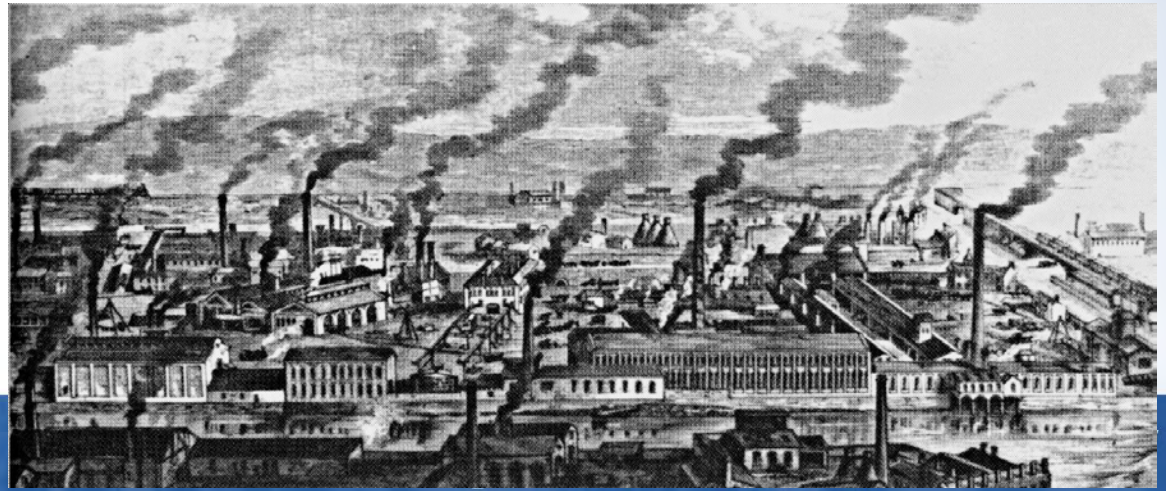
Physiognomische Fragmente zur Beförderung der Menschenkenntnis und Menschenliebe: 1775-1778

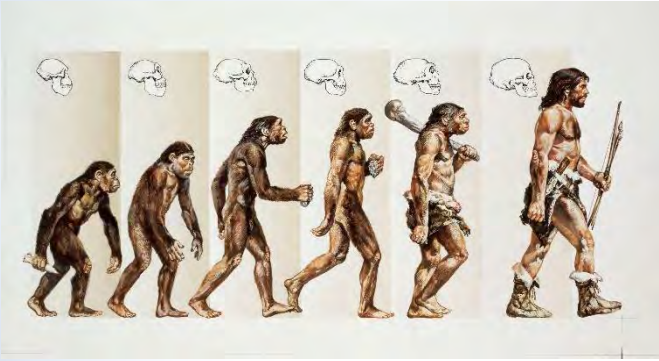
"the talent of discovering the, the interior man by the exterior appearance"

Johann Kaspar Lavatar: (15 November 1741 – 2 January 1801): Swiss poet, physiognomist, writer and theologian


The
Industrial Revolution
1760-1990

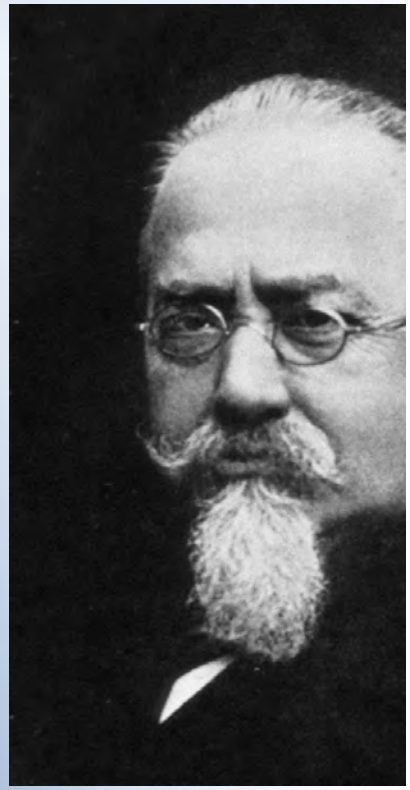


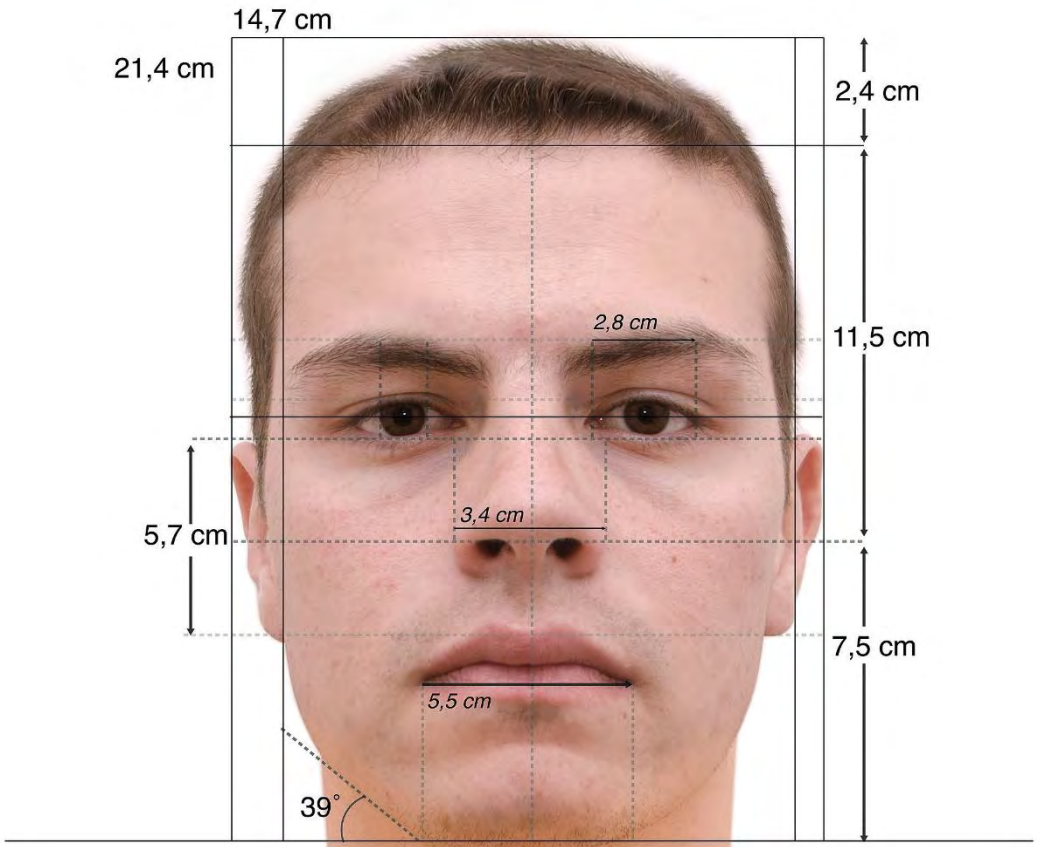




Todorov

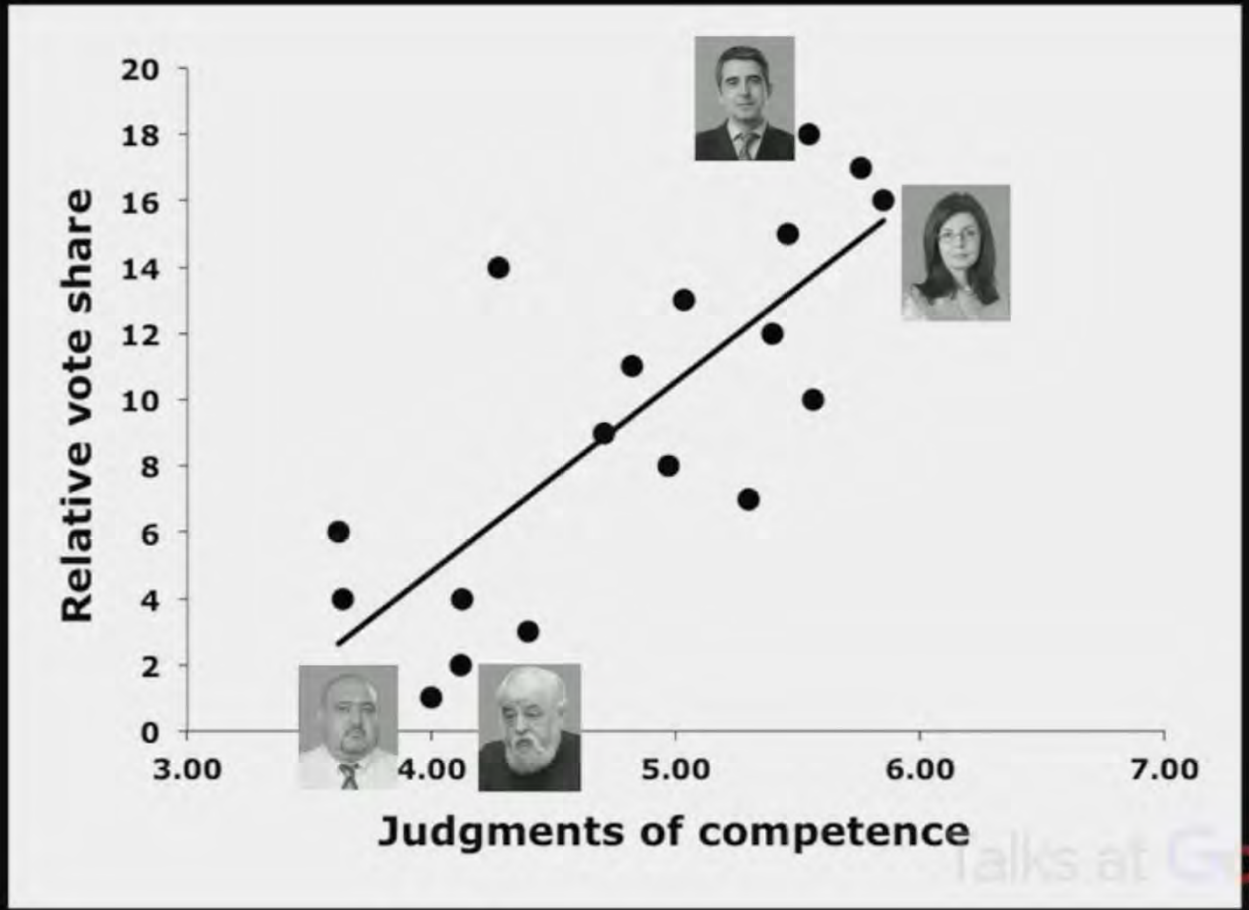
- Evolution of humankind in 24 hours
- Small-scale societies: no reliance on appearance to make inferences about character as everyone knew each other
- Concept of “living with strangers” – started with the migration
- Larger societies populated with strangers last 5 minutes/24 hours which forced a reliance on appearance information
- We all do it despite the inaccuracies



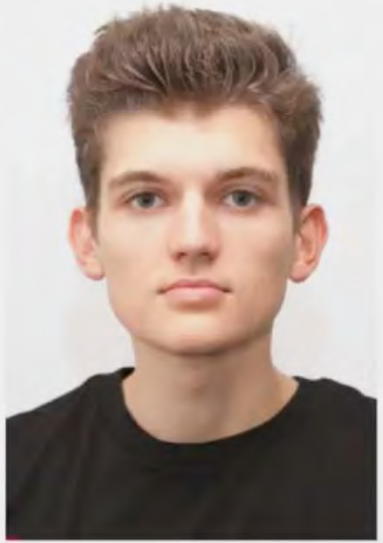

14,7 cm
21,4 cm
2,4 cm
11,5 cm
2,8 cm
3,4 cm
5,7 cm
7,5 cm
5,5 cm
39°

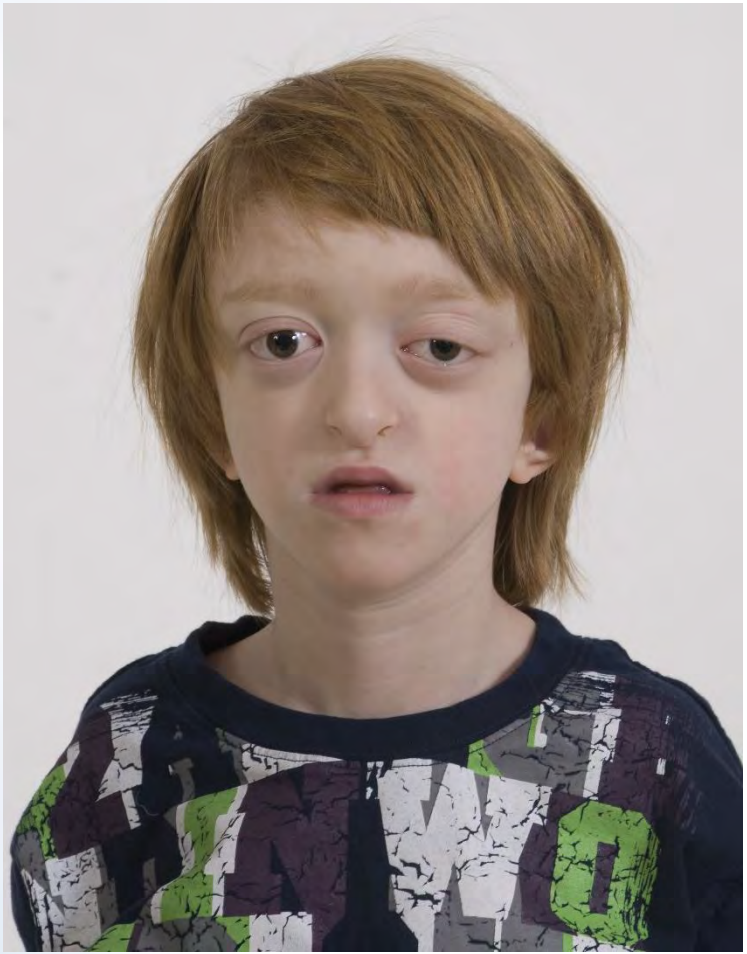

Pl. XLIX
— MATTOÏDES ET FOUS MORAUX.

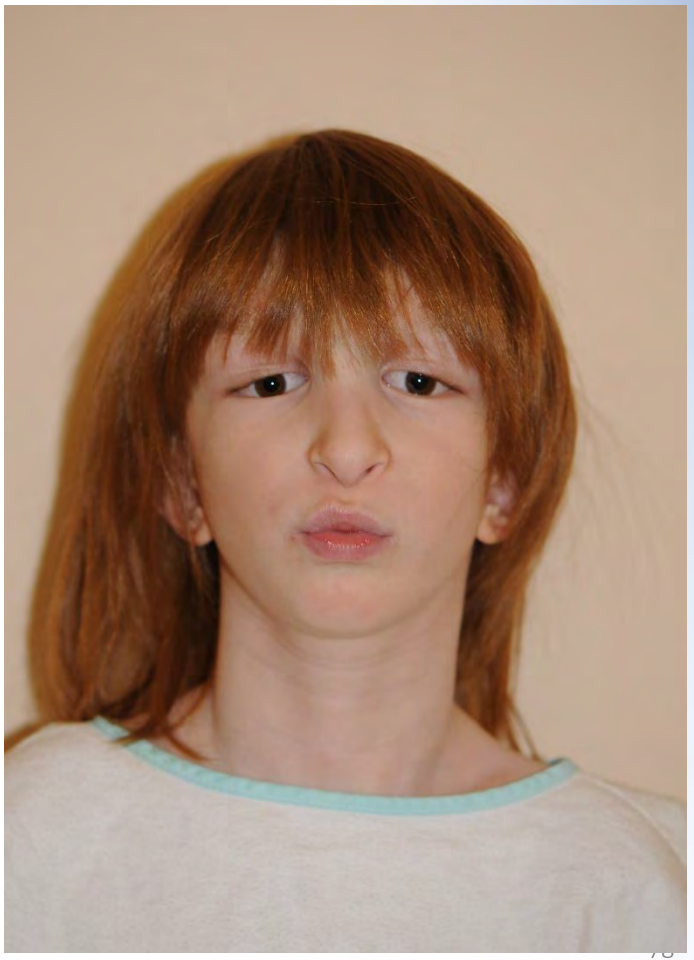
Cesare Lo an


Relative vote share
20
18
16
14
12
10
8
6
4
2
0
3.00
4.00
5.00
6.00
7.00
Judgments of competence

Who is more likely to have committed a violent crime?





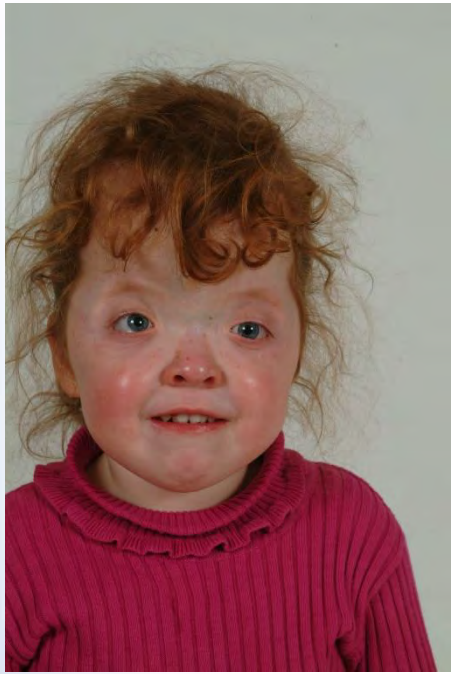


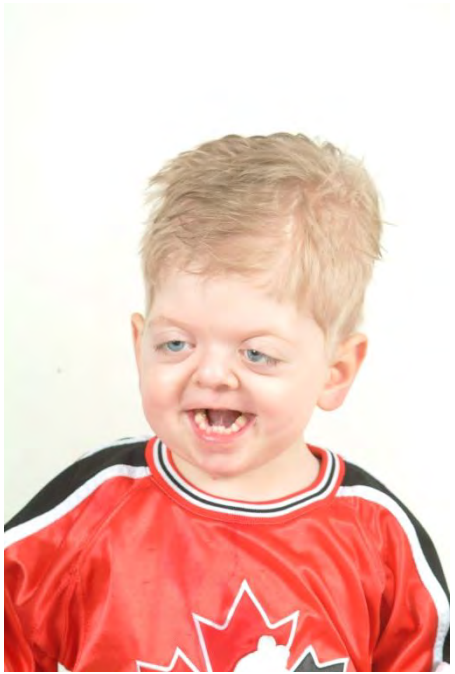

HD
TRAILER

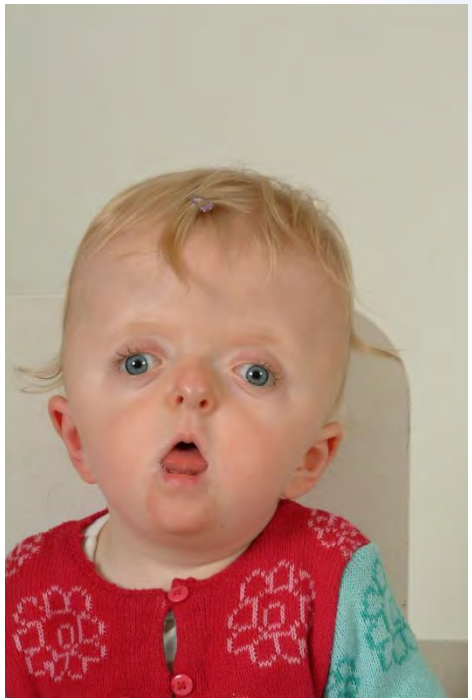
October 2016

November 2017

The importance of the face

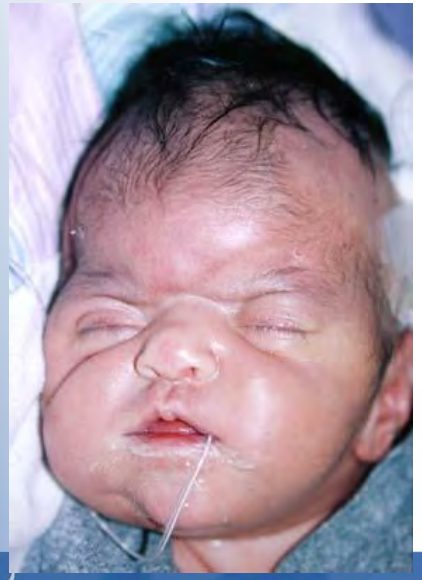


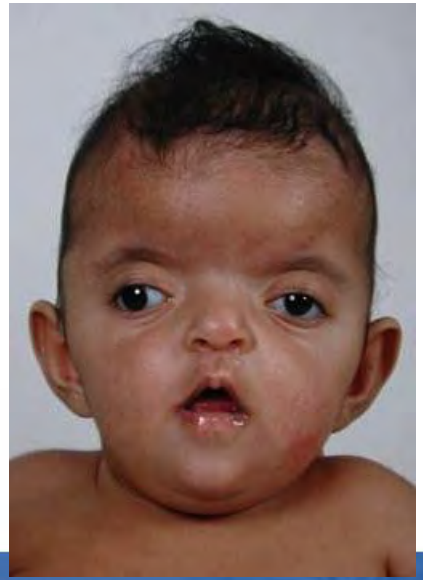


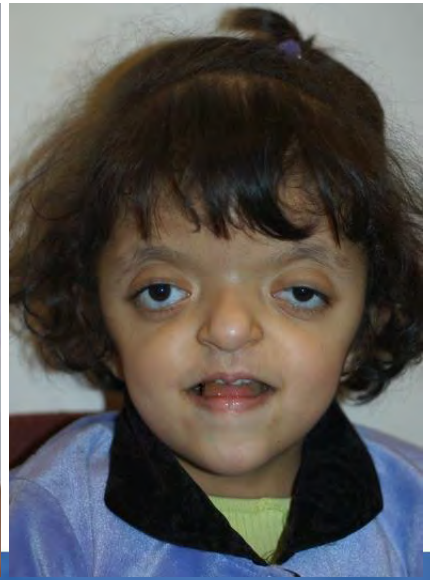


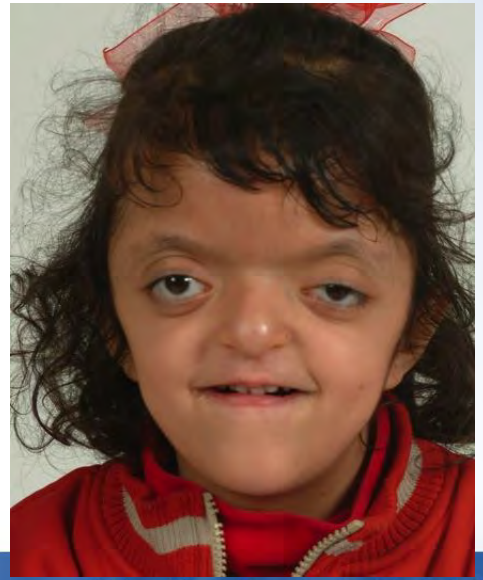
Craniosynostosis:

- Functional concerns
  - ICP
  - Airway
  - Visual
  - Neurodevelopmental
  - Aesthetic/Psychosocial
  - Natural History – no spontaneous improvement



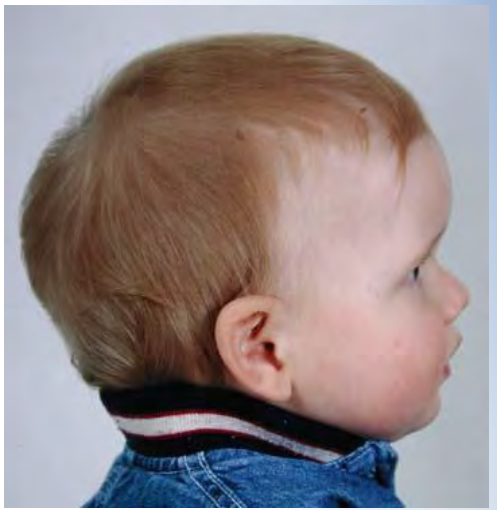


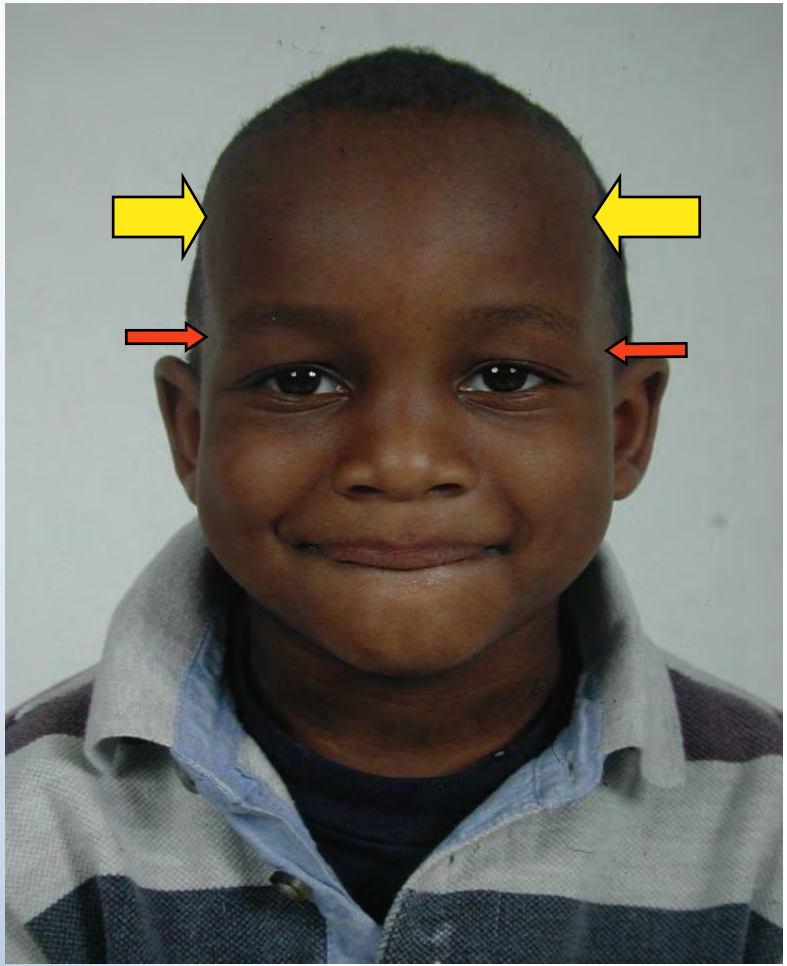


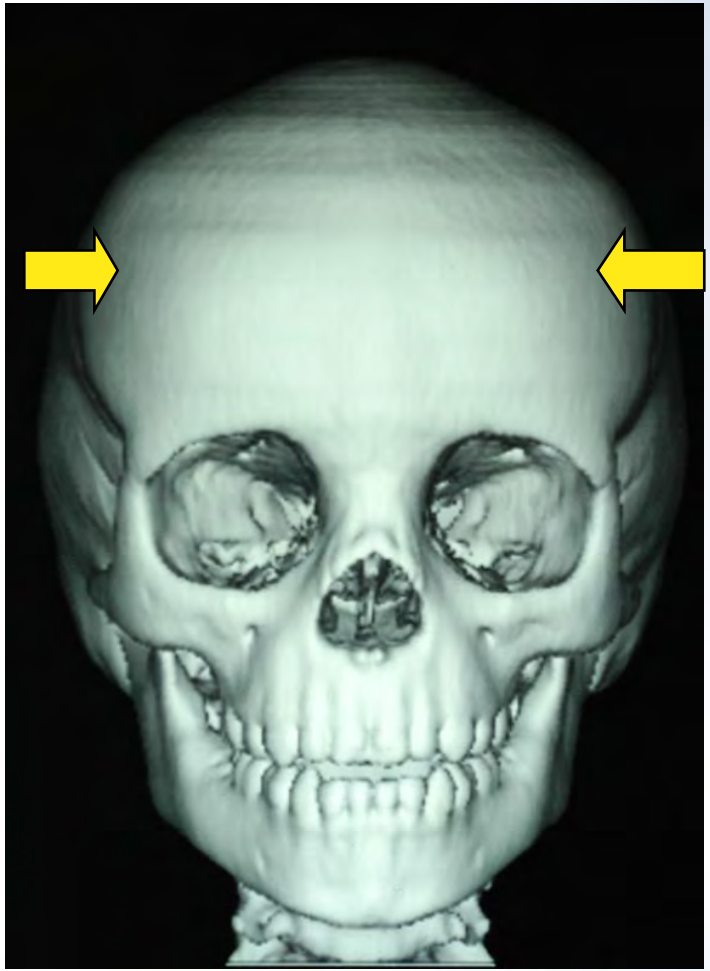


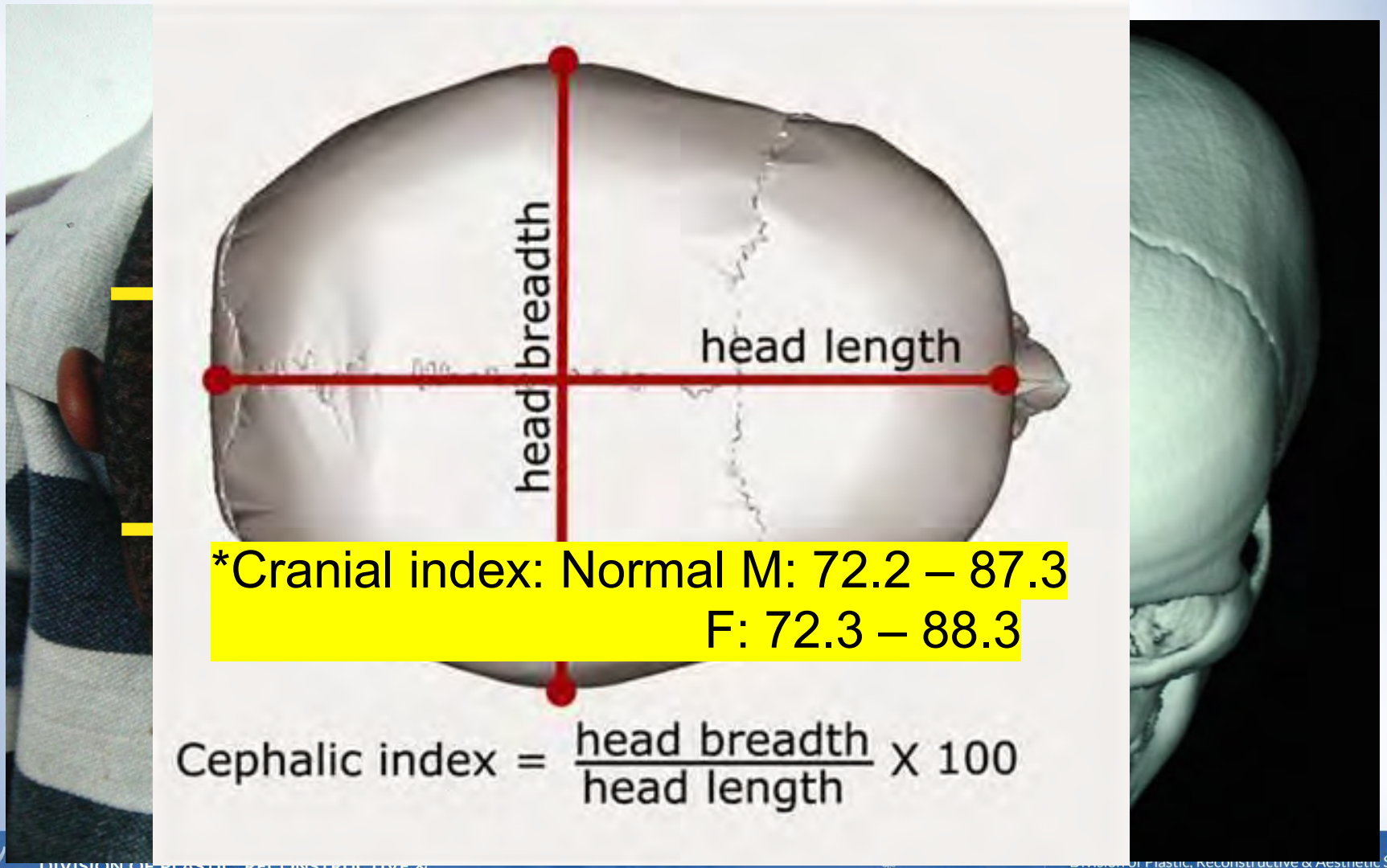
Sagittal Synostosis

- 1 in 2,000 -5,000 live births
- Commonest form of craniosynostosis
- Scaphocephaly - “keel-shaped”
- Male predominance
- Functional concerns
  - ICP elevation: 10 - 18% (?)
  - Speech issues: 29%
- Genetic basis – 2 to 8%
- Clinical features
  - AP elongation
  - Transverse narrowing
  - Bitemporal pinching
  - Forehead bossing
  - High forehead
  - Prominent occipital cap
  - Saddle deformity – mid skull
  - Palpable ridging along suture



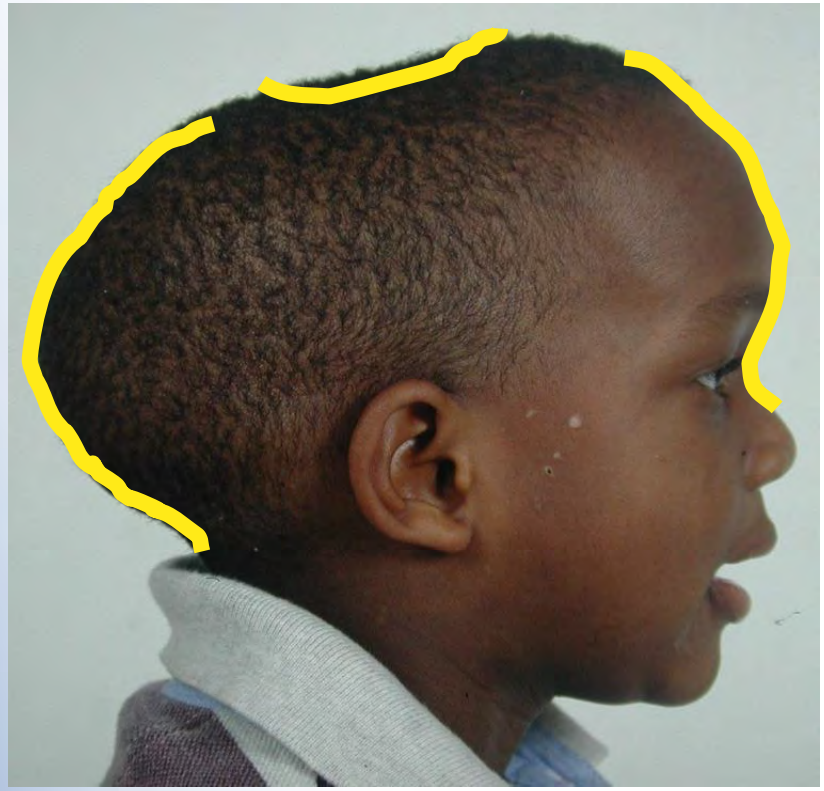


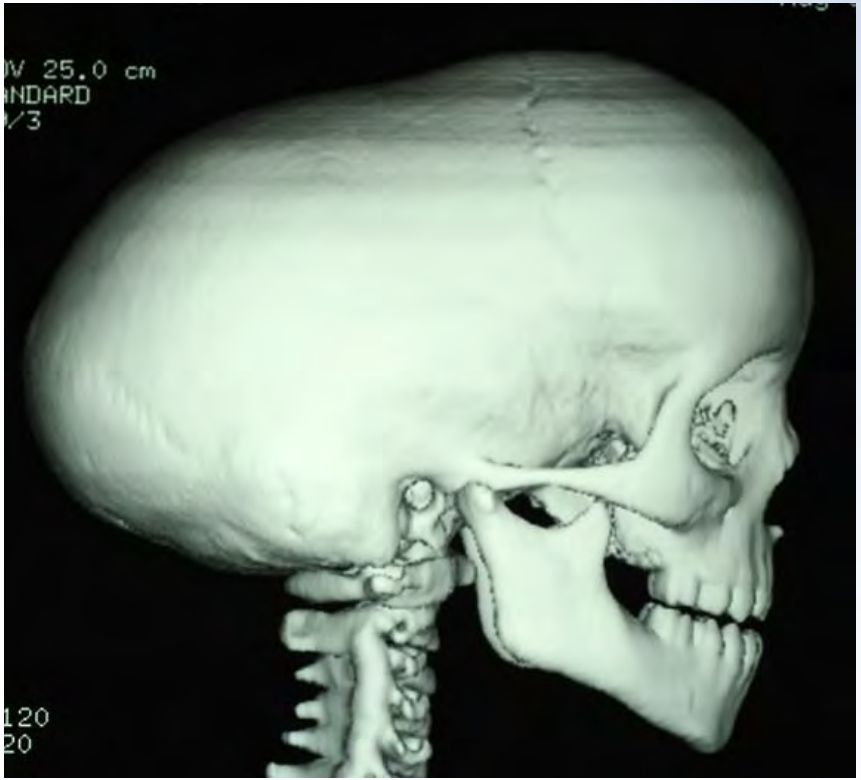




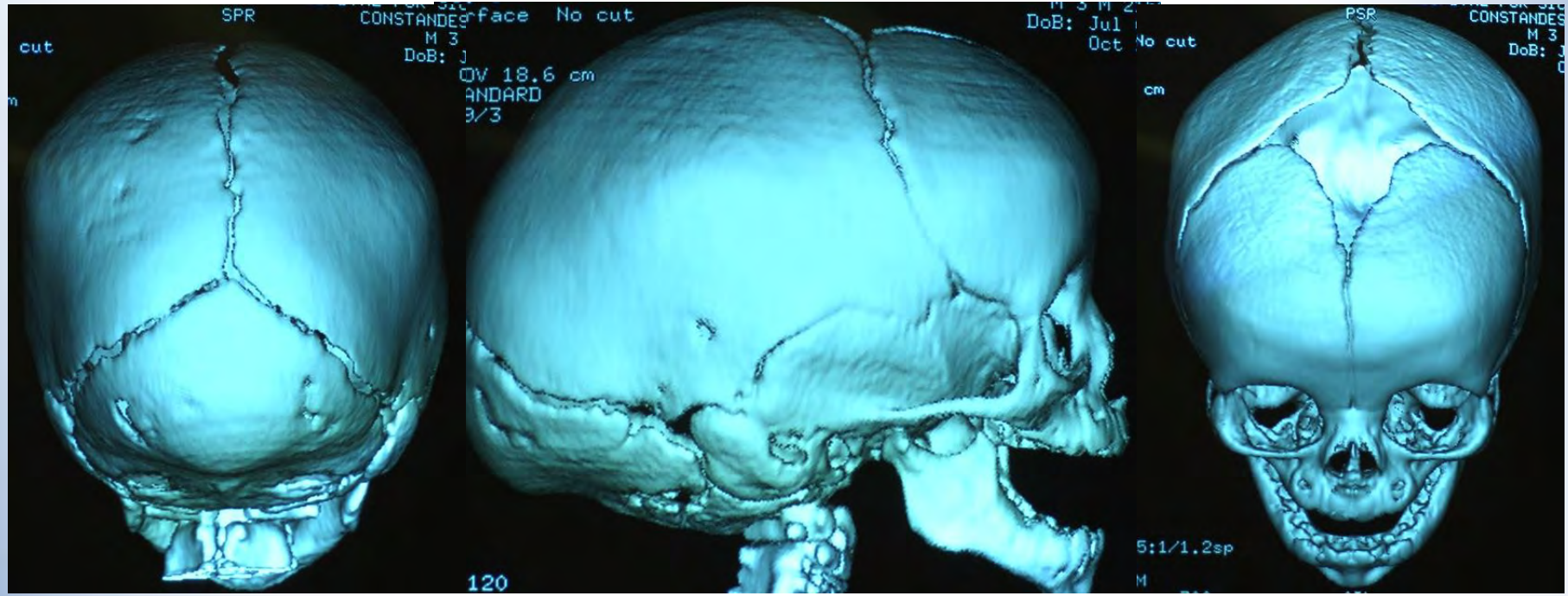
*Cranial index: Normal M: 72.2 – 87.3
F: 72.3 – 88.3

$$\text{Cephalic index} = \frac{\text{head breadth}}{\text{head length}} \times 100$$

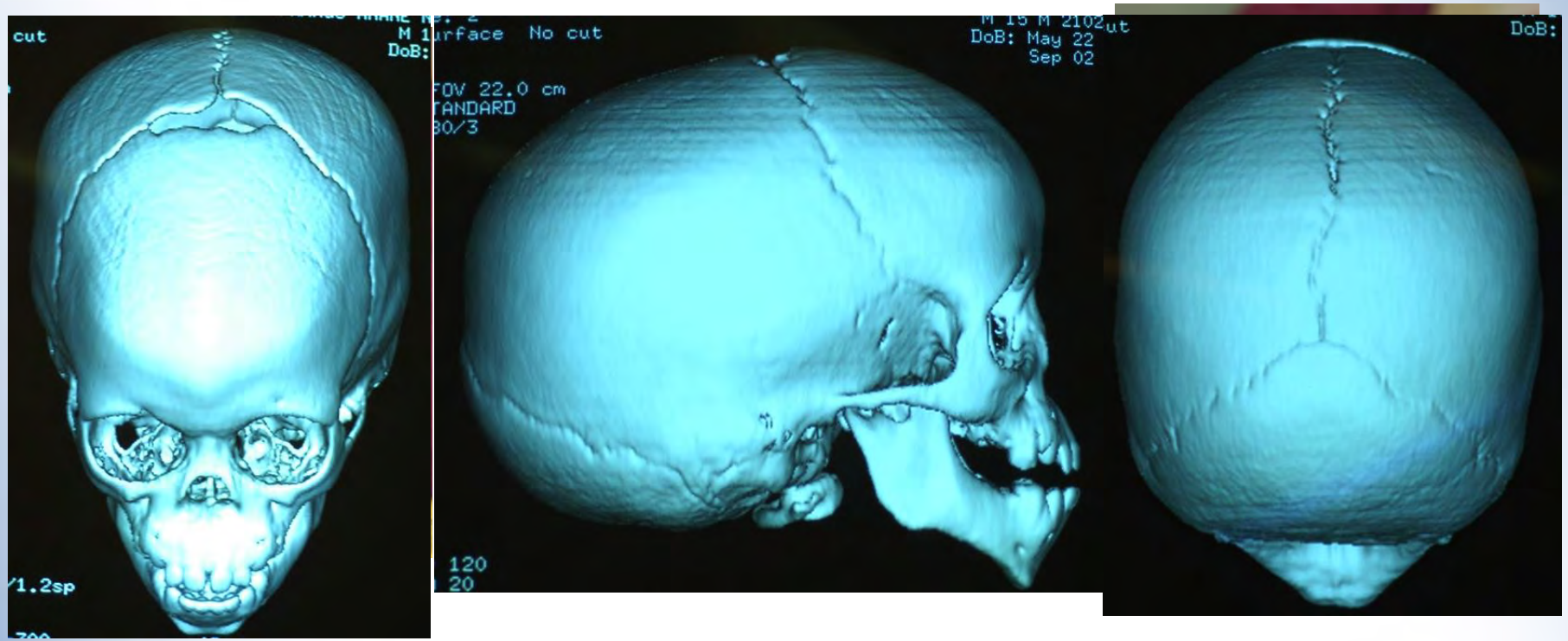



V 25.0 cm
NDARD
/3
120
20

But not all scaphocephaly is due to sagittal synosytosis…

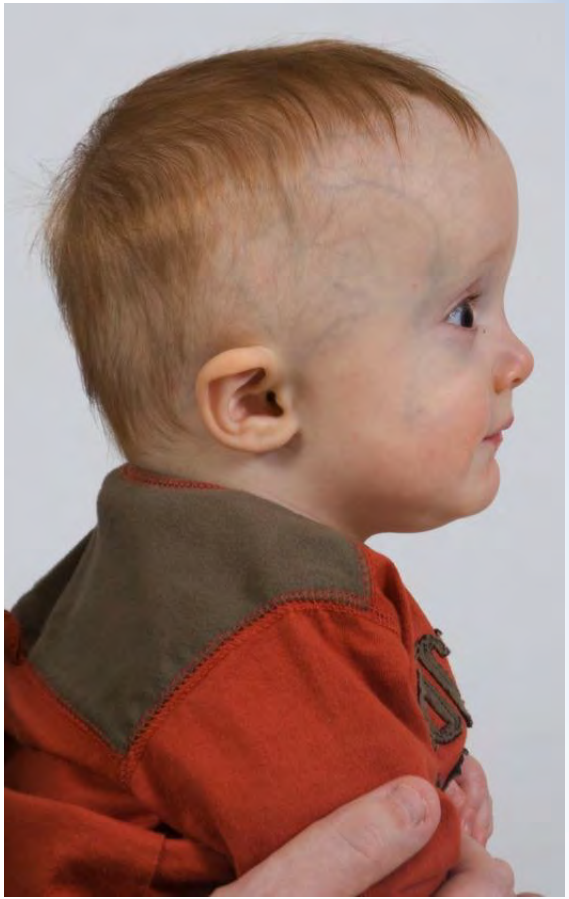


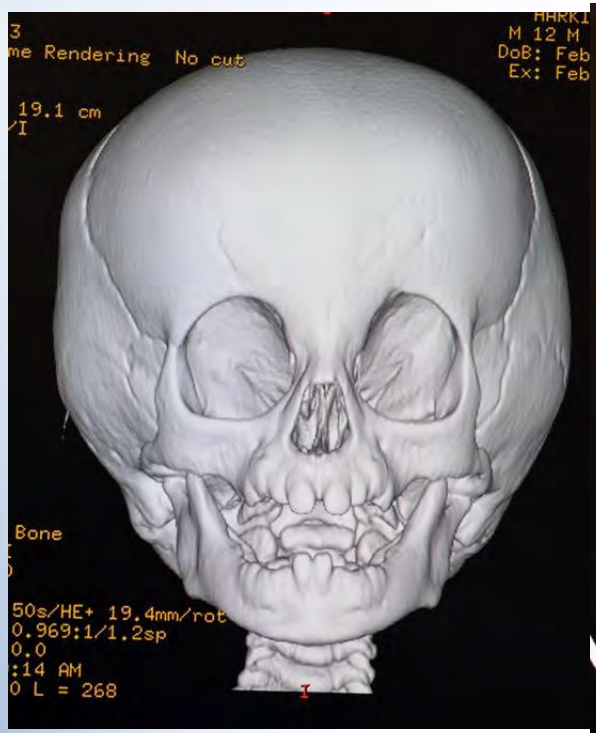
Positional scaphocephaly

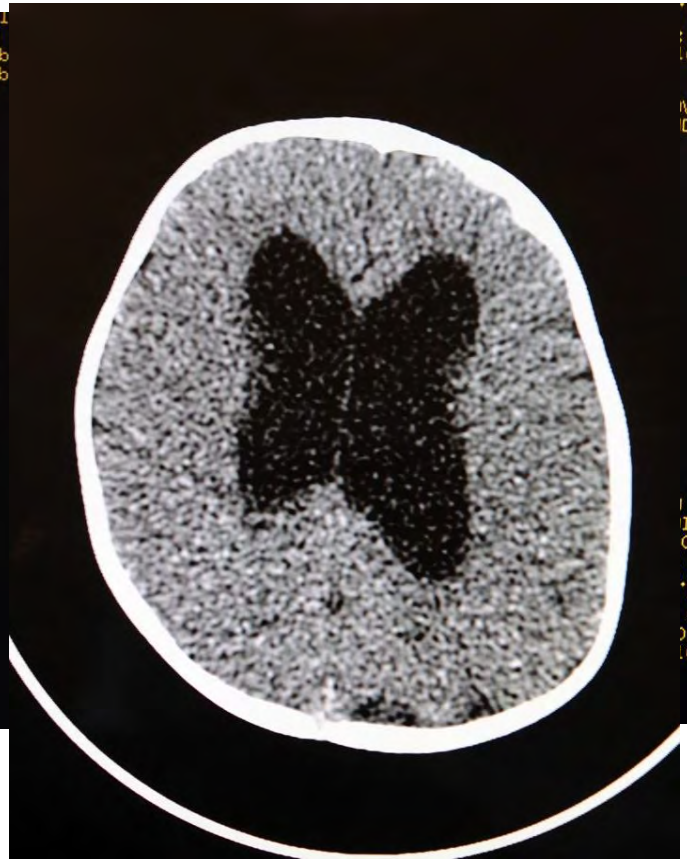


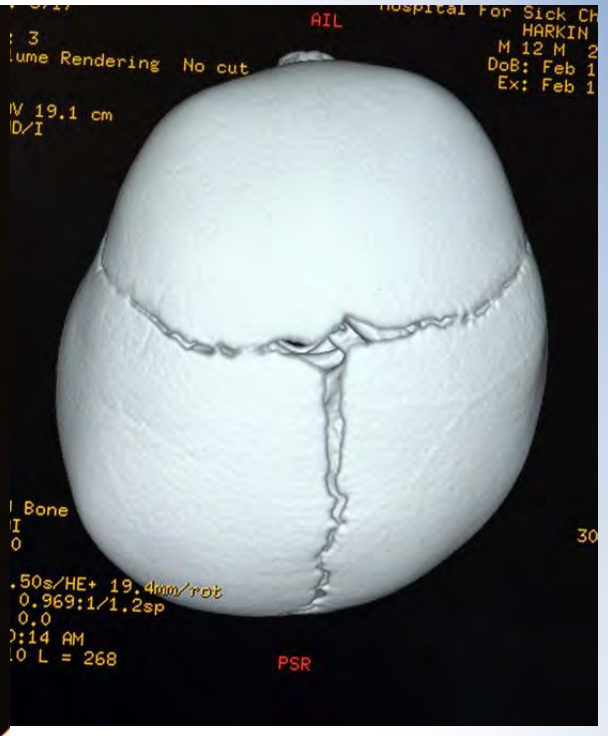
Vitamin D deficiency - Ricketts


HOCKEY





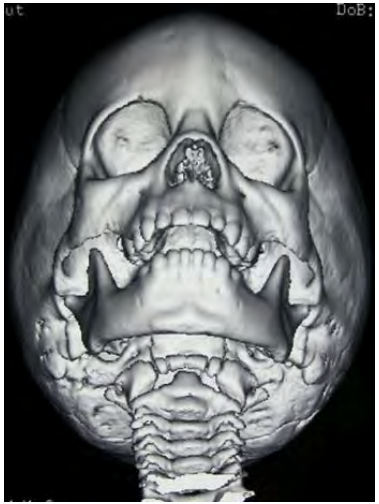


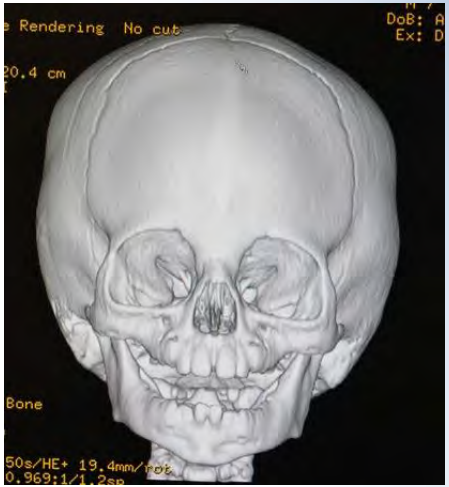


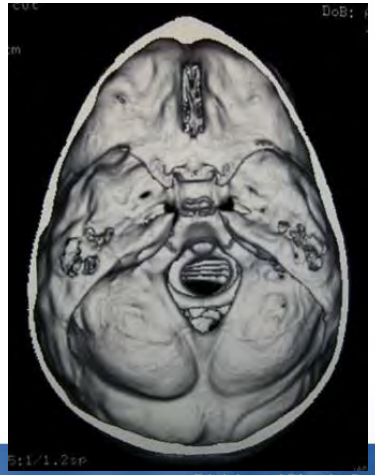
Extra-ventricular obstructive hydroceph

Metopic Synostosis

- Metopic suture fused by 8 months
- First suture to normally fuse
- Diagnosis made at birth
- 1 in 15,000 infants
- Change in demographics (more common than UCS)
- Males > females: 3:1
- Genetics
  - Positive family history 6% of cases
  - Associated with syndromes
    - Opitz trigonocephaly
    - Craniotelencephalic dysplasia
    - Say-Mayer syndrome
    - VSR syndrome
  - Associated anomalies 15% of cases
    - Agenesis of corpus callosum
    - Cardiac
    - Extremity
    - GU

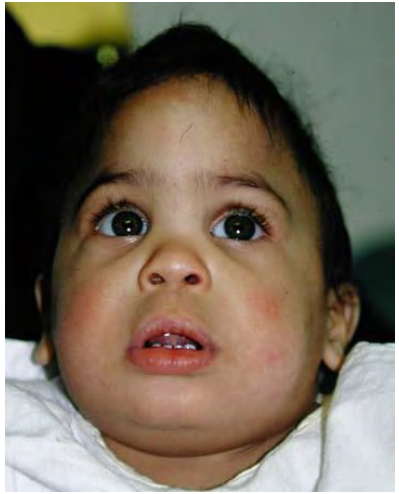


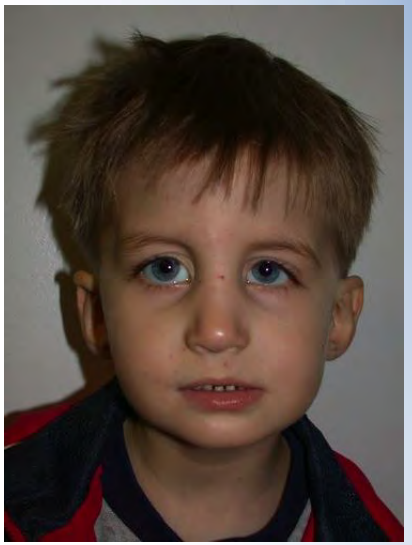


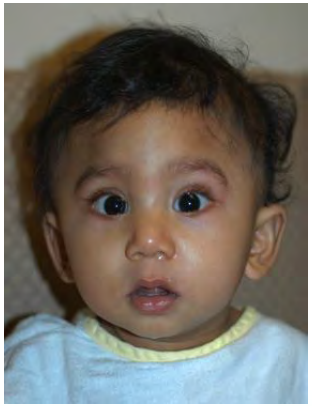


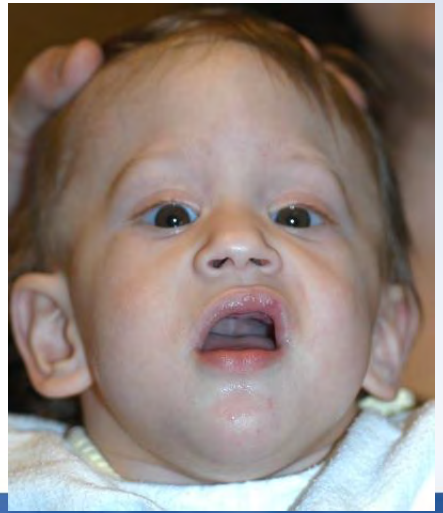
Metopic Synostosis

- Issues
  - ICP
    - 8% - 15% affected (?)
  - Aesthetic
  - Developmental
    - 31% - 33% measurable level of cognitive impairment c/w 10% in unaffected population (?)



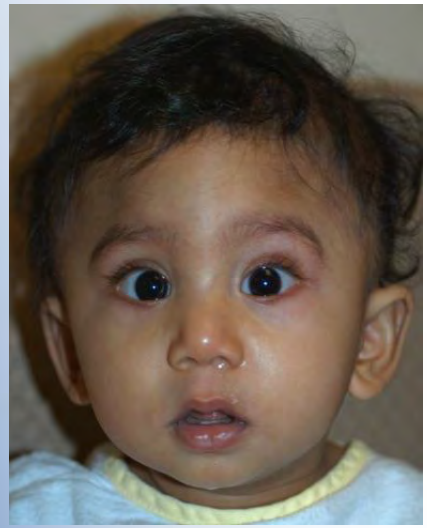
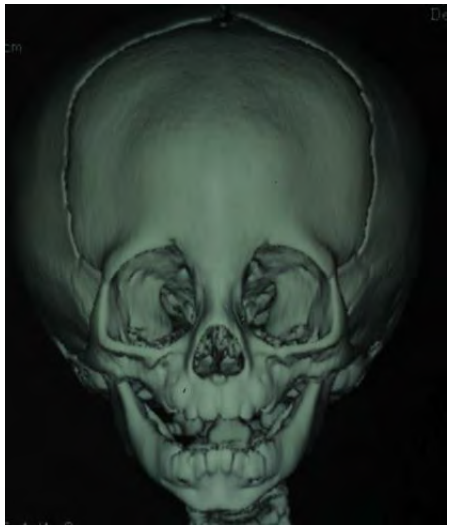


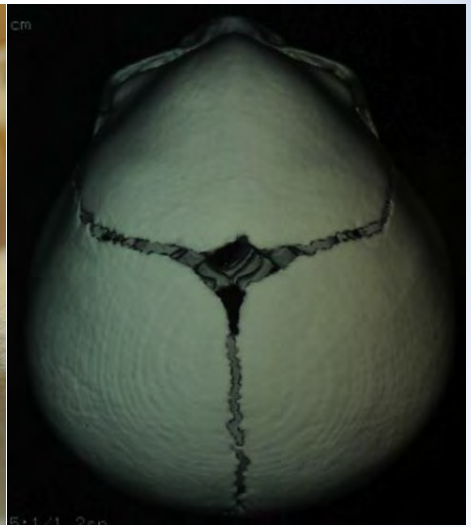




Metopic Synostosis

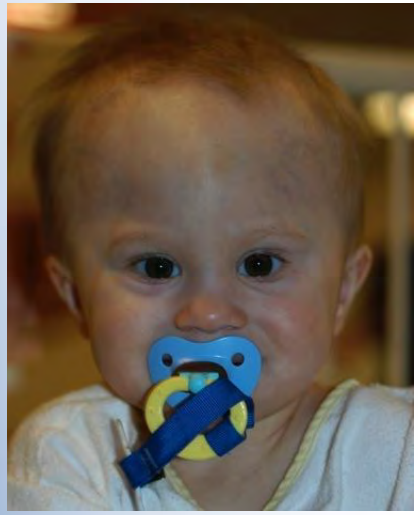
- Clinical Features
  - Forehead keel/ridge

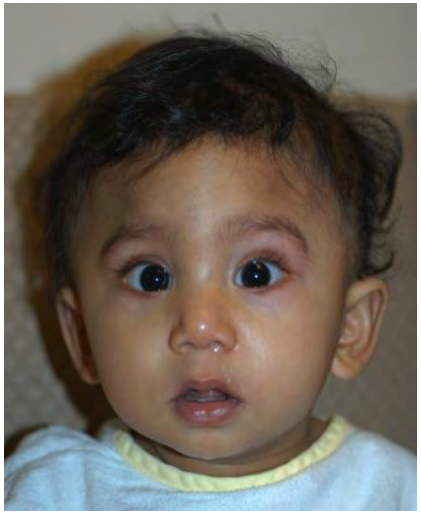



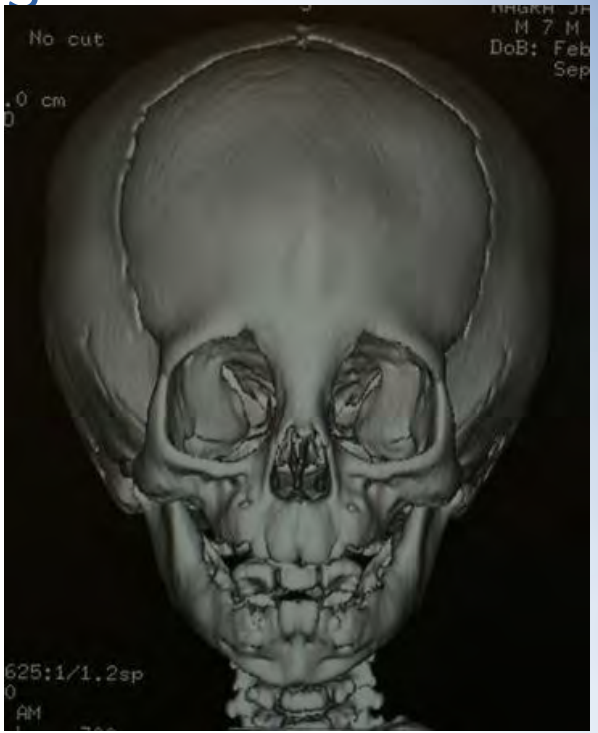


Metopic Synostosis

- Characteristics
  - Forehead keel/ridge
  - Hypotelorism

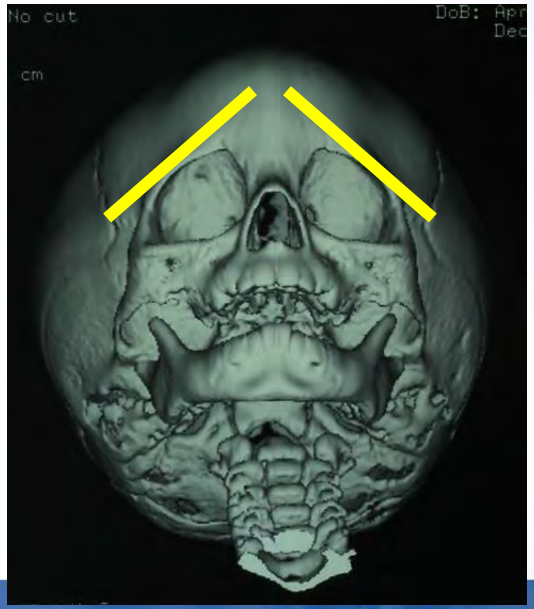






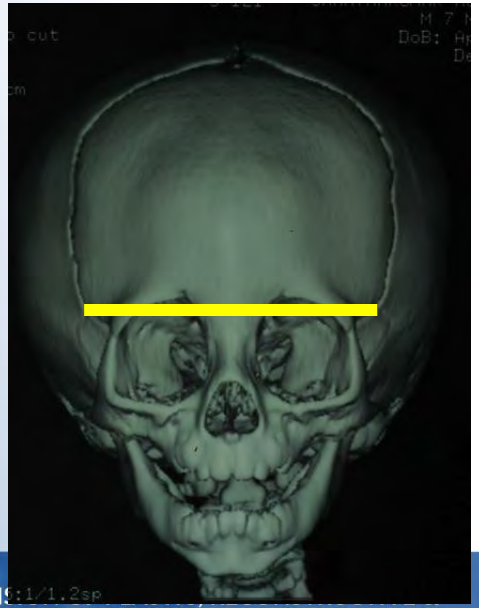
Metopic Synostosis

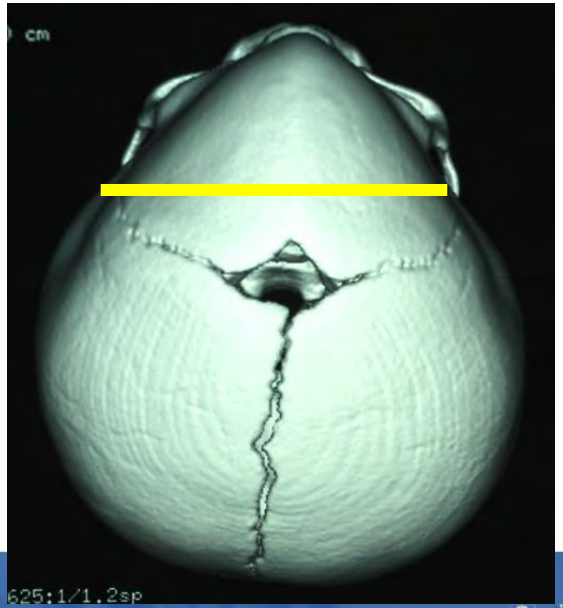
- Characteristics
  - Forehead keel/ridge
  - Hypotelorism
  - Lateral orbital recession

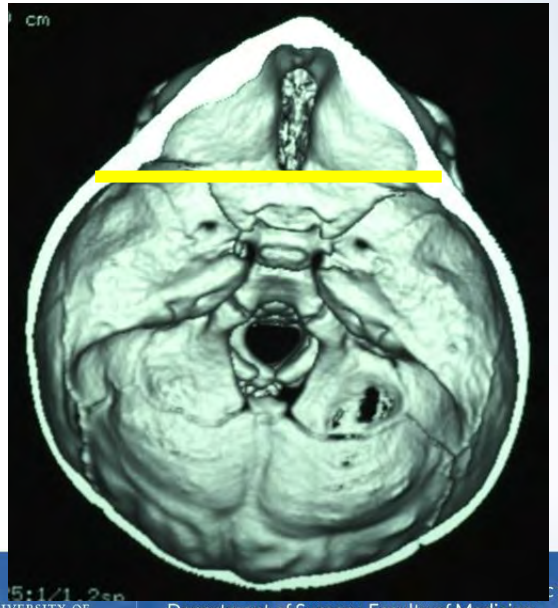


Metopic Synostosis

- Characteristics
  - Forehead keel/ridge
  - Hypotelorism
  - Lateral orbital recession
  - Bitemporal narrowing

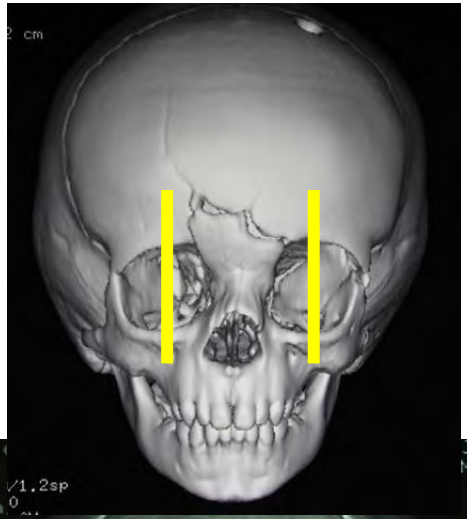


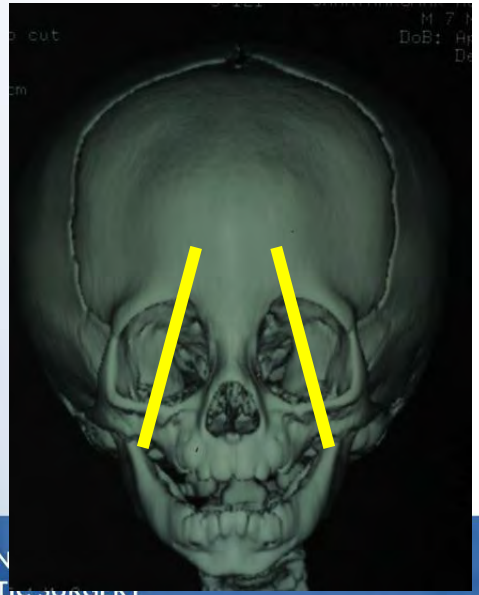


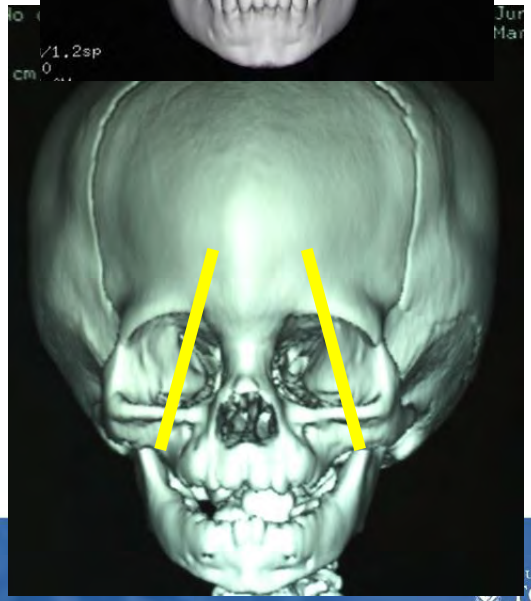


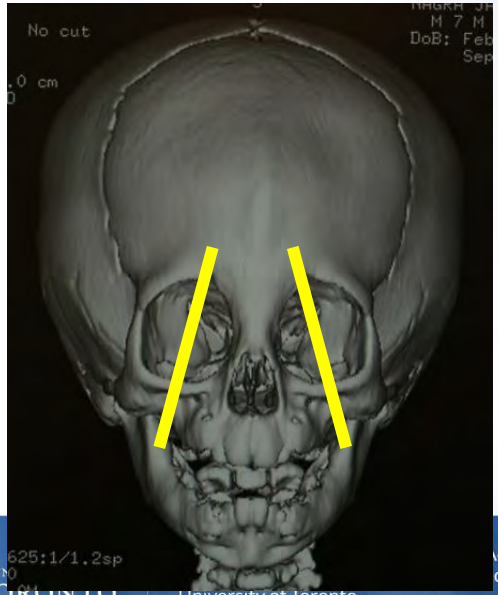
Metopic Synostosis

- Characteristics
  - Forehead keel/ridge
  - Hypotelorism
  - Lateral orbital recession
  - Bitemporal narrowing
  - Orbital configuration



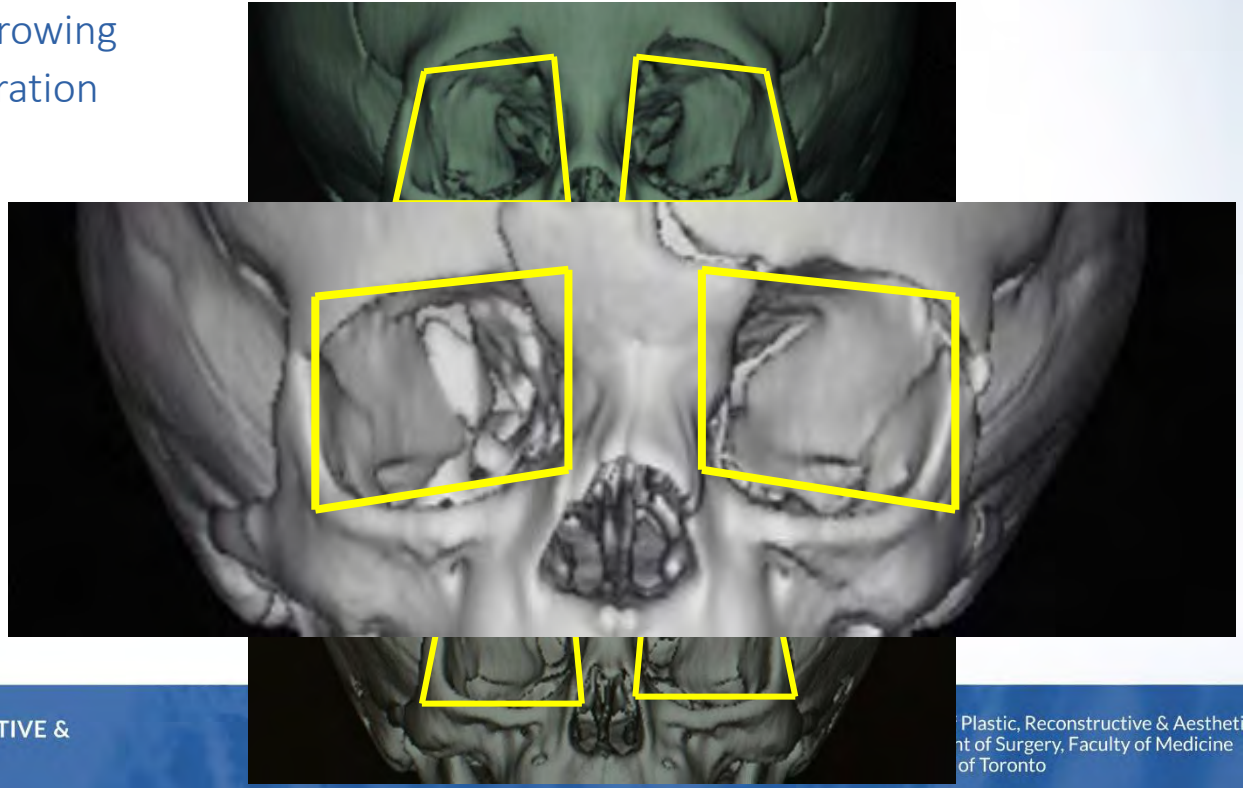






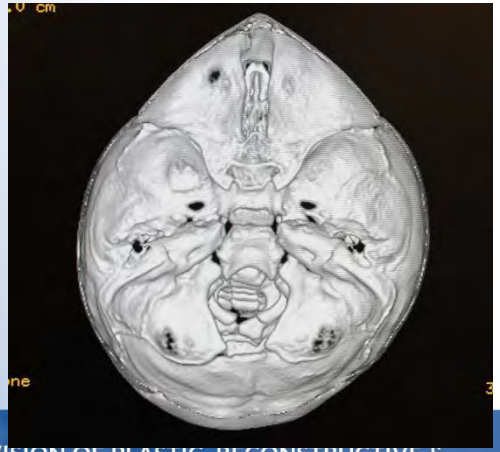
Metopic Synostosis

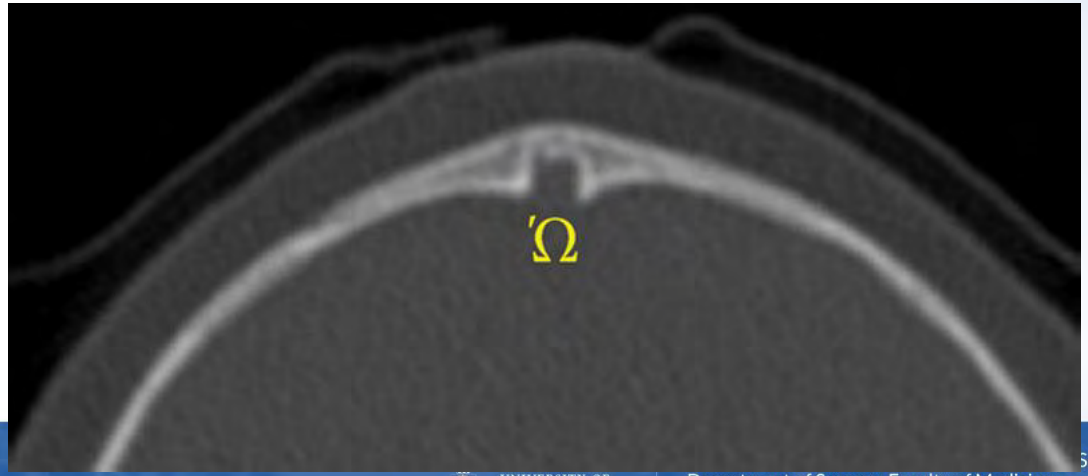
- Characteristics
  - Forehead keel/ridge
  - Hypotelorism
  - Lateral orbital recession
  - Bitemporal narrowing
  - Orbital configuration



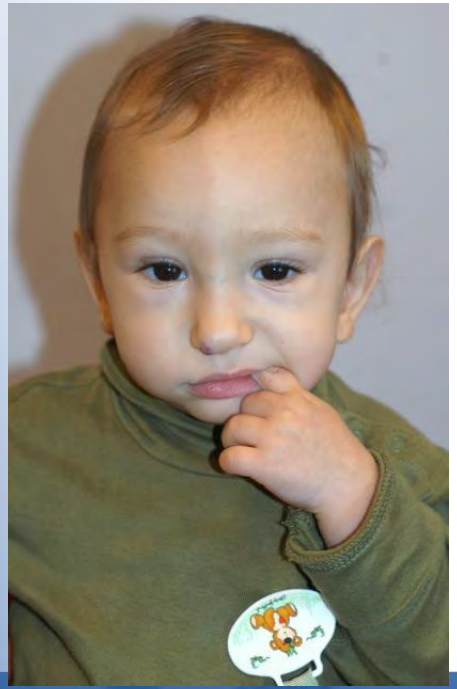
Metopic Synostosis

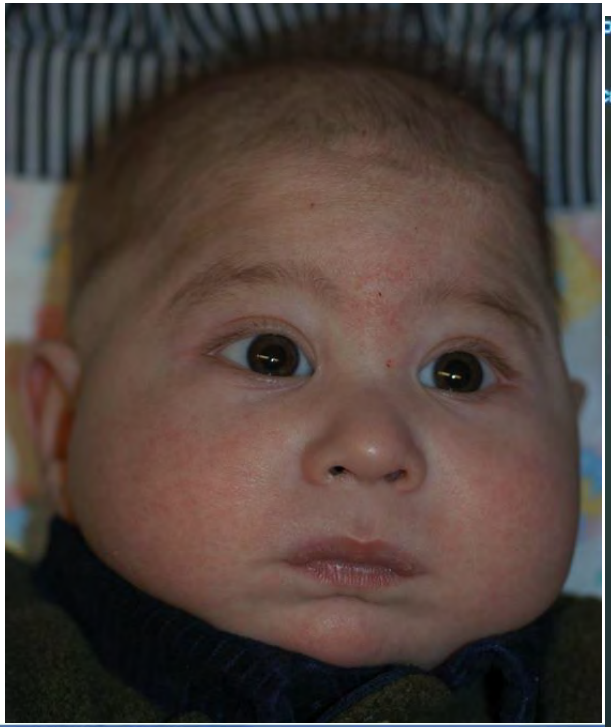
- Characteristics
  - Forehead keel/ridge
  - Hypotelorism
  - Lateral orbital recession
  - Bitemporal narrowing
  - Orbital configuration
  - Posterior cranial expansion
  - Omega sign on endocranial surface

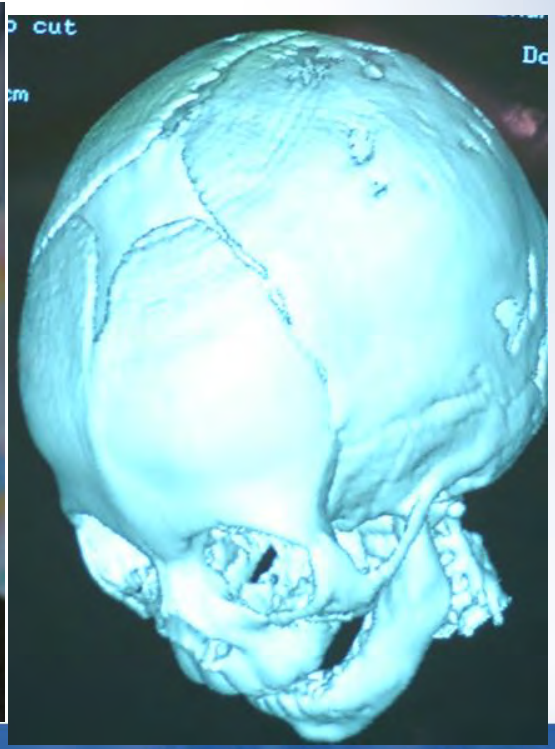




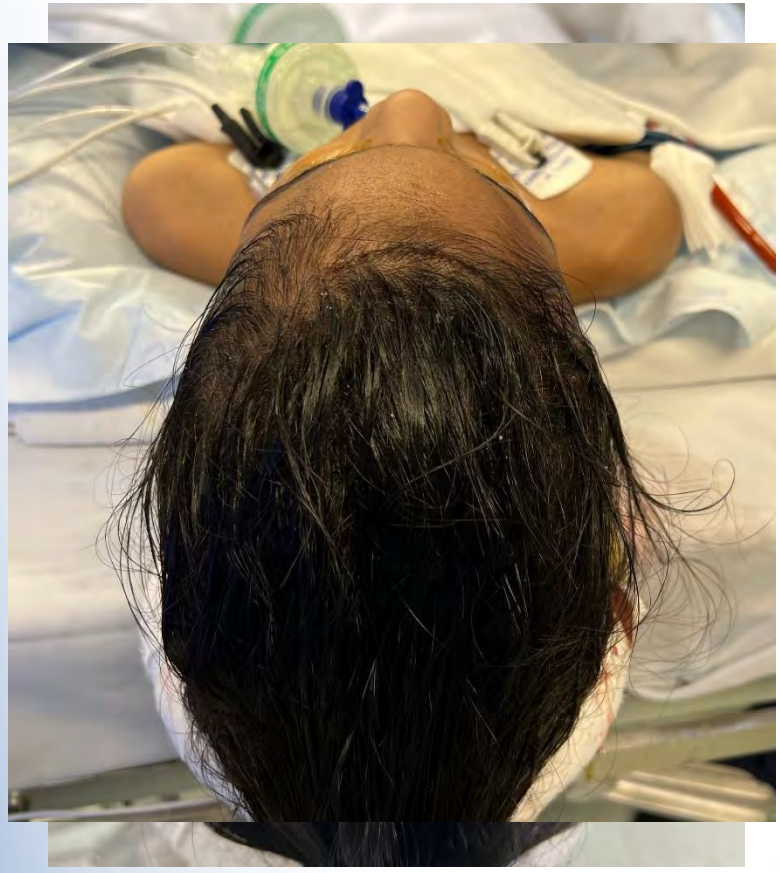
Metopic ridging: A form fruste of metopic synostosis?

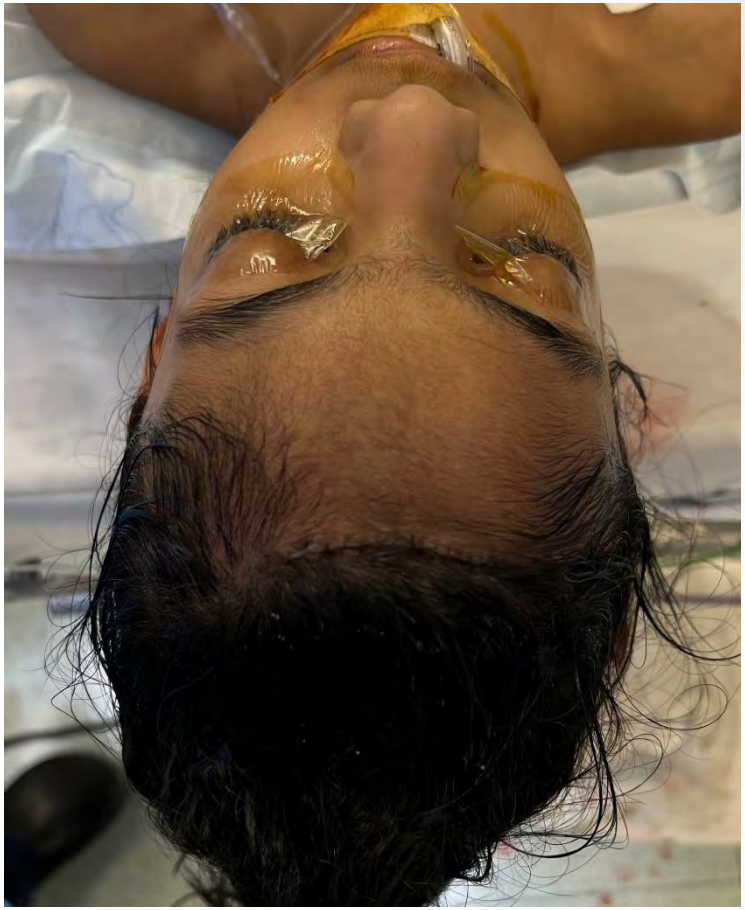




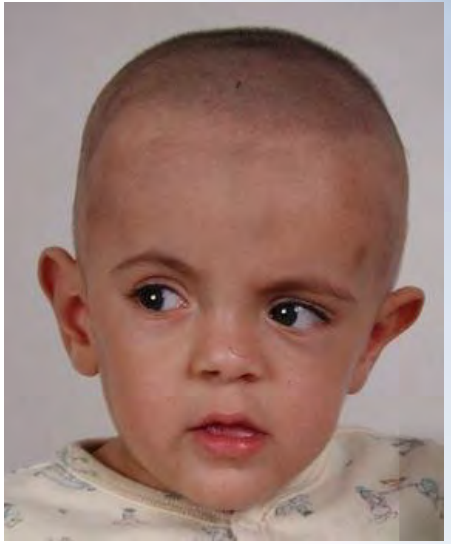


Metopic Ridge: My First Operative Case

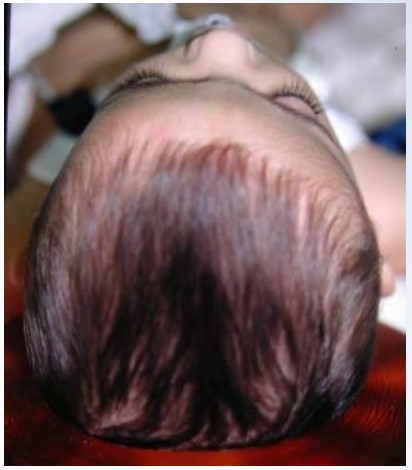


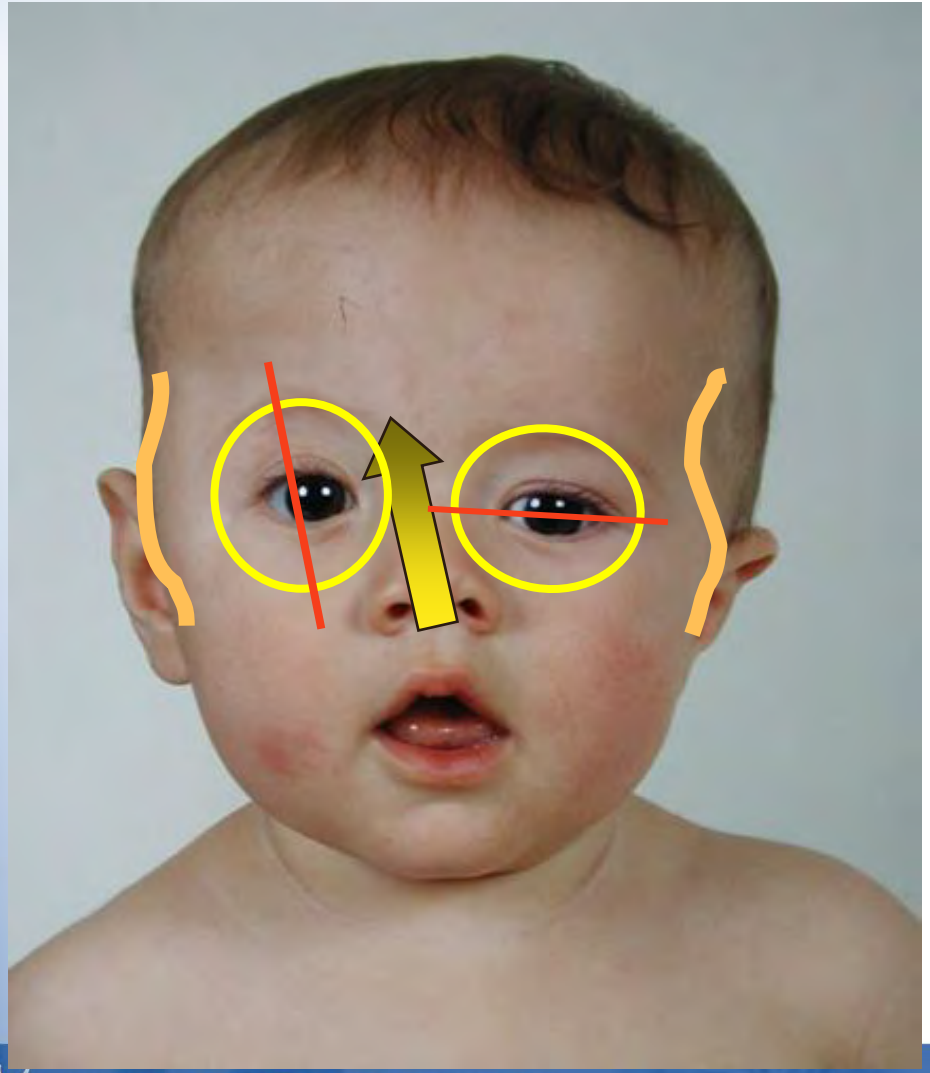


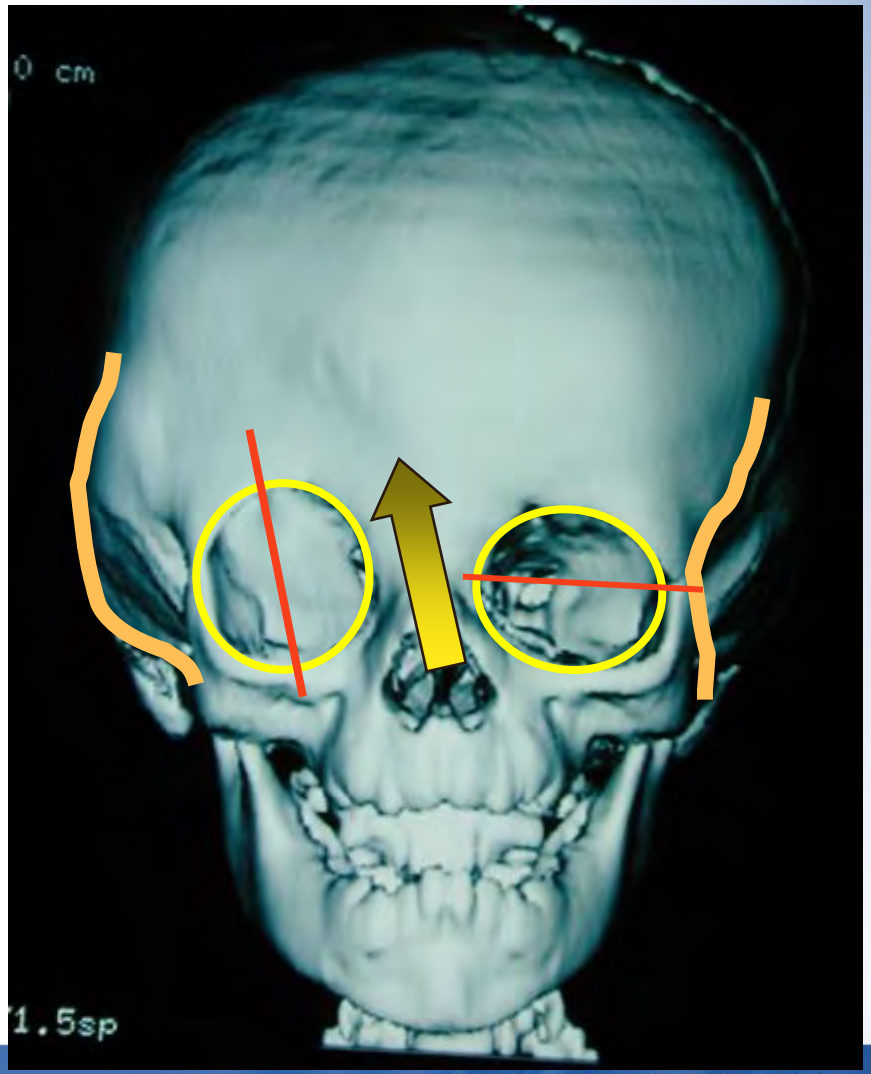
Unicoronal Synostosis

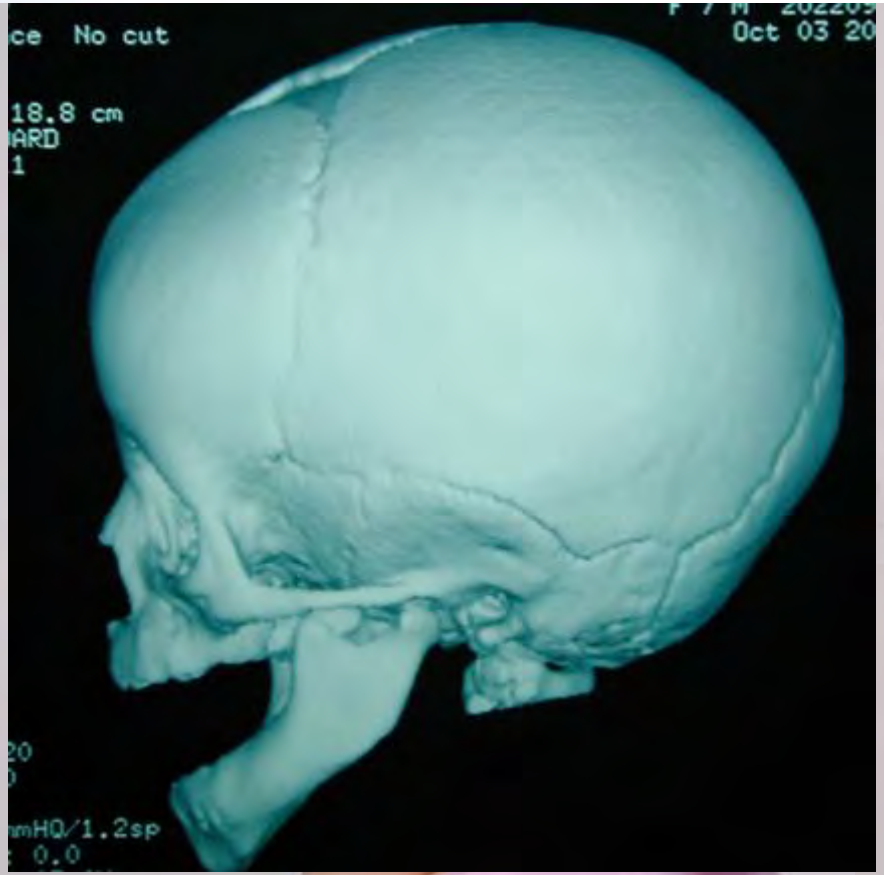


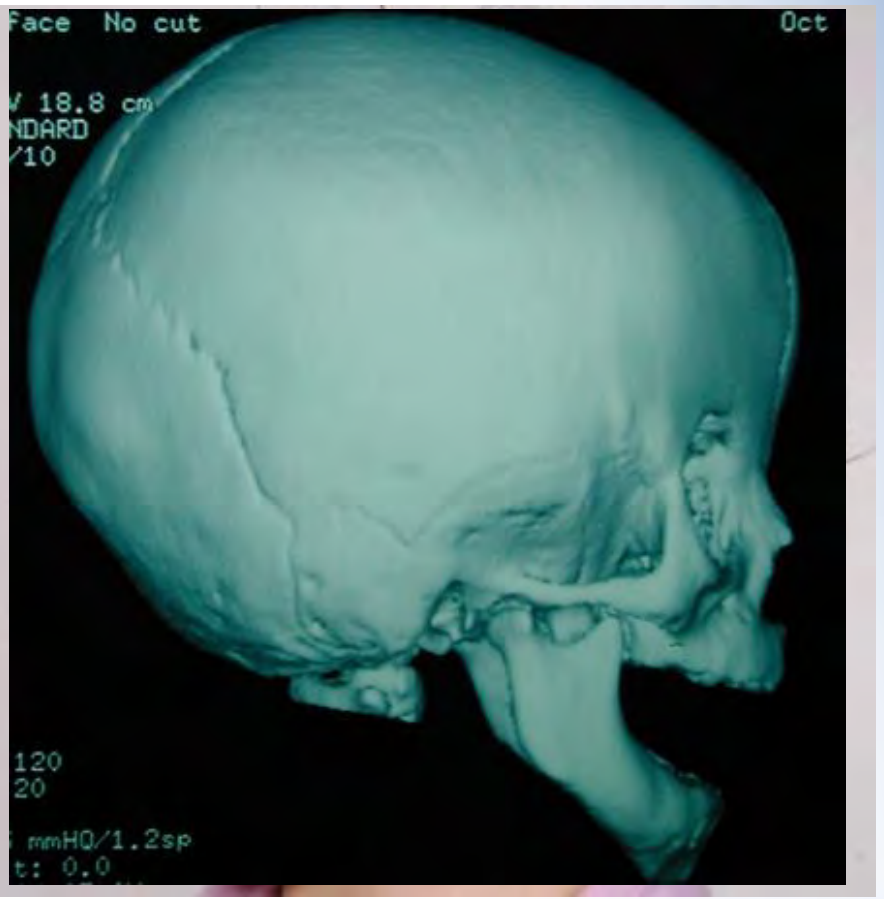
- Anterior plagiocephaly
- 1 in 10,000 births
- Female predominance
- Clinical features
  - No “normal side”
  - Ipsilateral forehead flattening
  - Ipsilateral temporal bulging
  - Nasal bridge deviation to synostosed side
  - Circular orbital configuration synostosed side
  - Transverse oval orbital configuration non-synostosed side
  - Contralateral forehead bossing
  - Ipsilateral lateral orbital wall posterior angulation
  - Ipsilateral “harlequin eye” on plain films
- Muenke (1997)
  - Pro250Arg substitution resulting from single point mutation in FGFR3 gene on chromosome 4p
  - Up to 20% of UCS patients



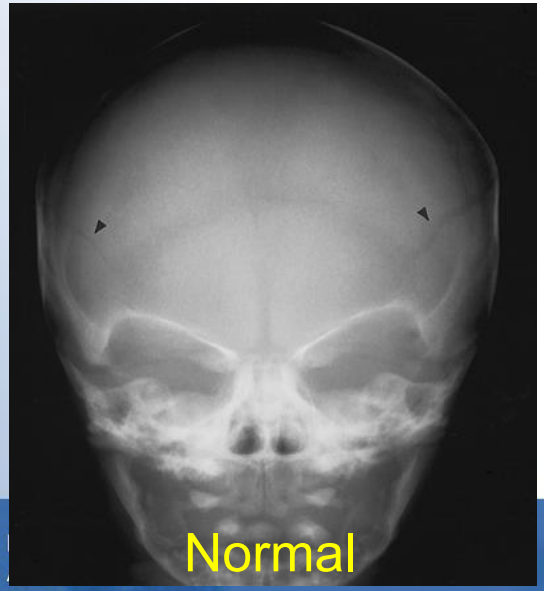


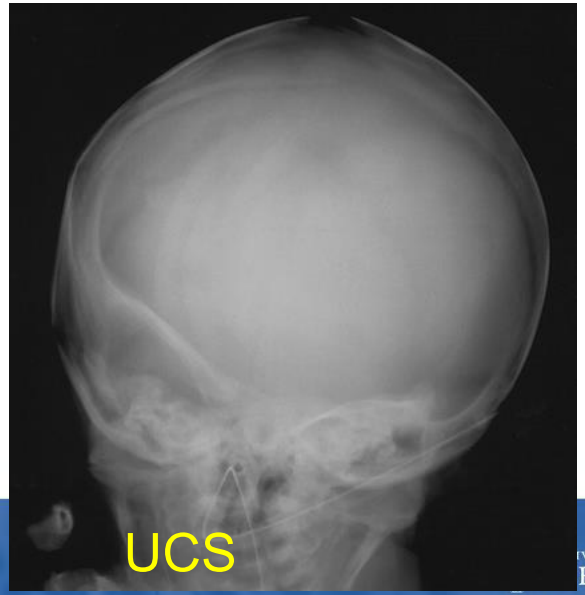

0 cm
1.5sp

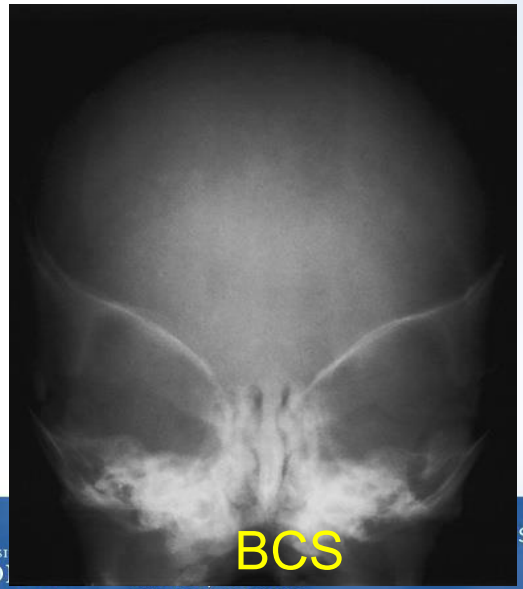

ce No cut
Oct 03 20
18.8 cm
ARD
1
20
mmHQ/1.2sp
0.0


Face No cut
Oct
18.8 cm
NDARD
/10
120
20
mmHQ/1.2sp
t: 0.0

Harlequin Eye Sign

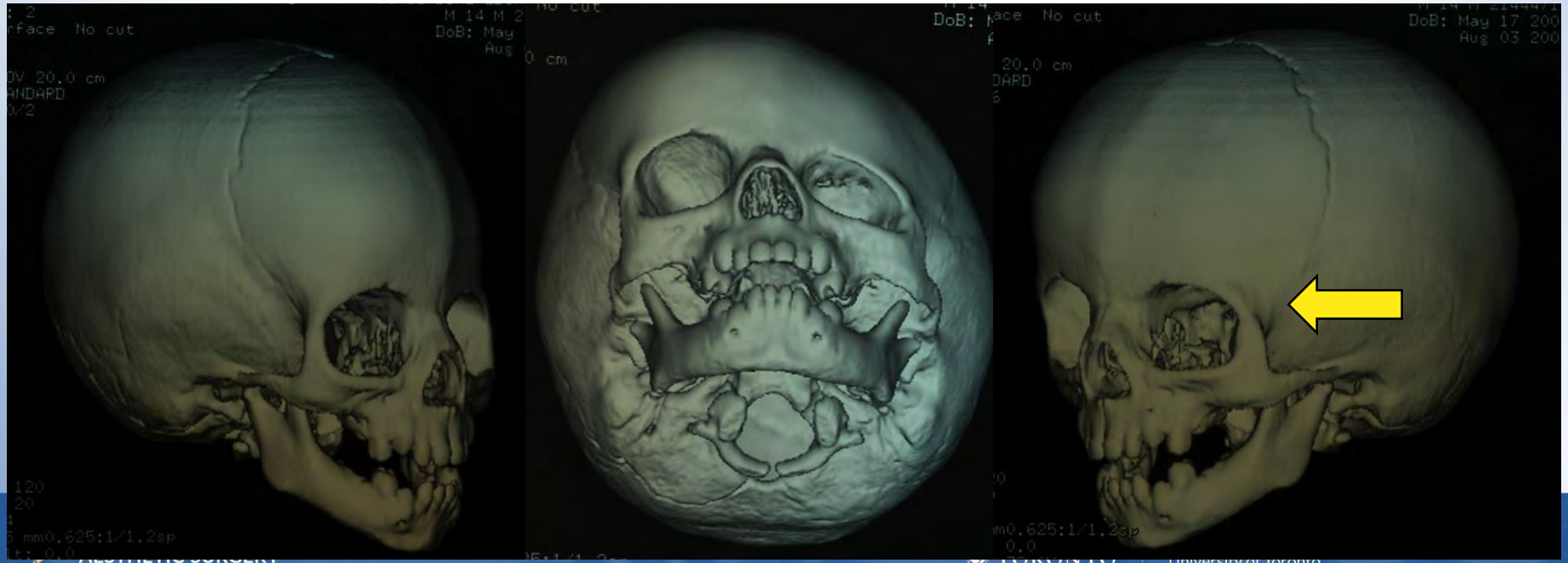






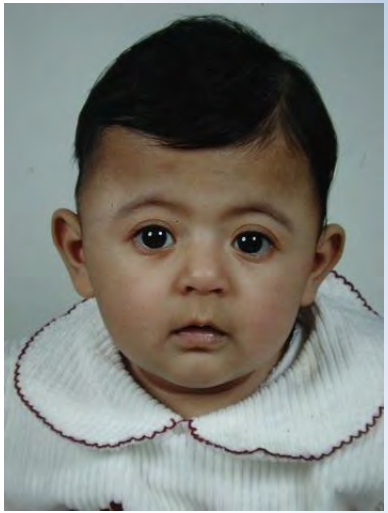
Anterior plagiocephaly variant

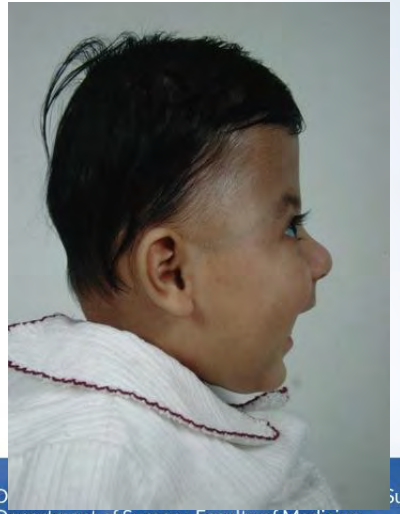
- Frontosphenoidal synostosis



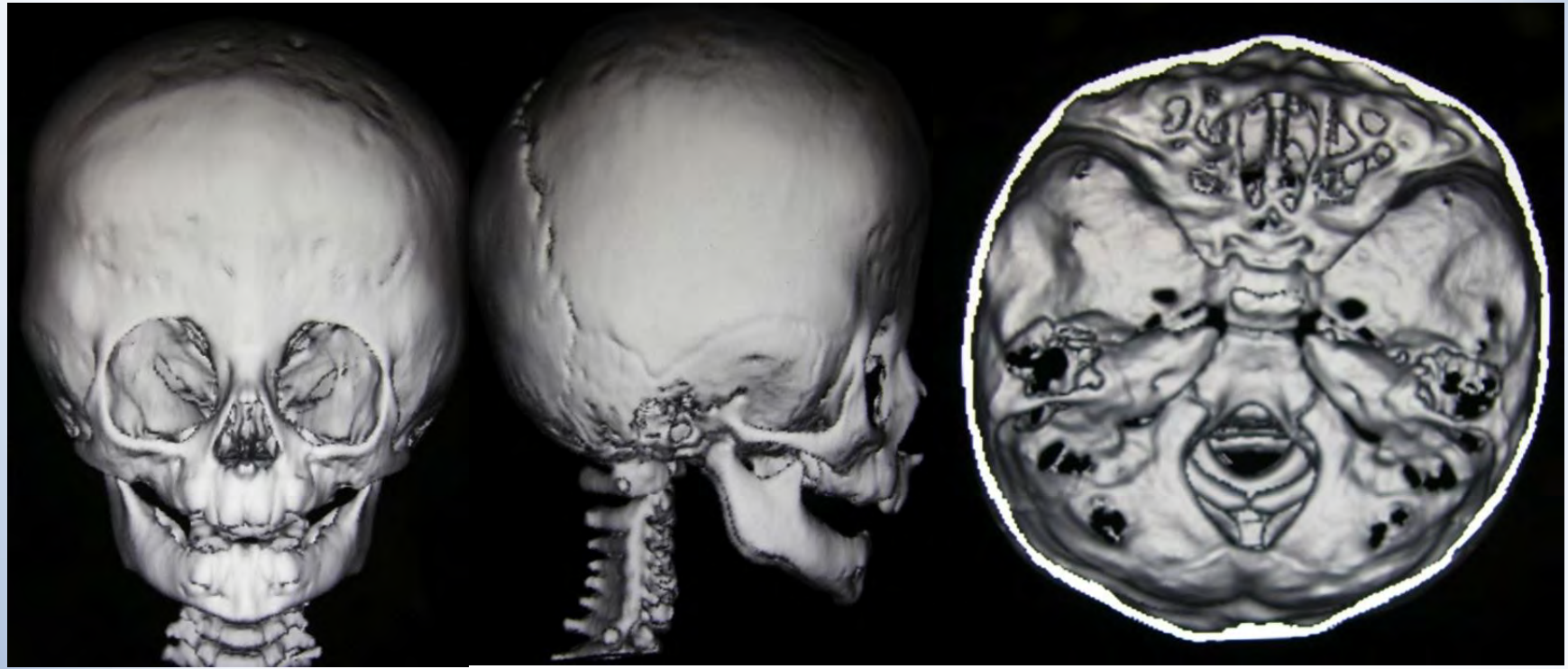
Bicoronal synostosis

- Turi-brachycephaly
- True non-syndromic uncommon – true frequency difficult to assess: 1 in 50,000?
- Distinction based on presence of midface retrusion
- Coronal ring: paired coronal and frontosphenoid sutures and the cartilaginous sphenoethmoidal synchondrosis
- Clinical features
  - Turri (tower) – brachy (short) skull
  - Shortened AP dimension
  - Flat forehead
  - Transverse widening
  - Increased vertical height
  - Superior orbital rim recession (cornea projects anterior to bone)
  - Bilateral temporal bulging



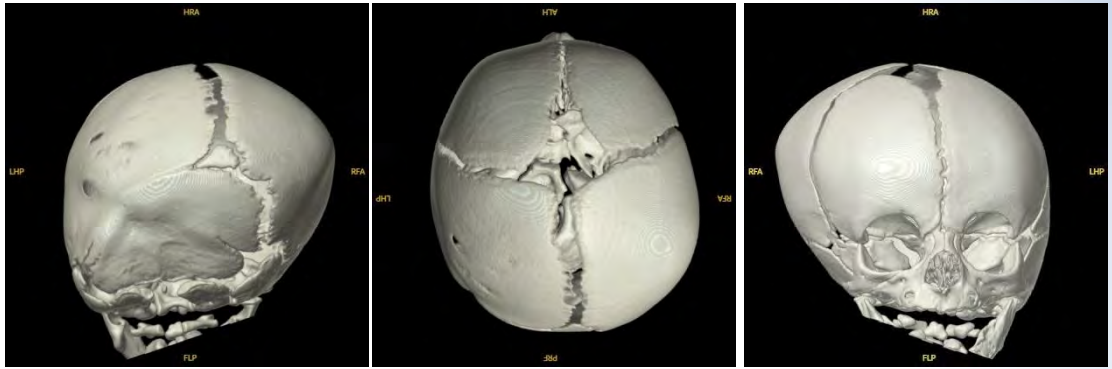


Bicoronal synostosis

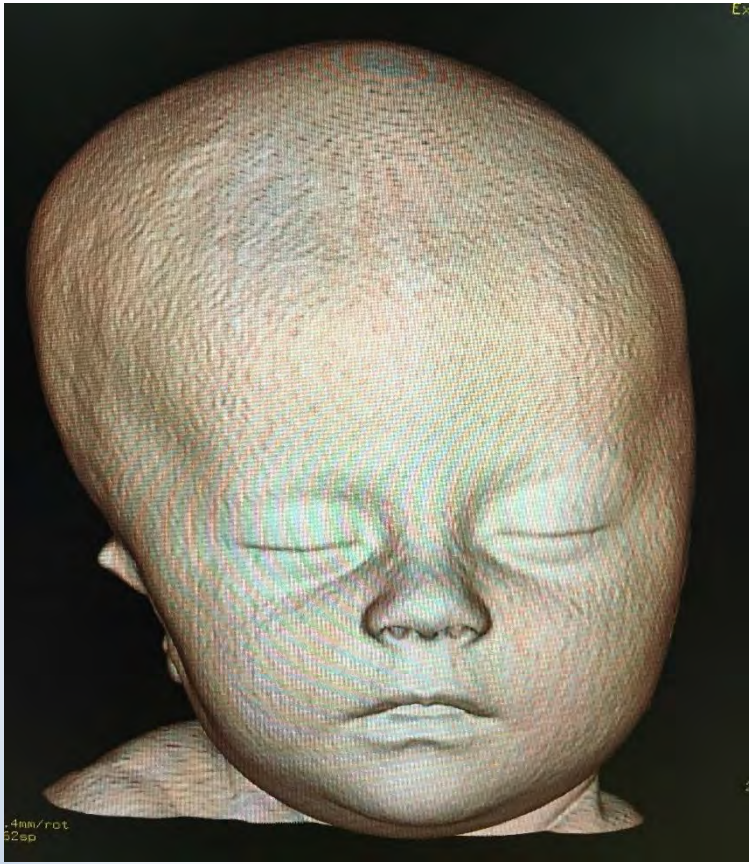


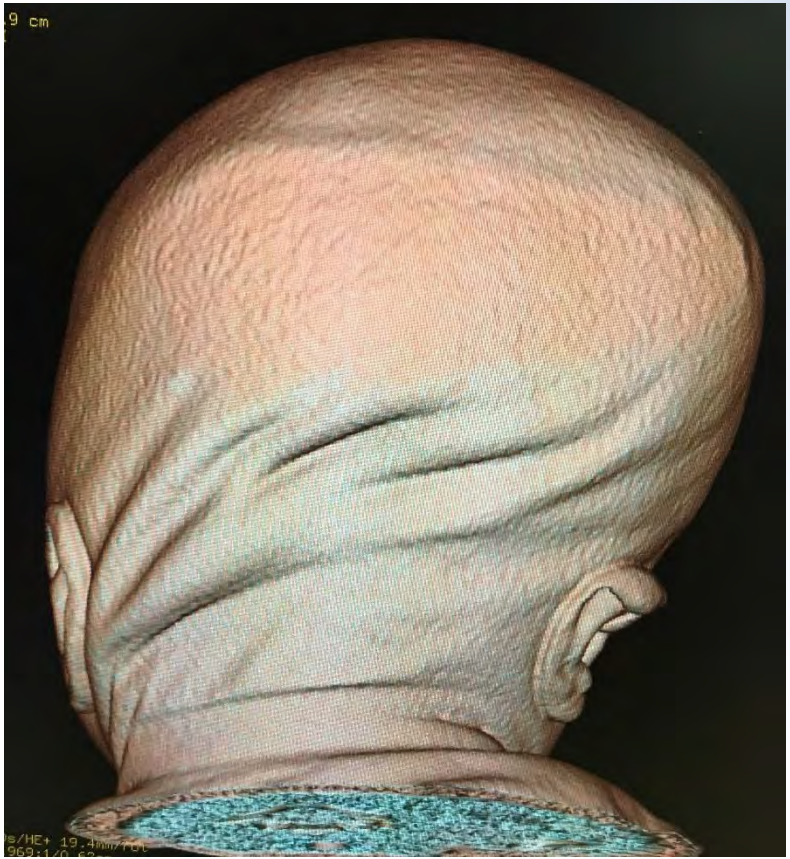
Lambdoid synostosis

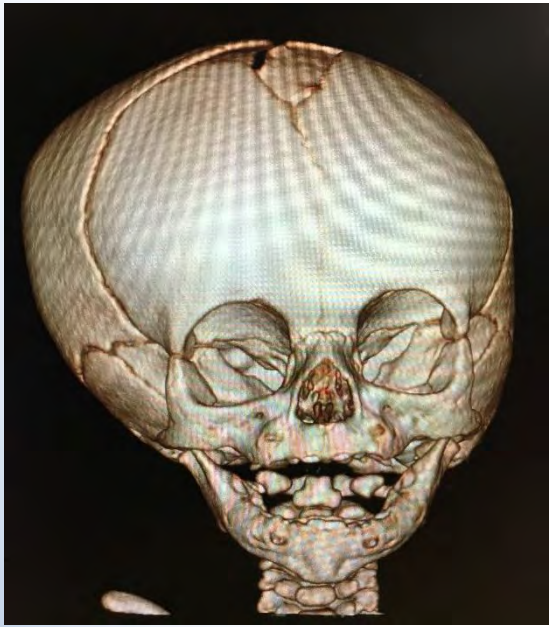
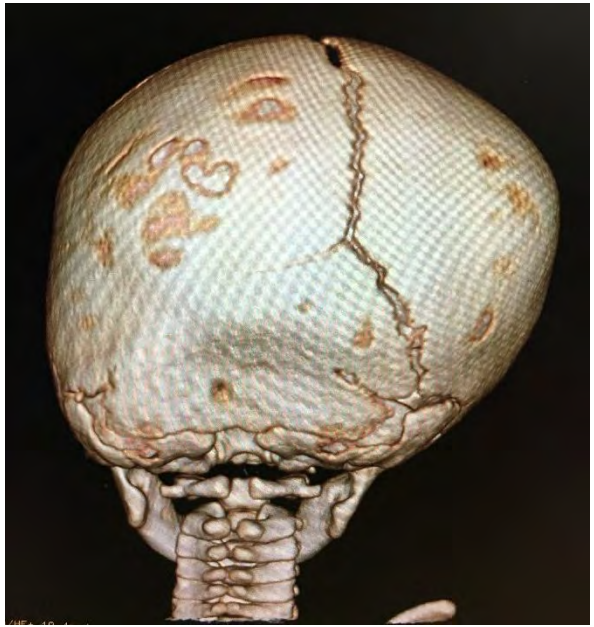
- Rare
- Posterior plagiocephaly
- 1 in 60,000 births
- Clinical features
  - Ipsilateral occipital flattening
  - Ridging along suture
  - Ear position not reliable**
  - Ipsilateral mastoid bulge
  - Canted cranial base
  - Trapezoidal head



J Craniofac Surg
. 2021 Jan-Feb;32(1):125-129.
Distinguishing Craniomorphometric Characteristics of Unilateral Lambdoid Craniosynostosis
Omar Allam[1], Kitae E Park, Navid Pourtaheri, Mohammad Ali Mozaffari, John Smetona, Xiaona Lu, Maham Ahmad, John A Persing, Michael Alperovich




9 cm

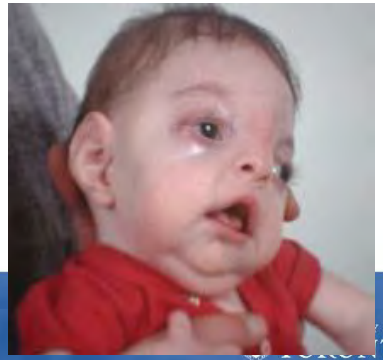



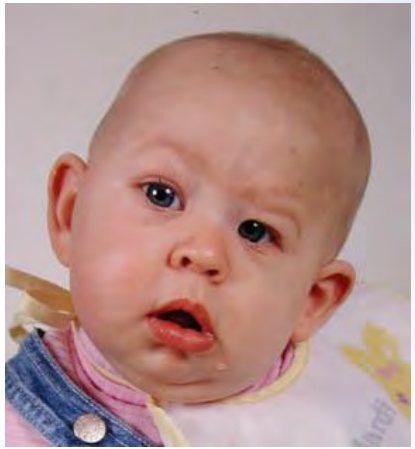

Lambdoid Synostosis

- Gumby

Deformational Plagiocephaly

- Epidemiology (prior to 1992)
  - Positional plagiocephaly : 1 in 300 infants
  - Risk factors
    - Prematurity
    - Constrictive intra-uterine environment
    - Multiple births
    - Torticollis
    - Birth trauma
    - Lack of full bone mineralization
    - Neurological deficits





PEDIATRICS®
OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

- Sleep Position and SIDS
  - AAP Task Force on Infant Positioning and SIDS
  - *Pediatrics:89, 1120, 1992*
  - Odds ratio: SIDS 1.3 to 11.7 times more likely with prone positioning

"healthy infants, when being put down to sleep, be positioned on their side or back"

1994: Back to Sleep Campaign launched!

Deformational Plagiocephaly

- The ripple effect of the Back to Sleep campaign.


The "Back to Sleep Campaign" and Deformational Plagiocephaly: Is There Cause for Concern?

1996


Observations on a Recent Increase in Plagiocephaly Without Synostosis

1996


The "Back to Sleep Campaign" and Deformational Plagiocephaly: Is There Cause for Concern?

1996

Deformational Plagiocephaly

- 2002:
  - 300% increase in referrals past 3 years
  - 300 to 400 new cases per year at HSC (estimate)
    - Plastic Surgery
    - Neurosurgery
    - Physiotherapy

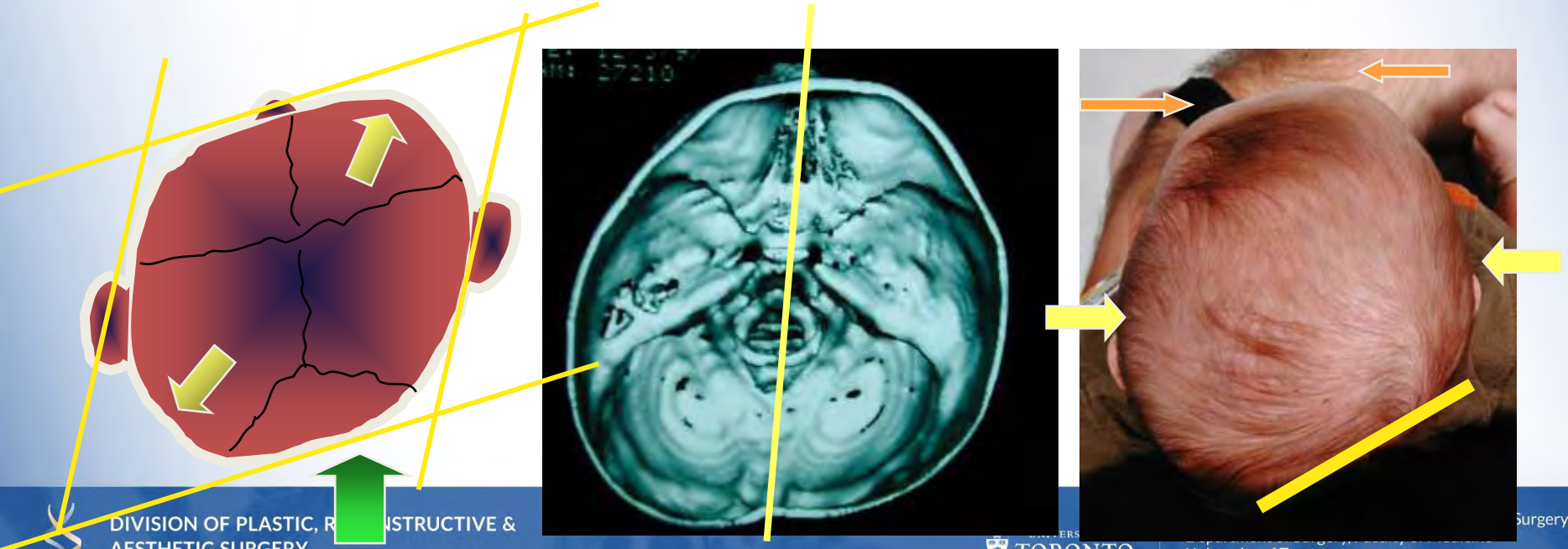
  - Incidence: 1 in 60 to 1 in 2 infants
  - Birth stats:
    - Ontario: 1999: 130,000 births
    - Potentially 2,500 to 65,000 new patients per year!

Deformational Plagiocephaly

- Why does it happen?
  - Soft skull
  - Hard mattress
  - Car seats

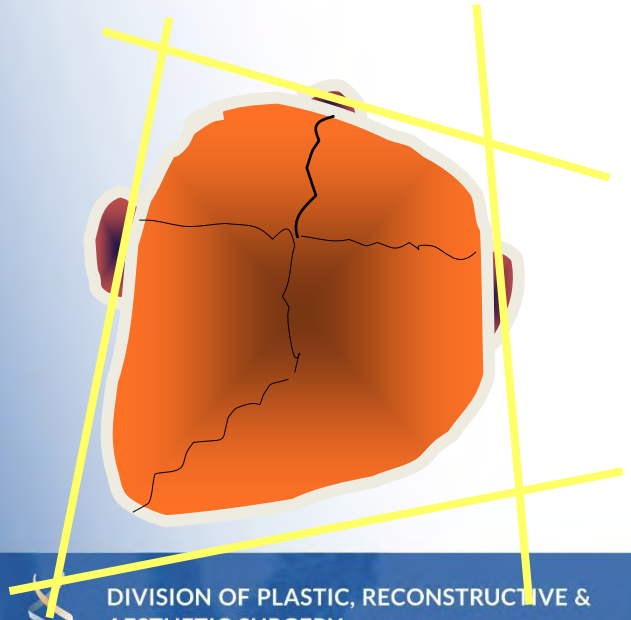
Deformational Plagiocephaly

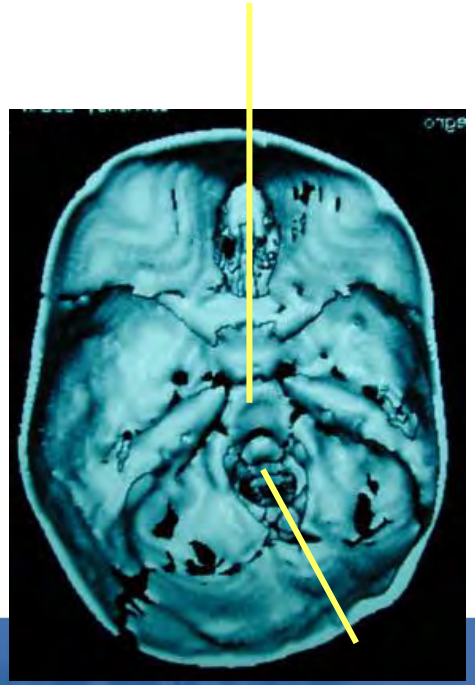
- Pathophysiology: Positional Plagiocephaly

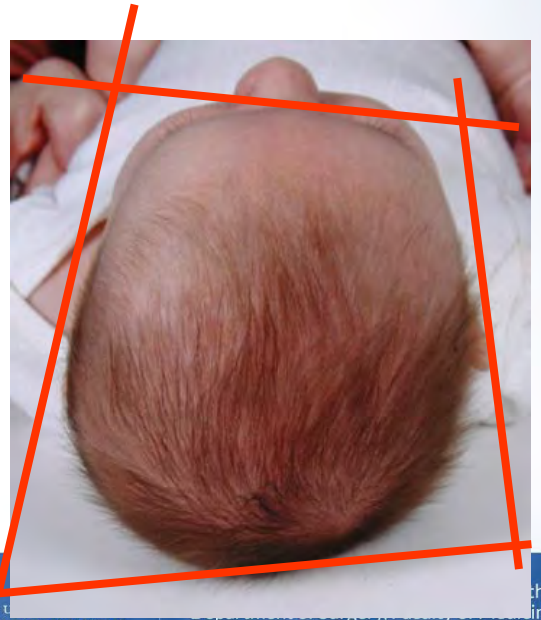


Deformational Plagiocephaly

- Differential Diagnosis:Lambdoid synostosis

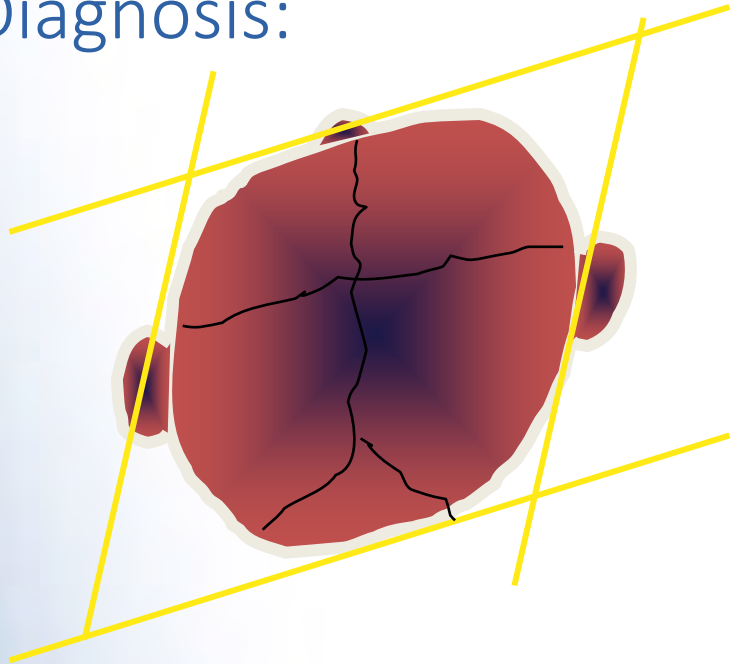




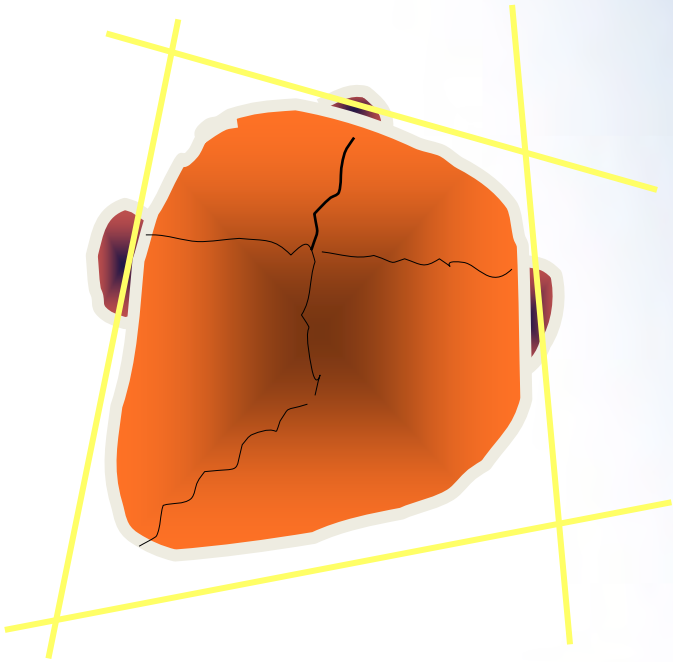


Deformational Plagiocephaly

- Diagnosis:



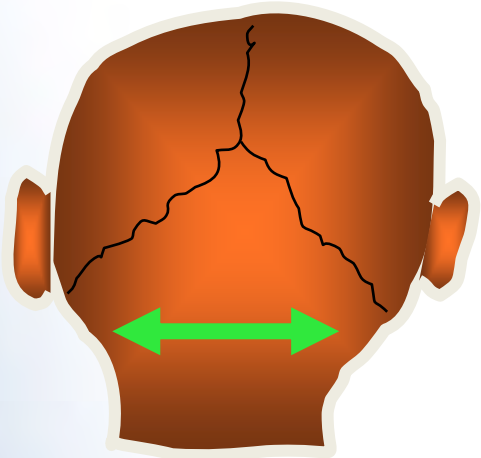
Positional: Parallelogram



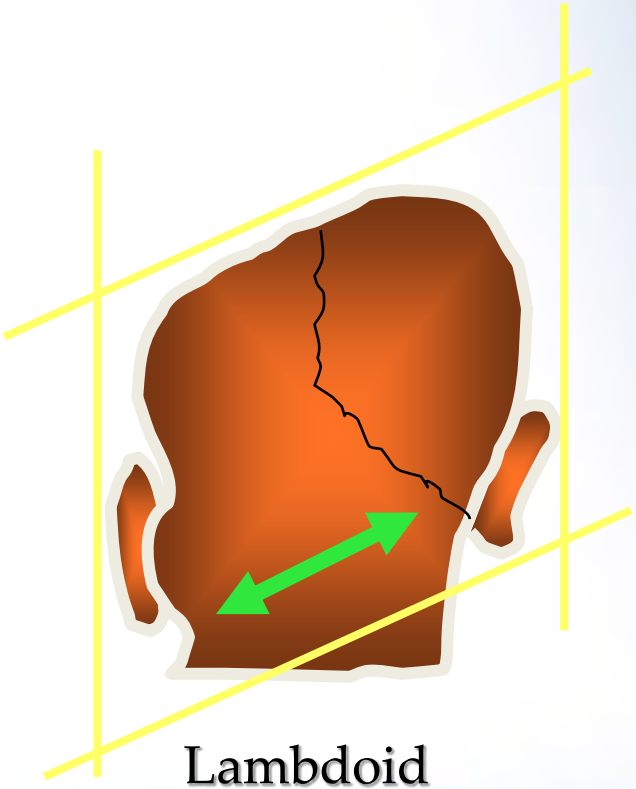
Lambdoid: Trapezium

Deformational Plagiocephaly

- Diagnosis:



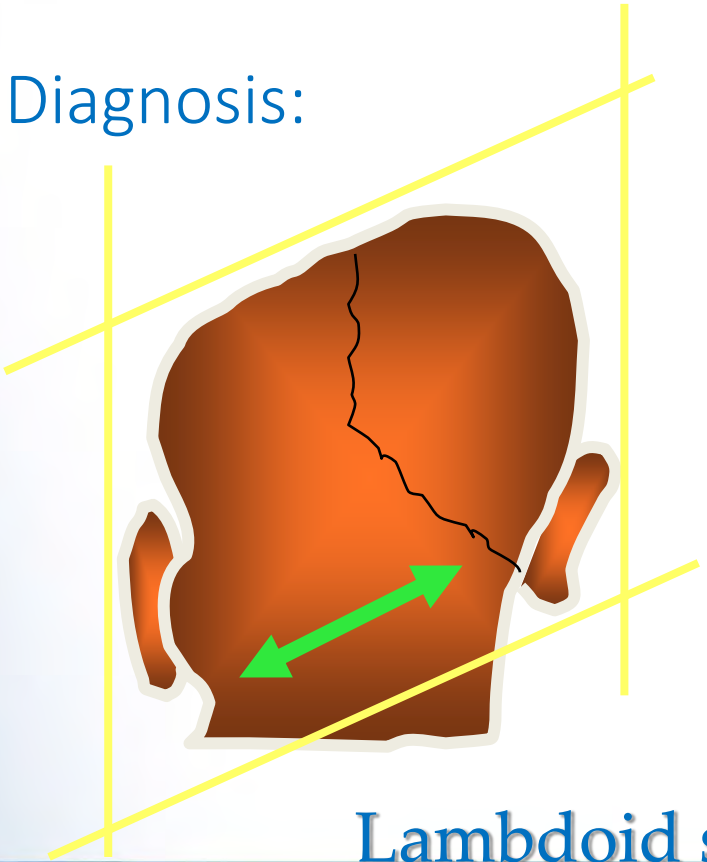
Positional

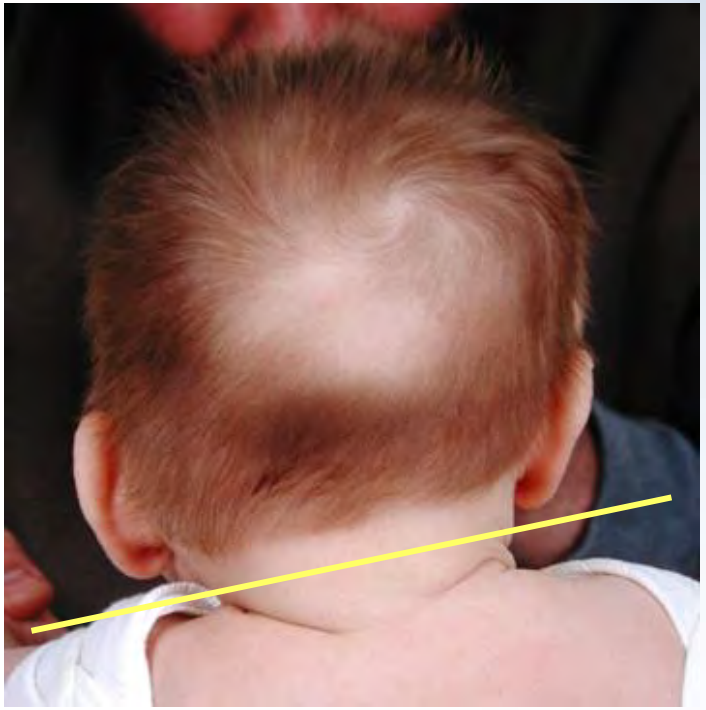


Lambdoid

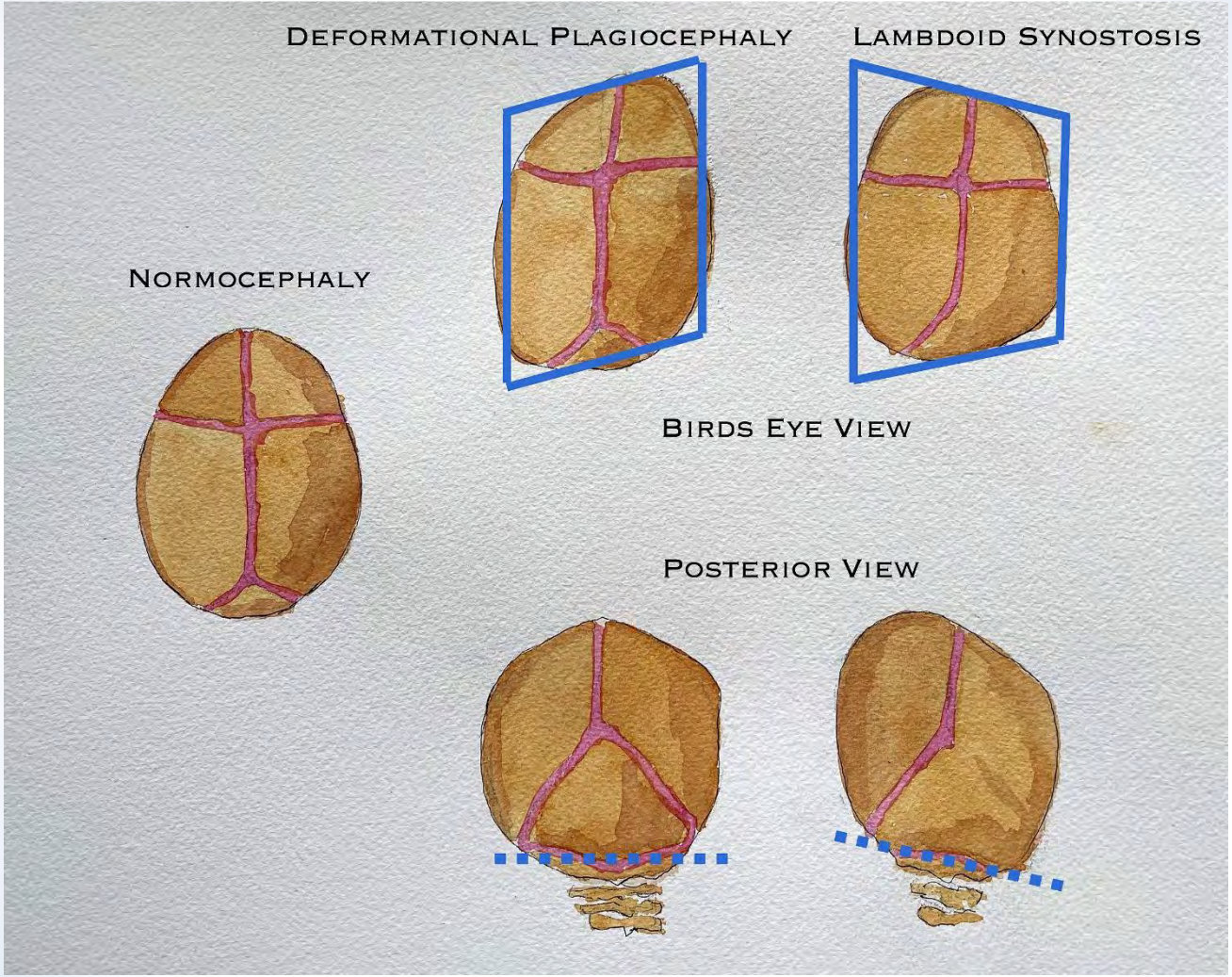
Deformational Plagiocephaly

- Diagnosis:





Lambdoid synostosis

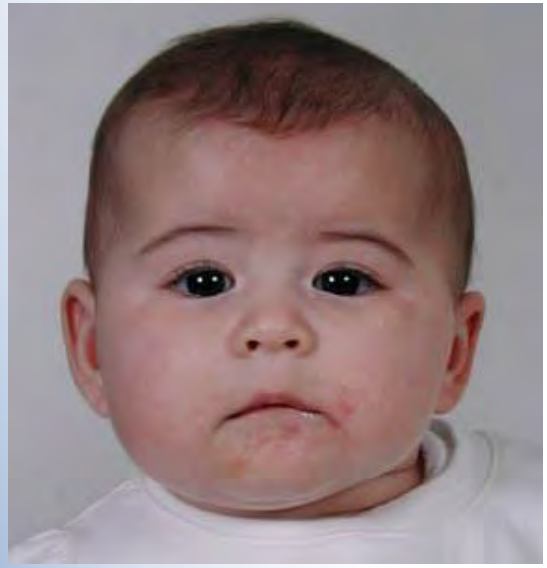

Deformational Plagiocephaly
Lambdoid Synostosis
Normocephaly
Birds Eye View
Posterior View

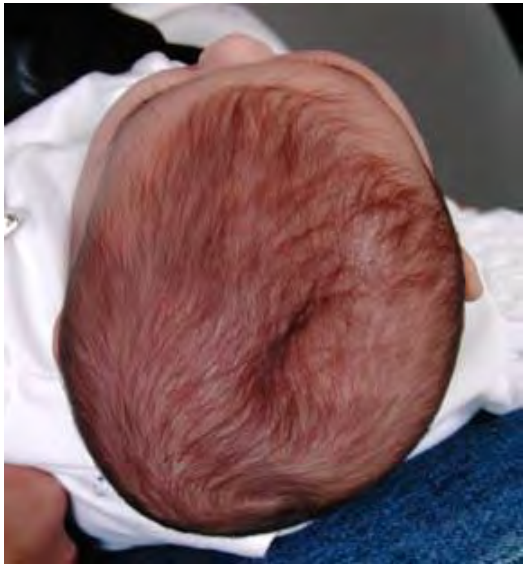
Deformational Plagiocephaly

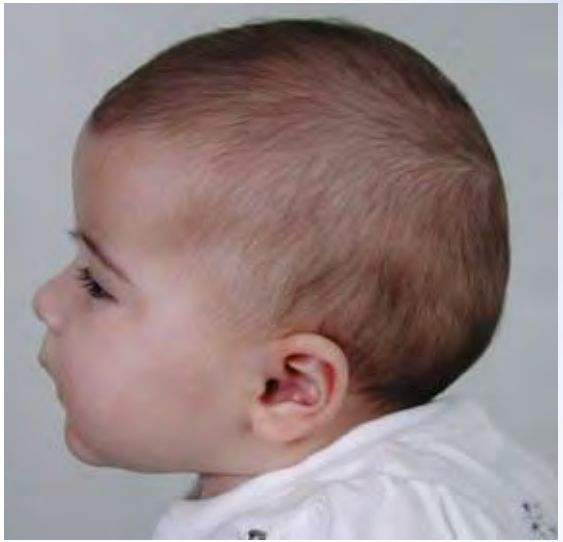
- How do you make the diagnosis?
  - 1. Look for the flat spot: right versus left versus midline
  - 2. Look at the position of the ipsilateral ear compared to the opposite side
  - 3. Check position of the forehead (it should follow the ear in positional)
  - 4. Check neck ROM
  - 5. Remember that unilateral lambdoid synostosis affects 1 in 60,000 live children (it is pretty rare!) (bilateral is even rarer!)
  - 6. If it doesn't make sense, then refer!

Deformational Plagiocephaly

- Clinical Subtypes:
  - Right Forward - 85%

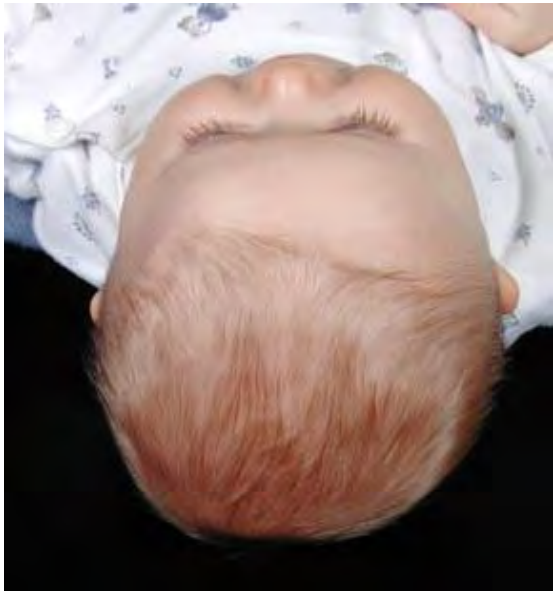
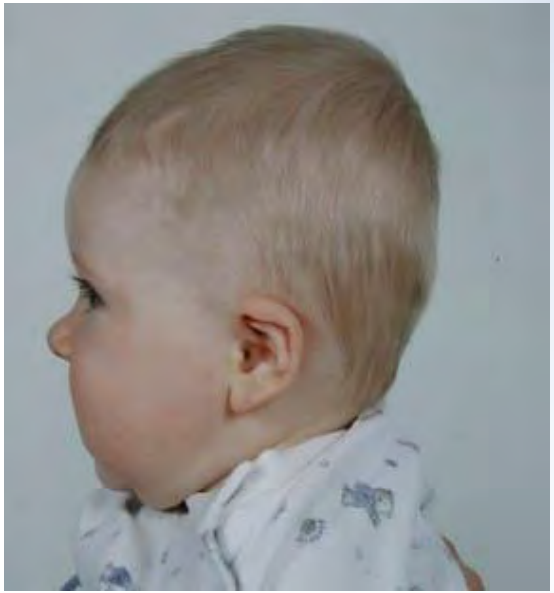






Deformational Plagiocephaly

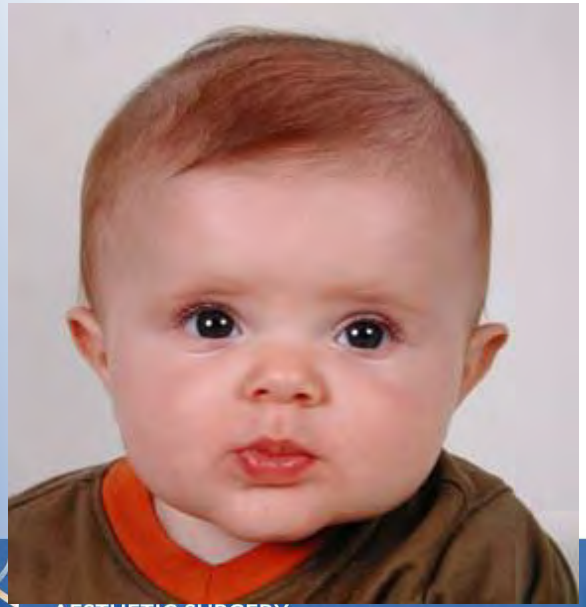
- Clinical Subtypes:
  - Flat - 10%*

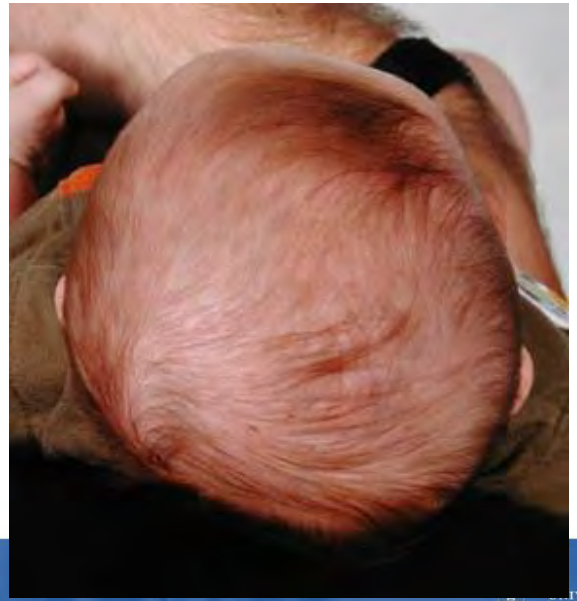




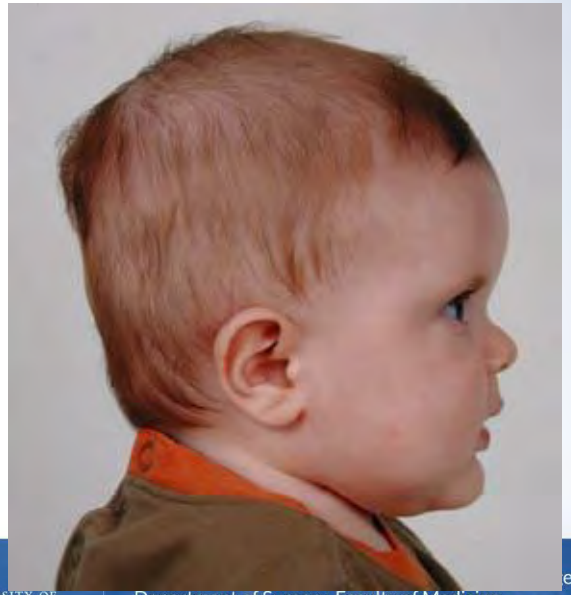
* Check for neurodevelopmental delay

Deformational Plagiocephaly

- Clinical Subtypes:
  - Left Forward - 5%

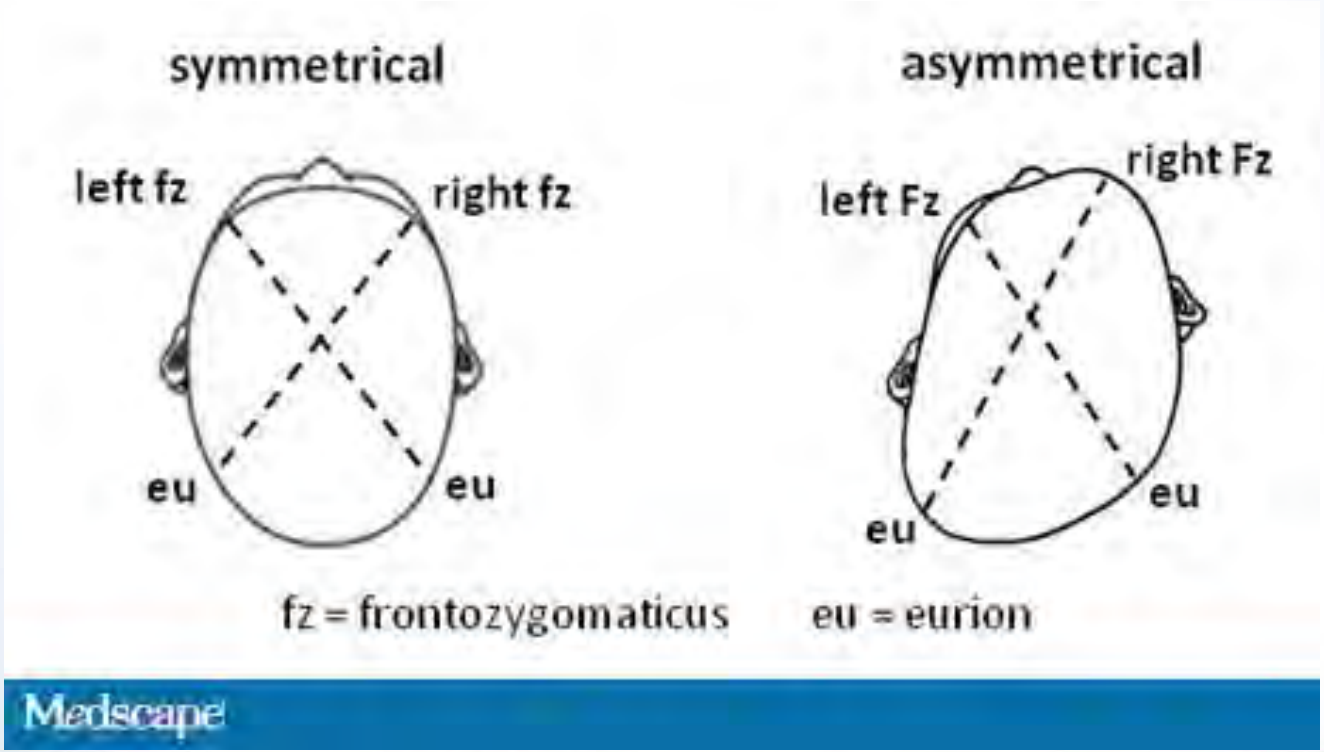






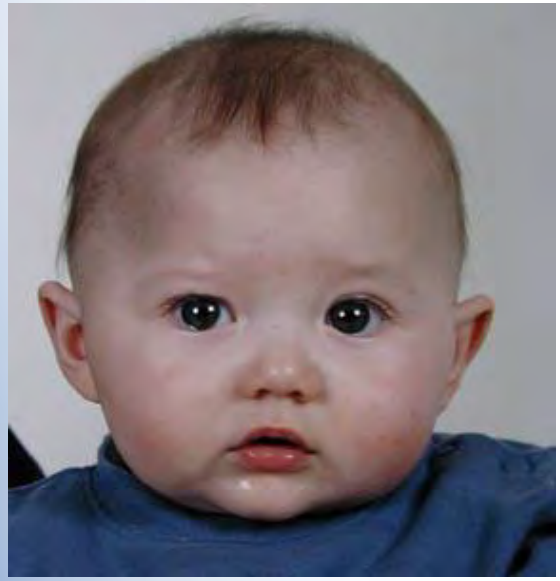
Deformational Plagiocephaly

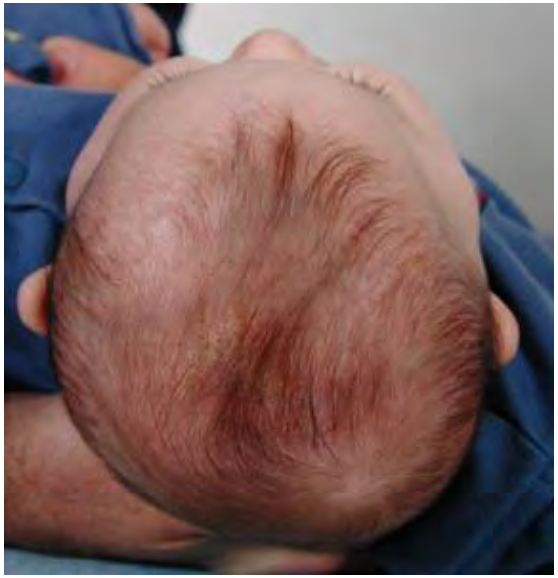
- Grade:

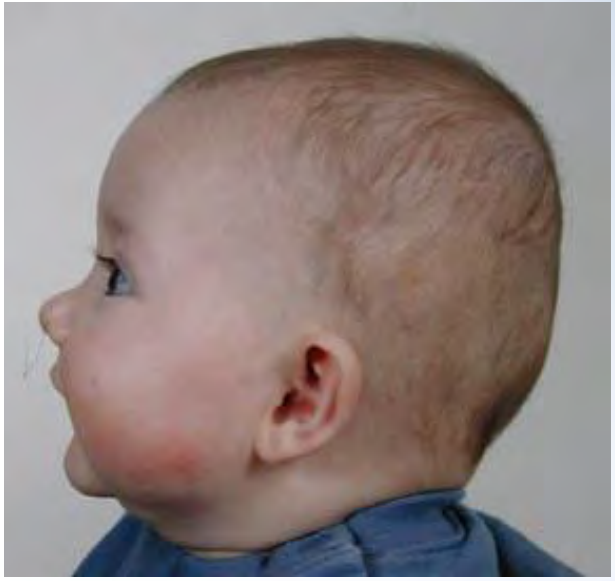


Deformational Plagiocephaly

- Grade:
  - Mild

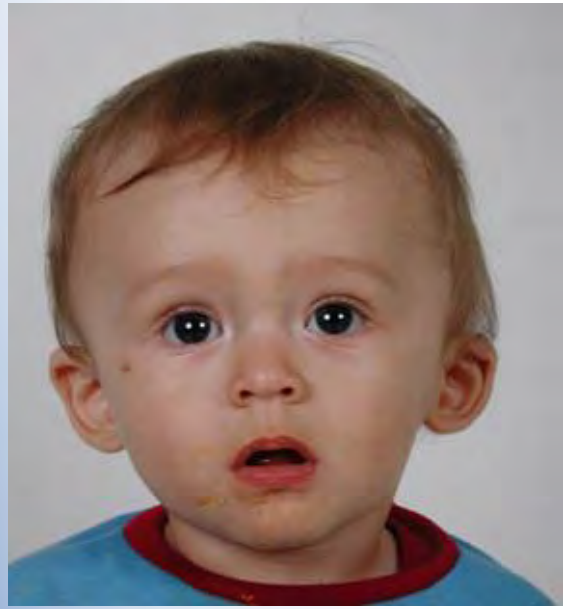


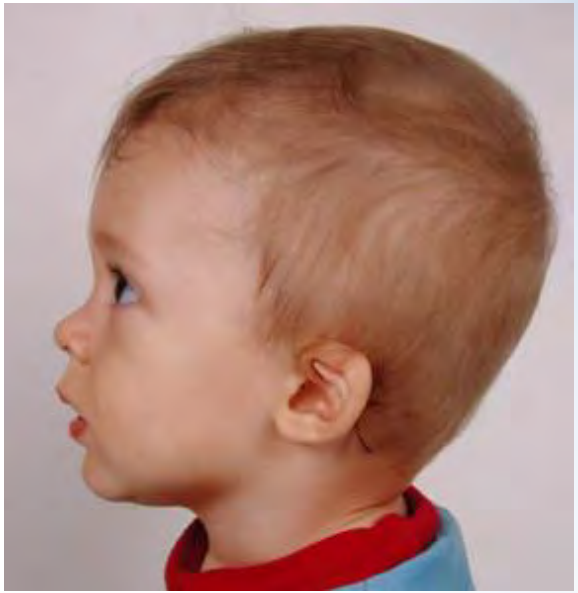




Deformational Plagiocephaly

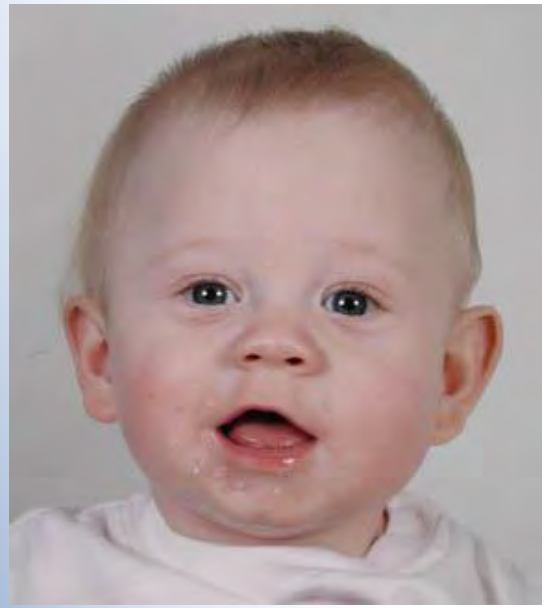
- Grade:
  - Moderate

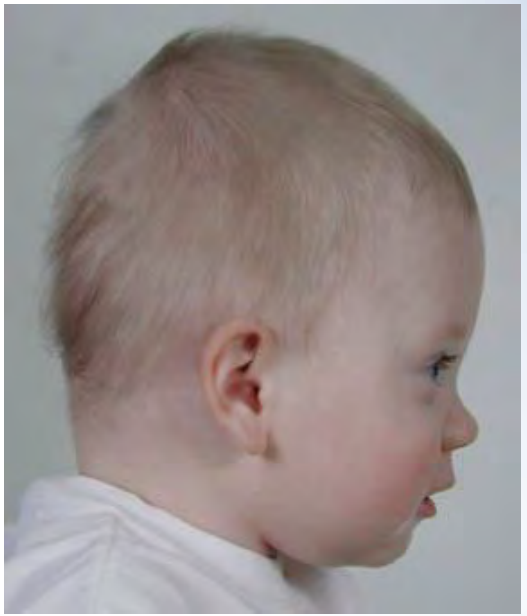




Deformational Plagiocephaly

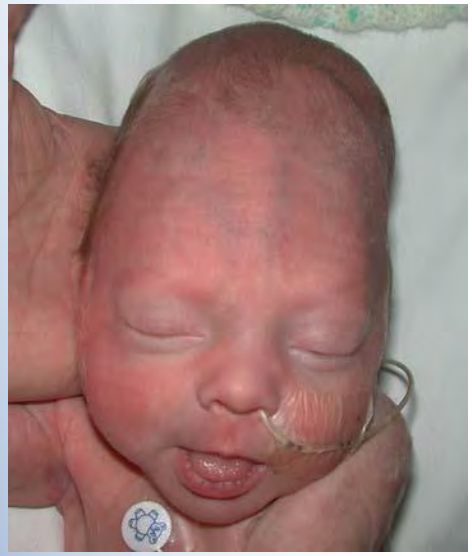
- Grade:
  - Severe

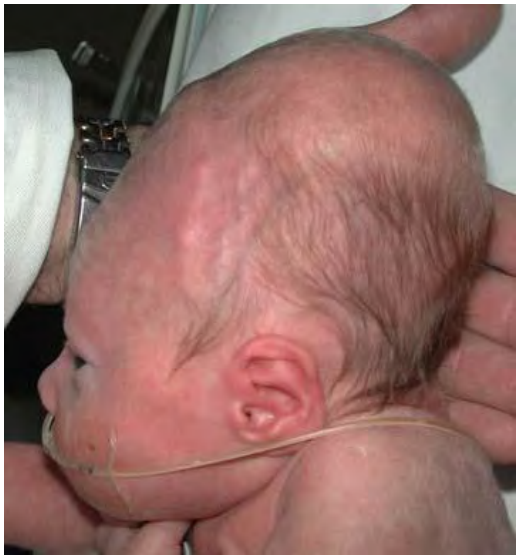


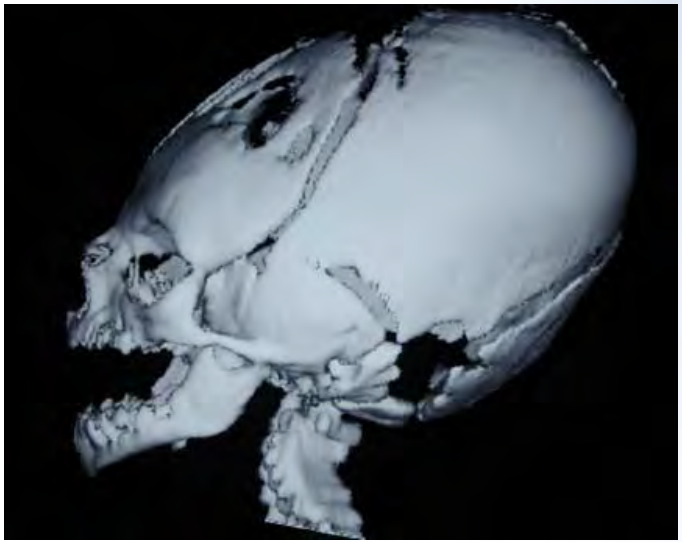


Deformational Plagiocephaly

- Grade:
  - Really, really bad

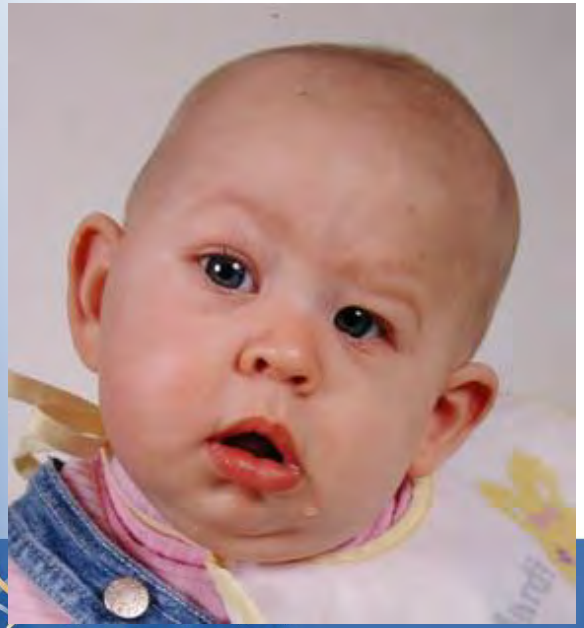


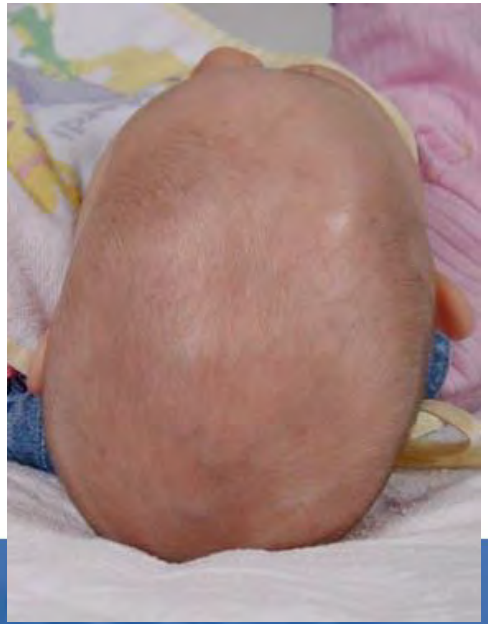




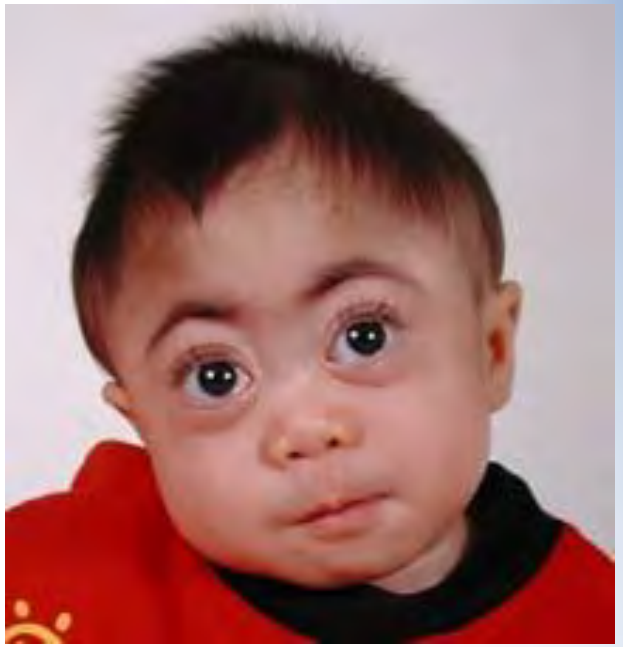
Deformational Plagiocephaly

- Torticollis:
  - Cranial and facial asymmetries

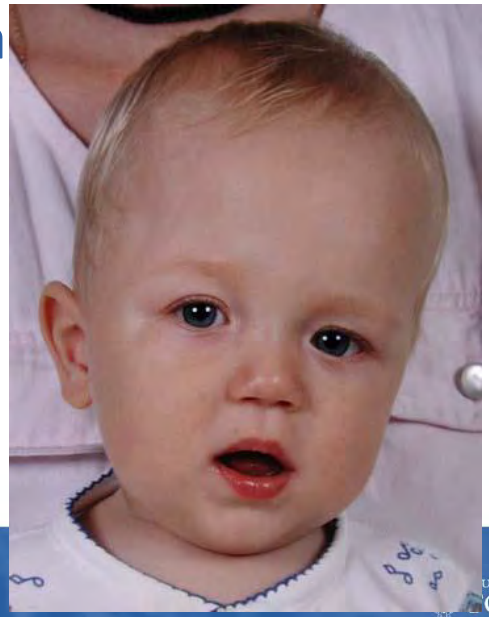


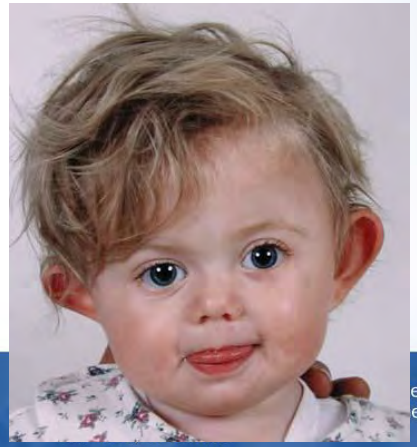


Deformational Plagiocephaly



- But, don't forget……
  - Craniosynostosis
  - Cephalohematoma
  - Bone tumour
  - C-spine anomalies
  - Head tilt
    - Ocular
    - Neurologic

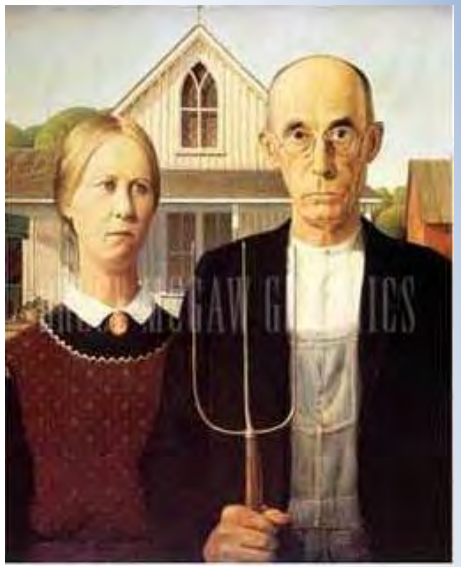




Deformational Plagiocephaly

- Management:
  - Interdisciplinary assessment:
    - Plastic Surgery
    - Physiotherapy
    - Occupational Therapy
  - Investigation:
    - Plain x-rays (occasionally)
      - C-spine
    - CT scan (only if pattern doesn't fit)
  - Treatment:
    - Active Counter-Positioning: < 6 months
    - Molding Helmet: 4 months to 12 months
    - Surgery: almost never

Deformational Plagiocephaly



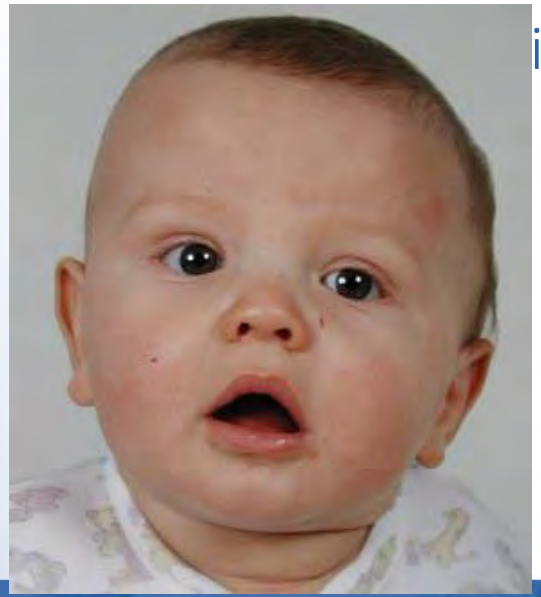
- Idiosyncracies of parents with affected children:
  - 1. First child
  - 2. Economic stability
  - 3. Undue concern regarding cosmetic deformity
  - 4. High maintenance
  - 5. Disproportionate amount of consult time required
  - 6. Disagreement between parents regarding best course of action
  - 7. Mother-son and Father-daughter preference for treatment

Deformational Plagiocephaly

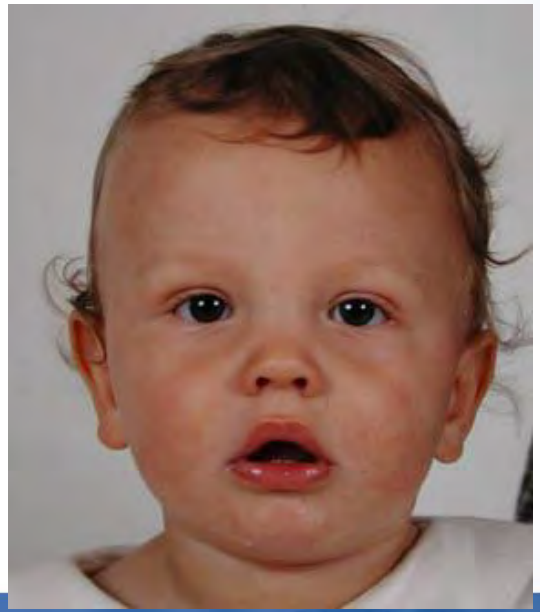
- Frequently asked questions:
  - 1. What is the natural history?
  - 2. Are there developmental concerns?
  - 3. How effective are the molding helmets?

Deformational Plagiocephaly

- Natural History of Positional Plagiocephaly:
  - ioning:

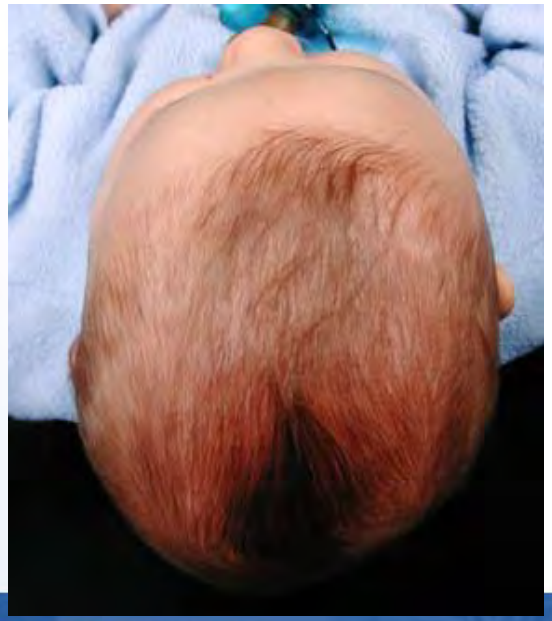


5 months

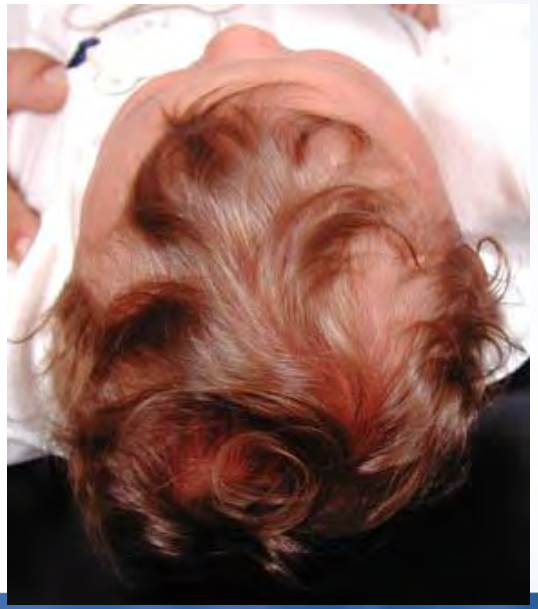


Deformational Plagiocephaly

- Natural History of Positional Plagiocephaly:
  - Active Counter Positioning:

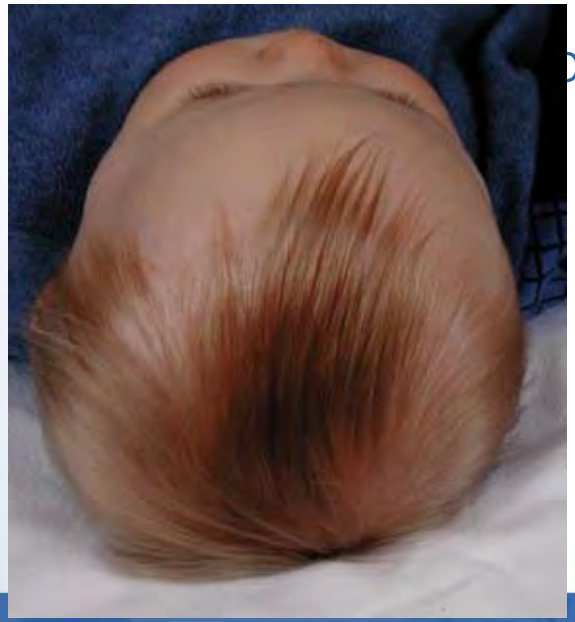


5 months

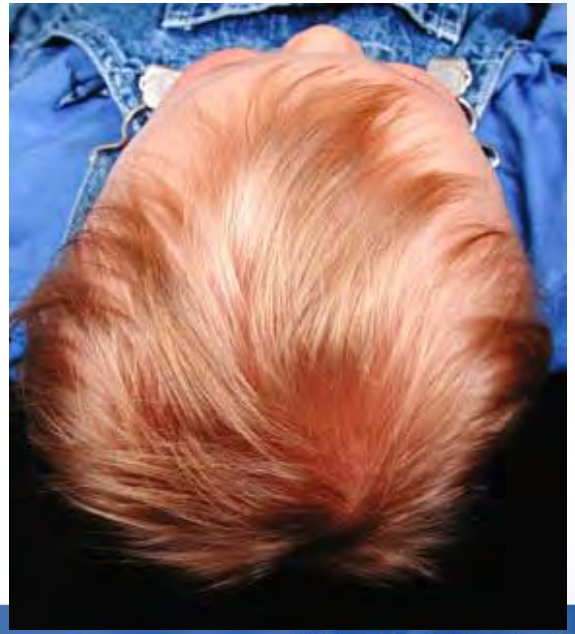


Deformational Plagiocephaly

- Natural History of Positional Plagiocephaly:
  - oning:



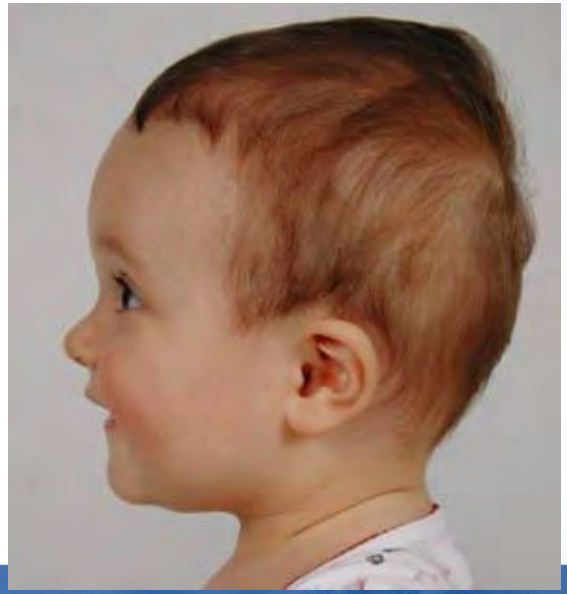
6 months



Deformational Plagiocephaly

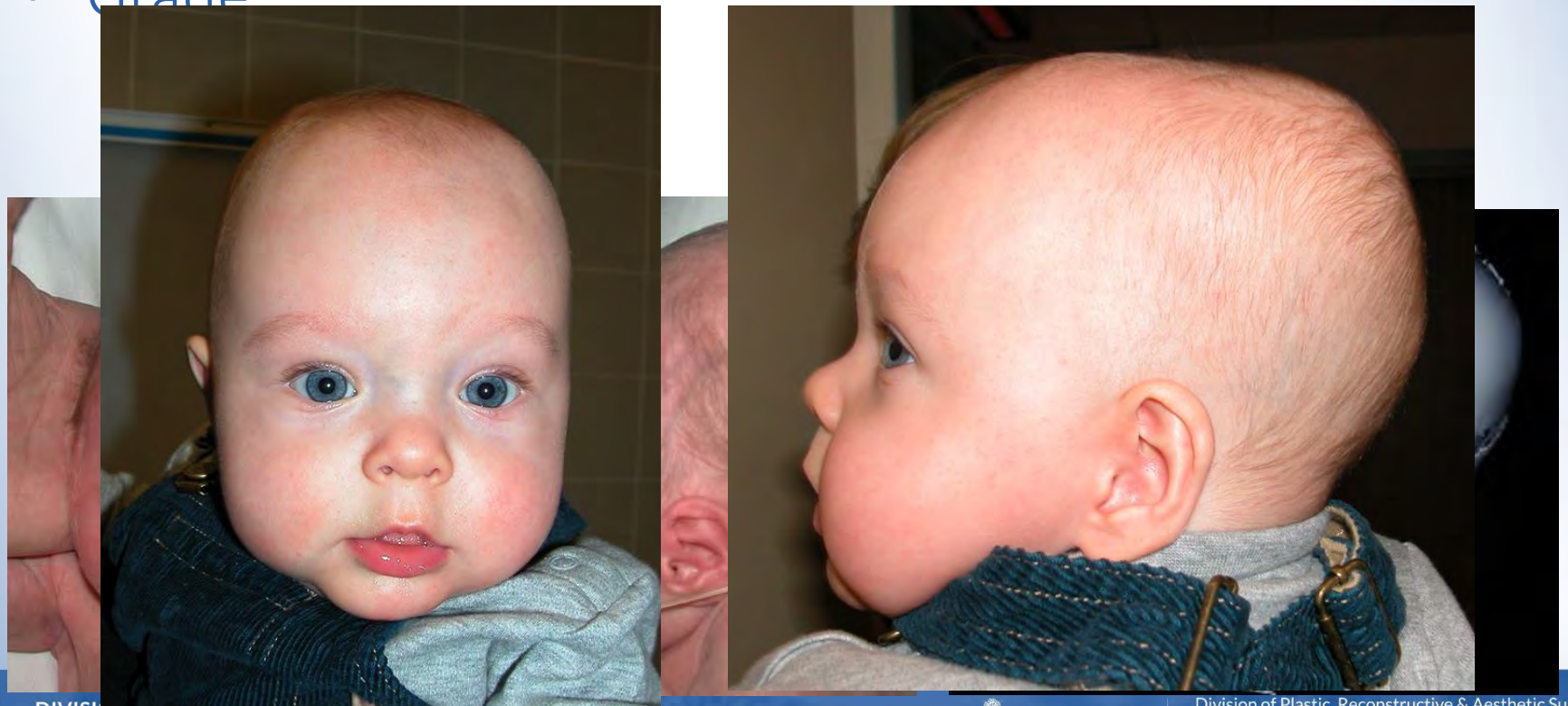
- Natural History of Positional Plagiocephaly:
  - A ing:

6 months

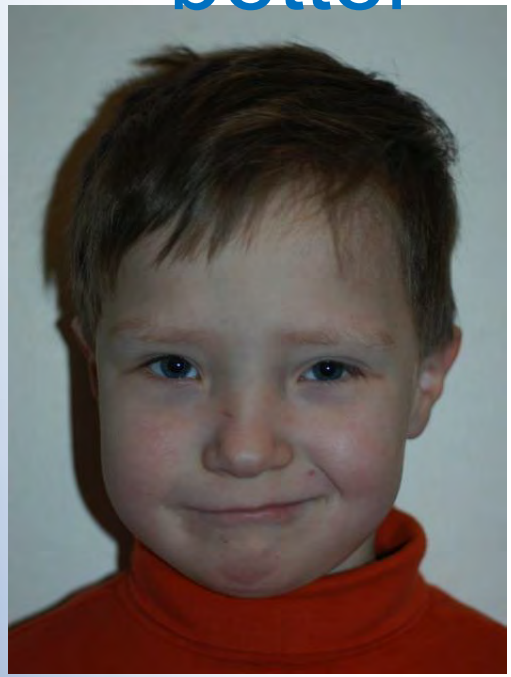


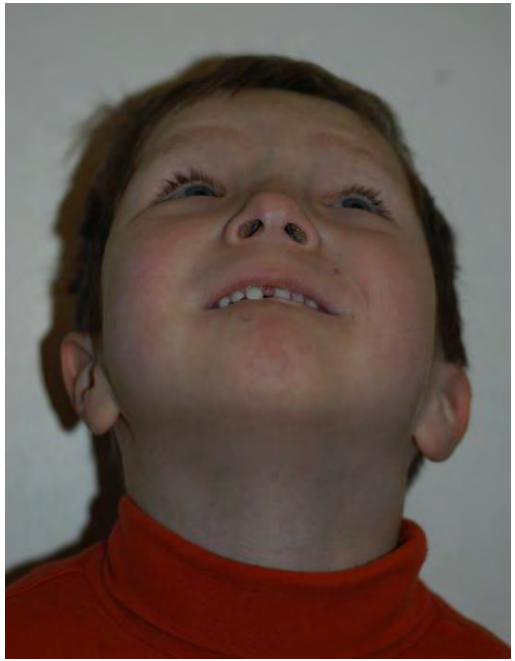
Deformational Plagiocephaly

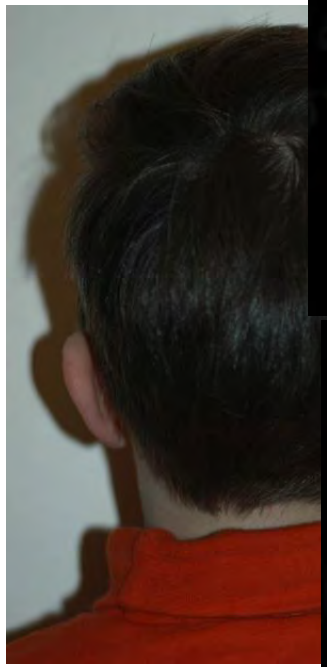
- Grade:

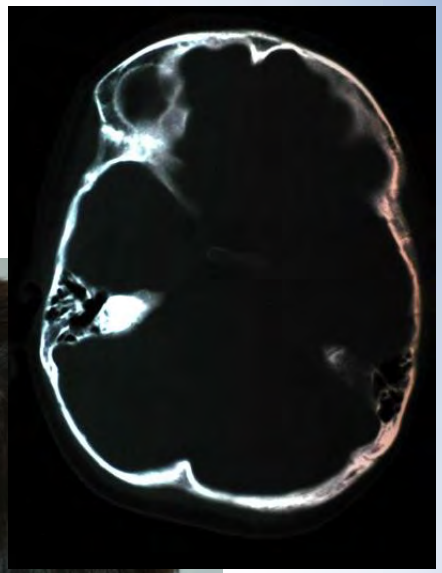


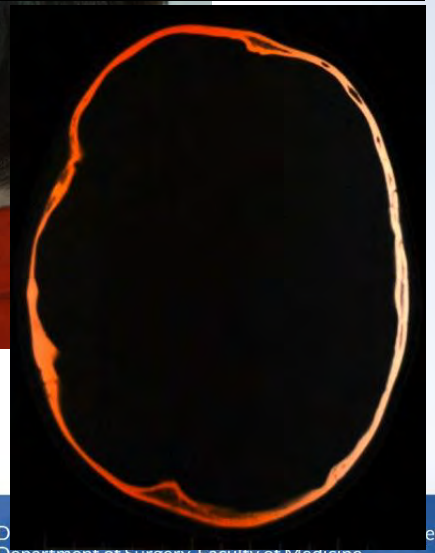
Not all cases get better











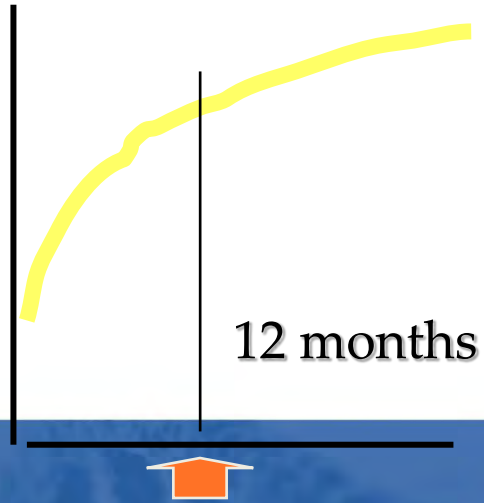
5 yo M

Deformational Plagiocephaly

- What is the natural history of the misshapen neonatal head?
  - Kane et al., Pediatrics, 97:877-885, 1996.
    - "If the calvarial shape abnormality is not corrected before 1 year of age, it remains into adulthood….."
  - Moss, J. Neurosurg. 87:667-670, 1997.
    - "66 (neonates), treated without orthotic devices, showed improvement in average cranial vault asymmetry from 9.2 to 4.7 mm over an average treatment period of 4.5 months that commenced when the average age of the patient was 6.4 months"

Deformational Plagiocephaly

- Cranial vault growth:
  - Cranial vault remodelling to skeletal maturity
  - What age does it essentially stop?



Deformational Plagiocephaly

- Are there any developmental concerns?
  - "the brain doesn't care what shape it is"
    - (C. R. Forrest, 1999 onwards)

  - Long-term developmental outcomes in patients with deformational plagiocephaly.
  - Miller and Claren
  - *Pediatrics 105, 2000*
  - Could deformational plagiocephaly represent a marker for earlyCNS problems subsequently manifested as school problems?

  - 40% (25/63) children with persistent deformational plagiocephaly required special education assistance

Deformational Plagiocephaly

- Molding Helmet Therapy:
  - Helmet treatment for plagiocephaly and congenital muscular torticollis
    - Clarren, Smith and Hanson
    - *J. Pediatrics, January 1979.*
  - *Passive helmet, only 4 of 7 patients compliant, safe and effective treatment*

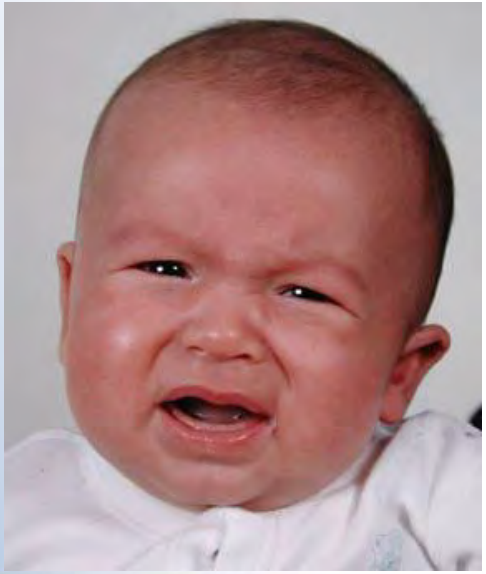
Positional Plagiocephaly

Deformational Plagiocephaly

- Molding Helmet Therapy:
  - Indications
    - Parental concerns
    - Mild-Moderate to Severe
    - Torticollis
    - Ideally 4 to 6 months of age
    - Likely not useful > 12 months

Deformational Plagiocephaly

- Molding Helmet:



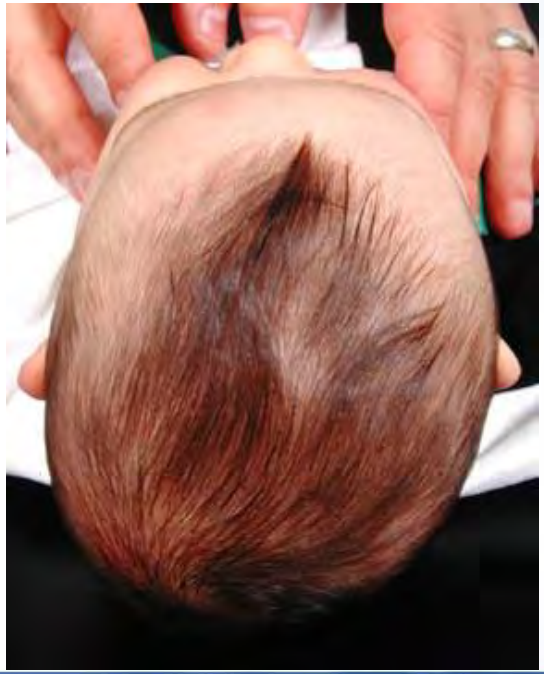
Dec 00

May 01

Deformational Plagiocephaly

- Molding Helmet:

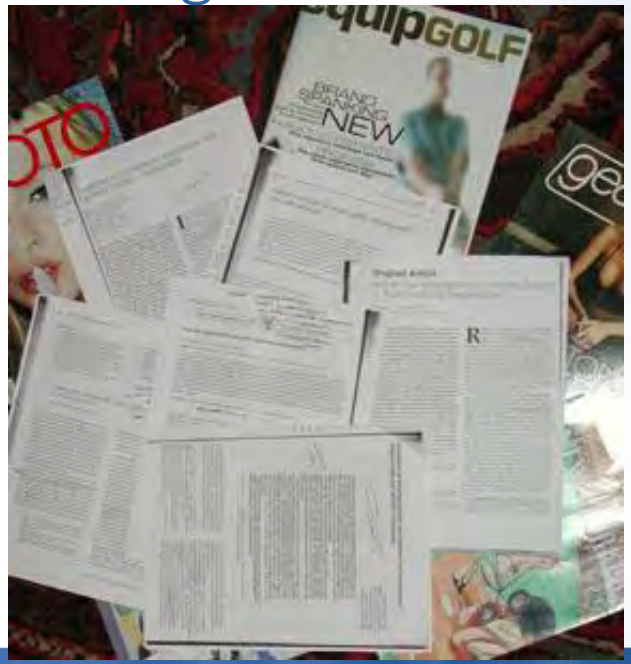
Dec '00



May '01

Deformational Plagiocephaly

- What is the effectiveness of molding helmet therapy?
  - Several articles
  - Industry driven
  - Very few good studies



THE JOURNAL OF CRANIOFACIAL SURGERY

Deformational Plagiocephaly

- Active counterpositioning or orthotic device to treat positional plagiocephaly?
  - Loveday and de Chalain
  - *J. Craniofacial Surgery, 12:308, 2001*

CI and CVAI measured in 29 infants (helmets) and 45 infants (ACP)

ACP versus helmets: 1.9% change versus 1.8% change in CVAI
Helmets 3 times faster (22 versus 64 weeks)

But: still residual asymmetry at end of study (5.4 vs 6.2 CVAI)

What Is the Optimal Time to Start Helmet Therapy in Positional Plagiocephaly?

Susanne Kluba, M.D., D.M.D.
Wiebke Kraut, M.D., D.M.D.
Siegmar Reinert, M.D., D.M.D., Ph.D.
Michael Krimmel, M.D., D.M.D., Ph.D.

Tübingen, Germany

Background: Although helmet therapy is widely accepted in the treatment of severe positional plagiocephaly, treatment regimens, especially regarding starting age, are controversial. This study investigated the importance of starting age to optimize the management of helmet therapy.

Methods: Sixty-two infants with severe positional plagiocephaly were enrolled in this prospective longitudinal study. Twenty-four started helmet therapy before 6 months of age (group 1) and 38 were older than 6 months (group 2). Cranial diagonal measurements were taken. Resulting differences and Cranial Vault Asymmetry Index values were compared and categorized by age at initiation of therapy. The Mann-Whitney *U* test was used for statistical analysis.

Results: Duration of therapy was significantly shorter in group 1 (14 weeks) compared with group 2 (18 weeks) ($p = 0.013$), with significantly better outcomes. The Cranial Vault Asymmetry Index in group 1 was reduced to a normal mean value less than 3.5 percent. Infants in group 2 did not achieve normal values (index value, 4.5 percent) ($p = 0.021$). The relative improvement in asymmetry was significantly better in group 1 (75.3 percent) compared with group 2 (60.6 percent) ($p = 0.001$). After 4 to 11 weeks of treatment, group 1 already showed a better absolute reduction ($p < 0.001$) and a better relative reduction ($p = 0.002$).

Conclusions: Optimal starting age for helmet therapy is months 5 to 6 of life, and early recognition of infants in need is essential. Delaying the onset of treatment significantly deteriorates the outcome. The still often-practiced regimen of starting helmet therapy after physiotherapy should be replaced by a combined therapy in severe cases. (*Plast. Reconstr. Surg.* 128: 492, 2011.)

CLINICAL QUESTION/LEVEL OF EVIDENCE: Therapeutic, II.

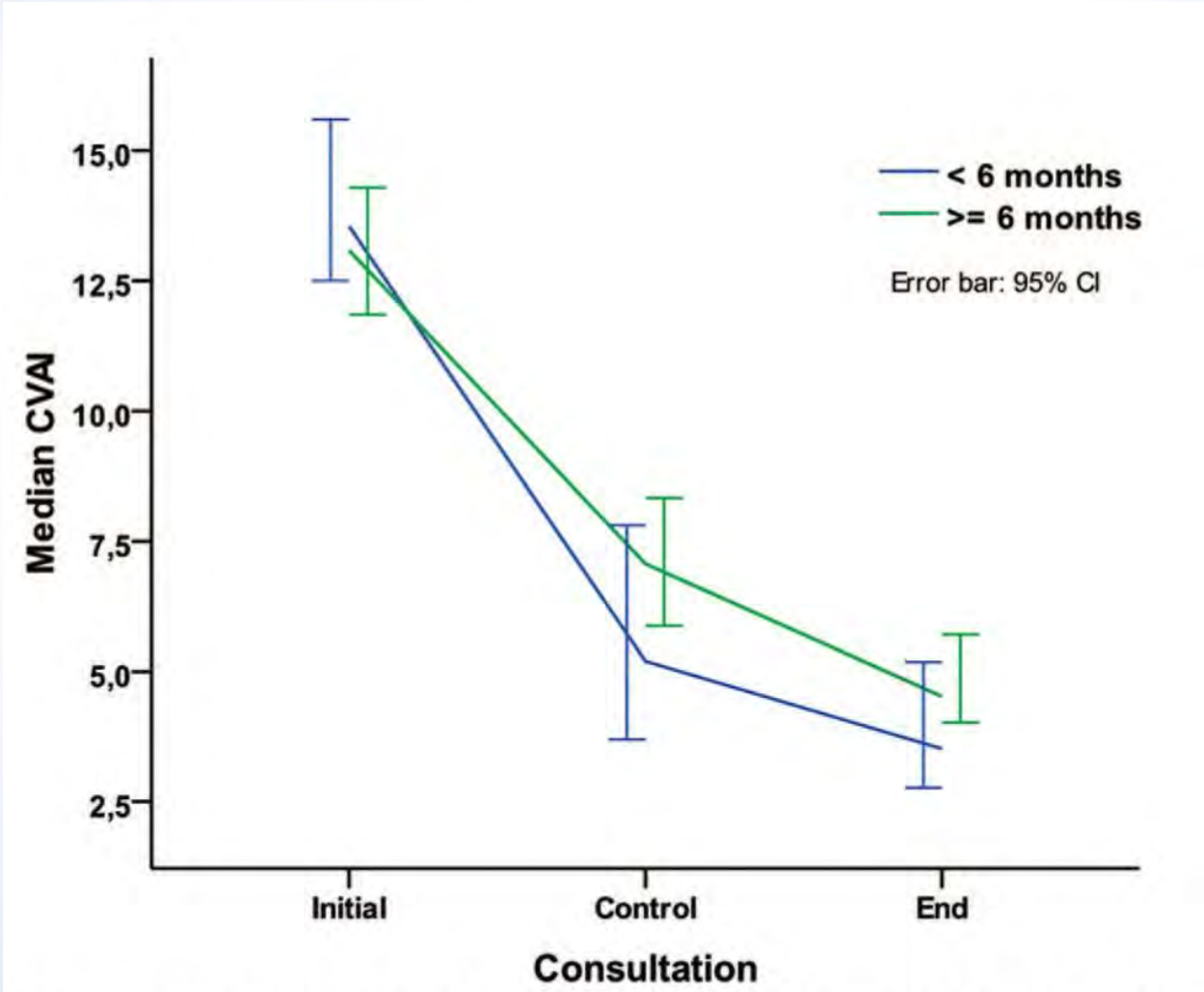


Fig. 3. Course of Cranial Vault Asymmetry Index (*CVAI*) values. *CI*, confidence interval.

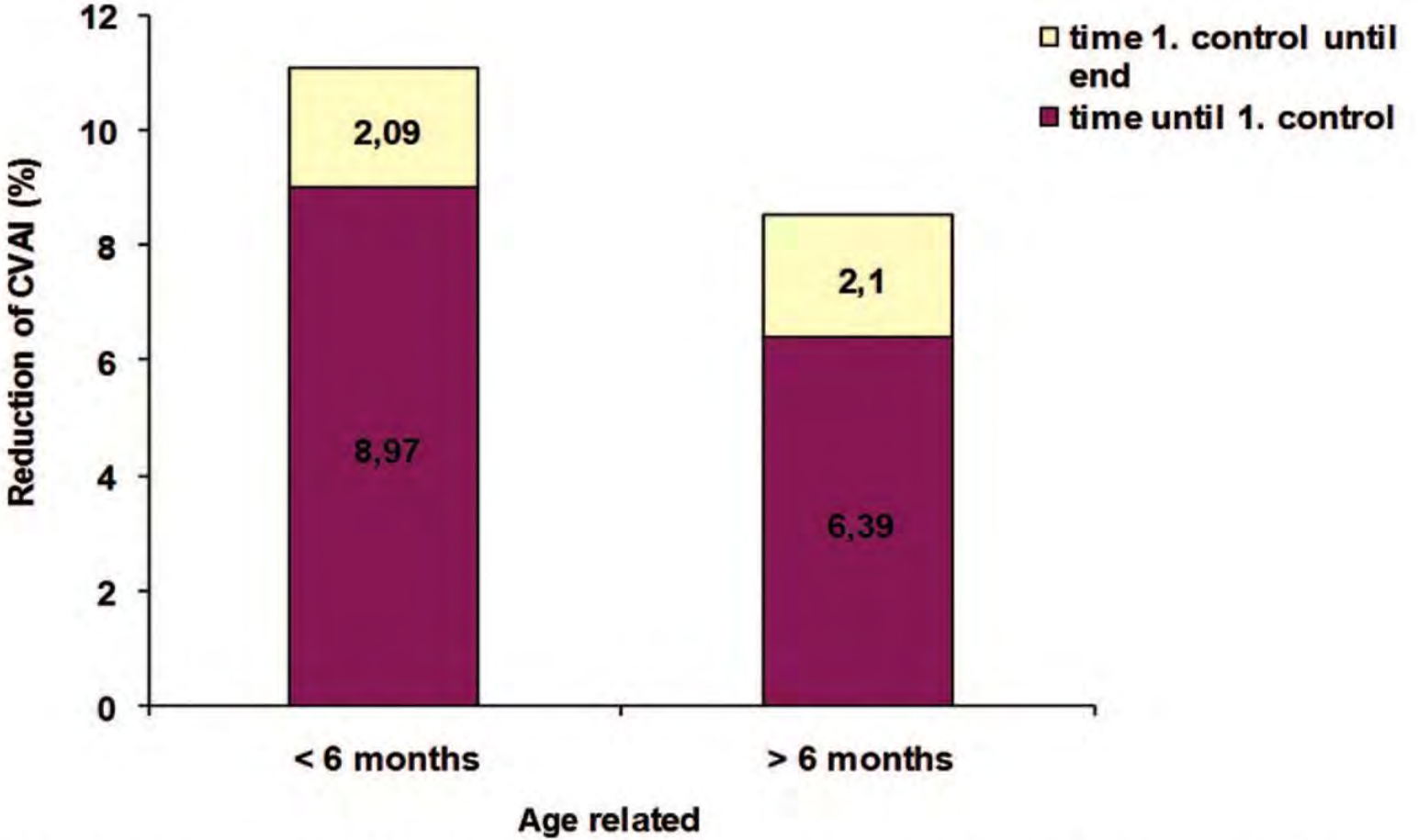


Fig. 4. Comparison of treatment dynamics. *CVAI*, Cranial Vault Asymmetry Index.

Positional Plagiocephaly

- Does surgery have a role?
  - Lambdoid synostosis. Review of cases managed at the Hospital for Sick Children, 1972-1982
  - Muakkassa et al.
  - *J. Neurosurgery 61: 340-347, 1984*

  - 74 cases, advocated early (< 6 months) surgery
  - "when the lambdoid suture closes, the occiput on the involved side is flat, the forehead on the same side tends to bulge forward and the ear on this side adopts a low and forward position"

Hmmmm.....

Surgery may ultimately be indicated for select cases with established deformation

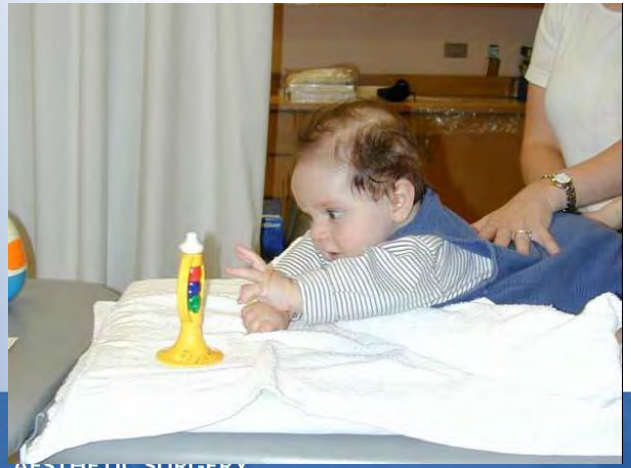
Deformational Plagiocephaly

- Guidelines for referrals:
  - 1. Torticollis
  - 2. Positional plagiocephaly not responsive to counterpositioning
  - 3. Parents interested in molding helmets
    - between 4 to 12 months of age
  - 4. Patient > 3 years (surgical candidate)

- Not good indications for referrals
  - 1. "to check the shape of the head"
  - 2. "to rule out craniosynostosis"
  - 3. "fontanelles closing fast"

Positional Plagiocephaly

- EDUCATION AND INFANT POSITIONING
  - Back to sleep
  - Reduce amount of time a baby spends on its back
  - Repositioning baby on a regular basis
  - Encourage prone play during wakeful times

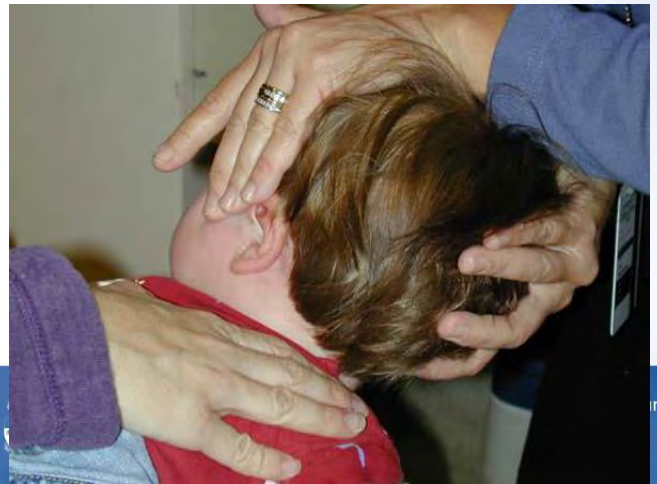


Positional Plagiocephaly

- EARLY DETECTION
  - During well baby checks screen for positional preference in terms of equal neck rotation
  - Identifying early signs of skull and facial asymmetry

Positional Plagiocephaly

- EARLY INTERVENTION
  - Active stimulation of neck rotation to opposite side
  - Rule out secondary torticollis
  - Once positional preference identified, recommend placing infant in 1/4 side sleeping position from supine for sleeping
  - Tummy time



Positional Plagiocephaly

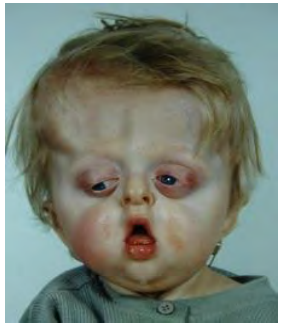
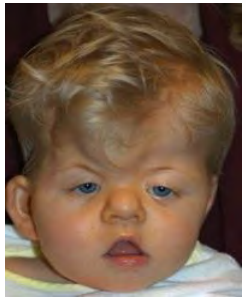
- Summary:
  - 1.Current "epidemic" of positional plagiocephaly
  - 2. Diagnosis clinical
  - 3. CT for rare cases (better than plain films)
  - 4. Refer cases
    - Pattern doesn't fit
    - Parents interested in helmet
    - Torticollis
  - 5. Prevention is key
  - 6. CHIP initiative
    - Multi-directed initiative designed to prevent the development of positional plagiocephaly
    - Paediatricians, Family Doctors, Nurses, Midwives, OTs, PTs
    - Linkage with Canadian Pediatric Society
    - Care not to confuse "Back to Sleep" message

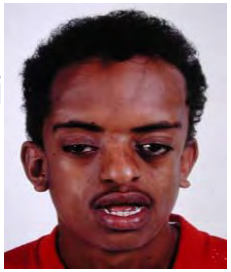

"When I'm awake with mummy, I'm on my tummy"
THE HOSPITAL FOR SICK CHILDREN
HELP PREVENT INFANT FLATHEAD SYNDROME, TURN YOUR BABY


Why time on his tummy is good for his head.
I'M HAVING TUMMY TIME NOW

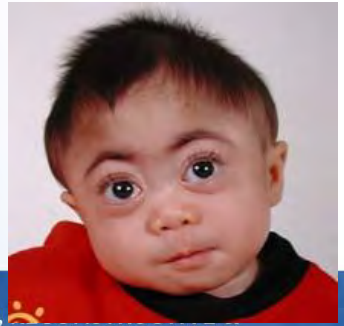
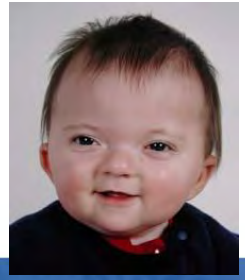
Syndromic Craniosynostosis

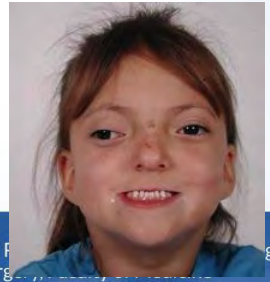
- Syndromic Craniosynostosis
- (180 different syndromes)
  - Crouzon
  - Apert
  - Saethre-Chotzen
  - Muenke
  - Pfeiffer
  - Craniofrontonasal dysplasi



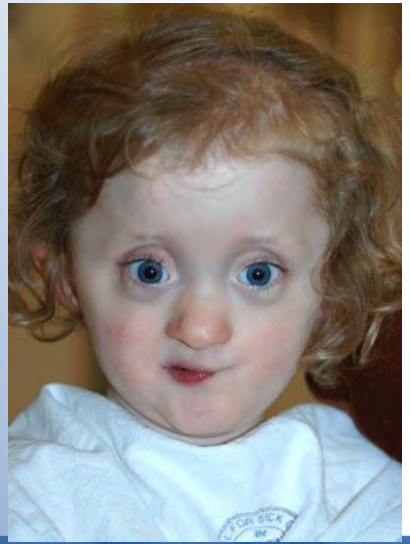
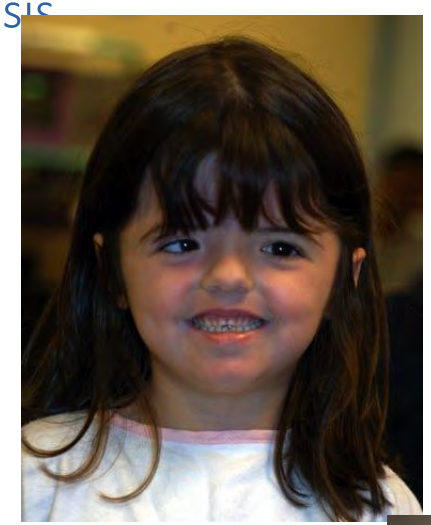
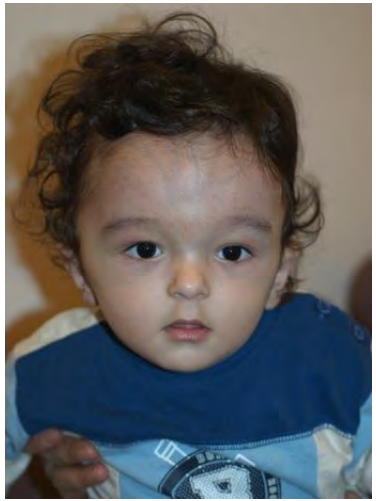
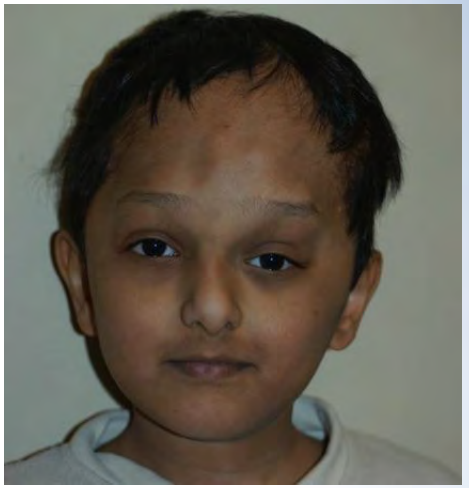
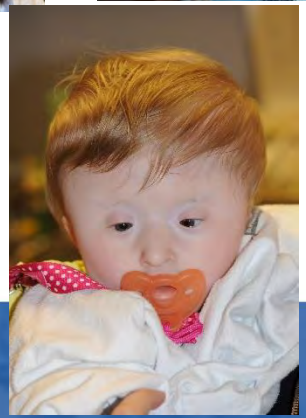


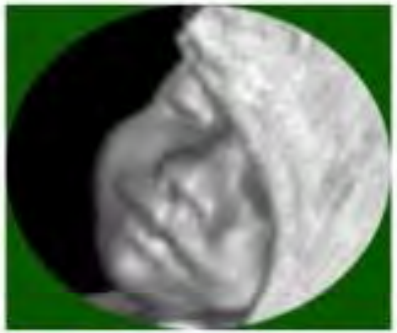
Syndromic Craniosynostosis

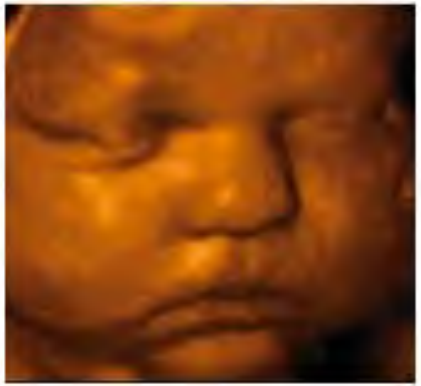
- Unrecognized: facial dysmorphism associated with craniosynostosis

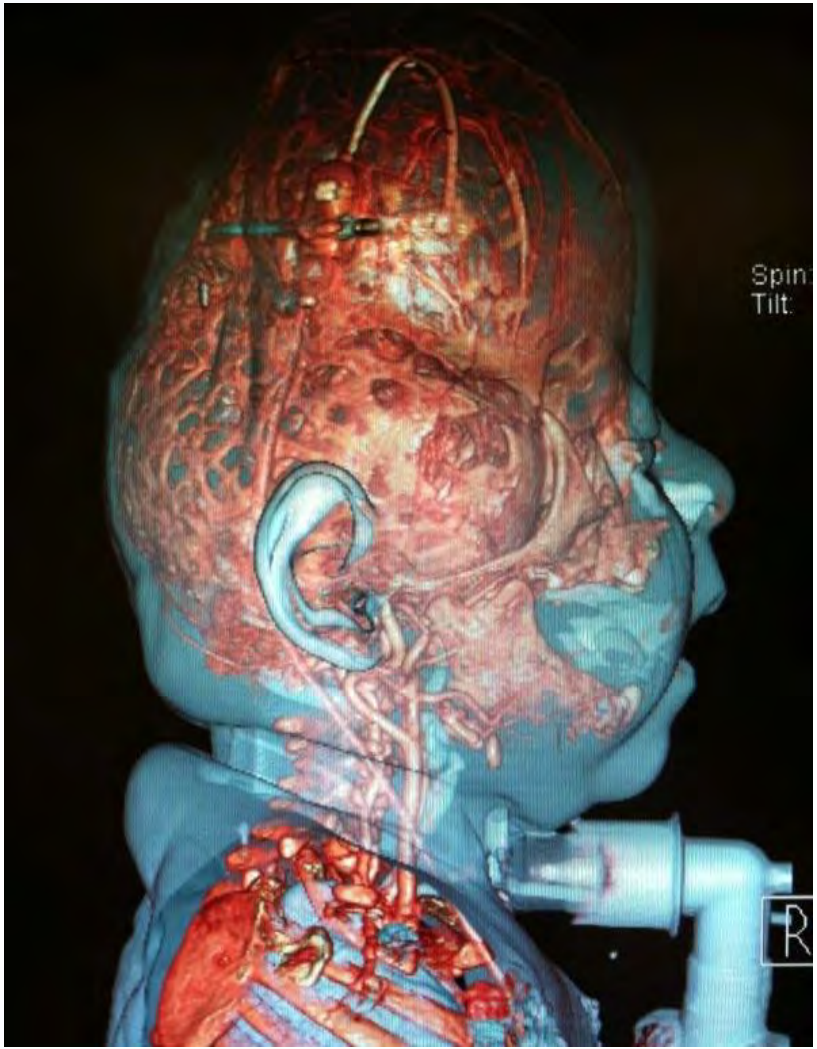

Syndromic Craniosynostosis

Prenatal diagnosis and intervention





High parental expectations


Spin
Tilt:
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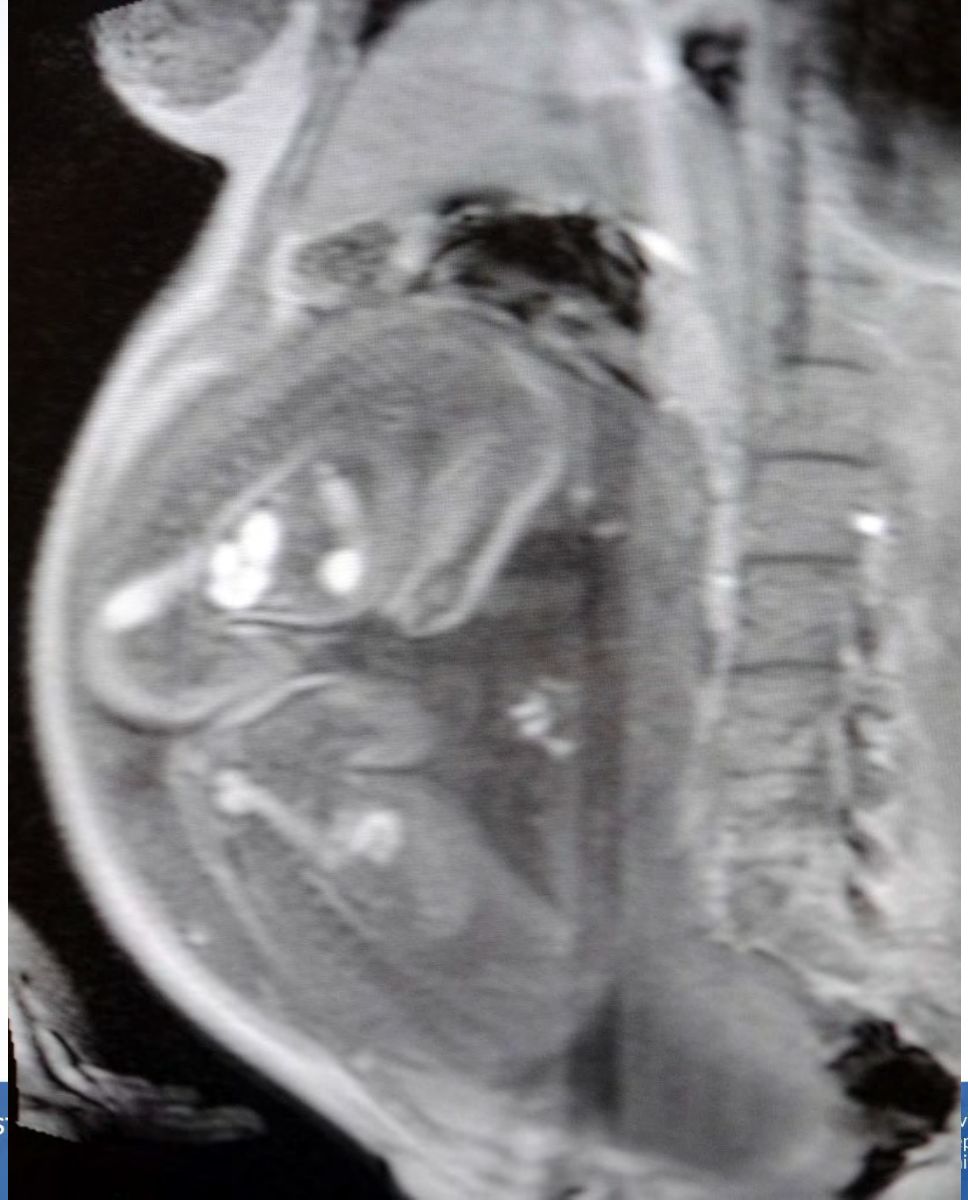

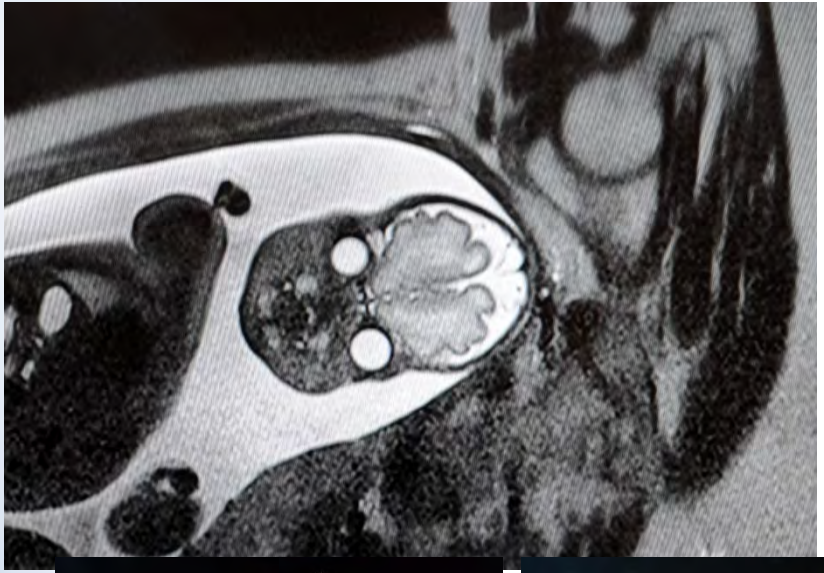

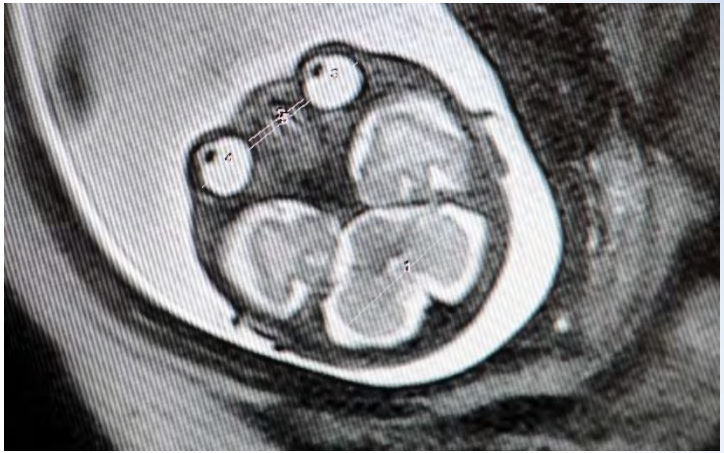

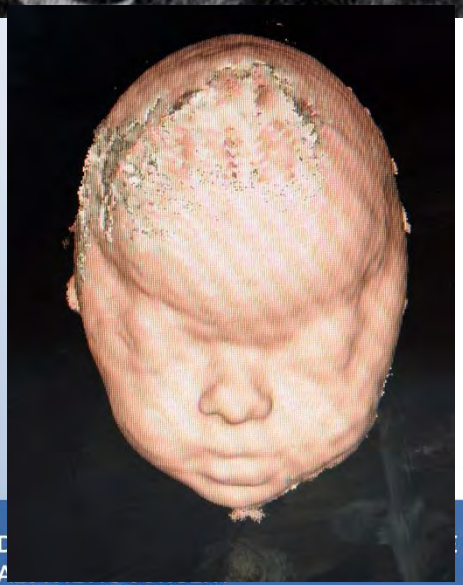

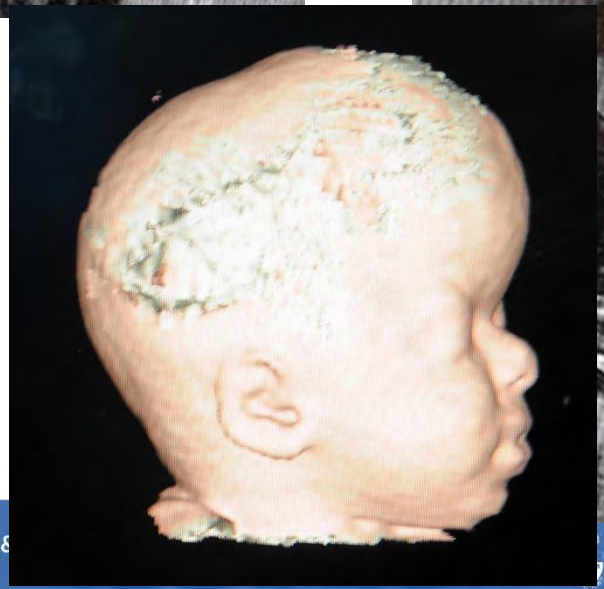

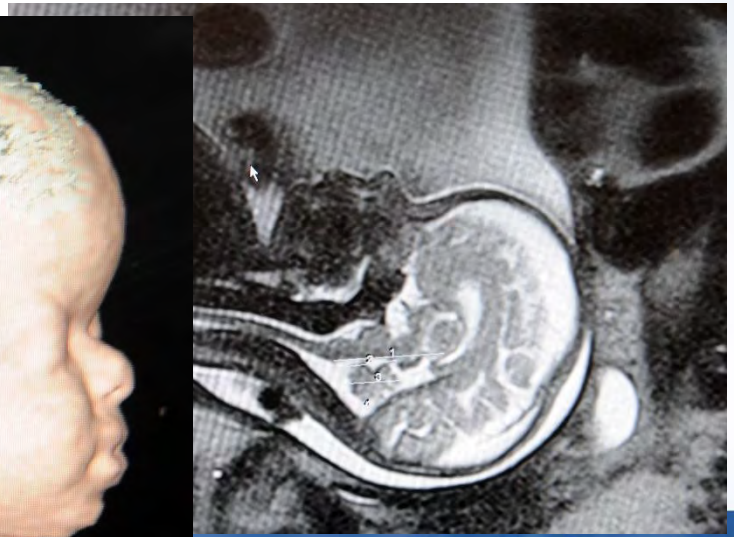

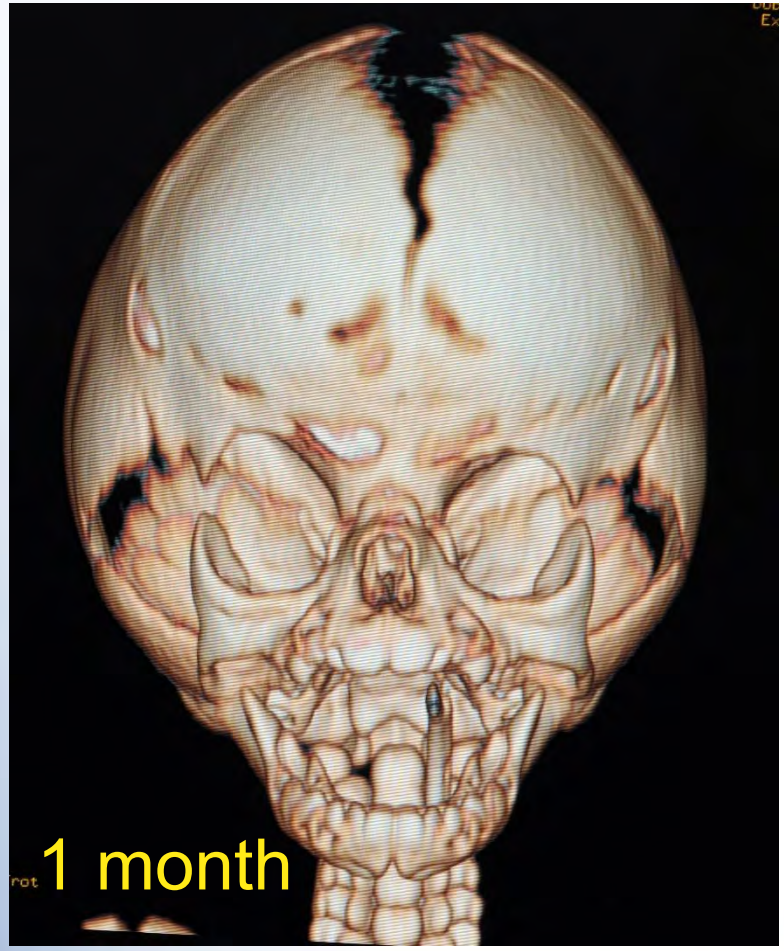
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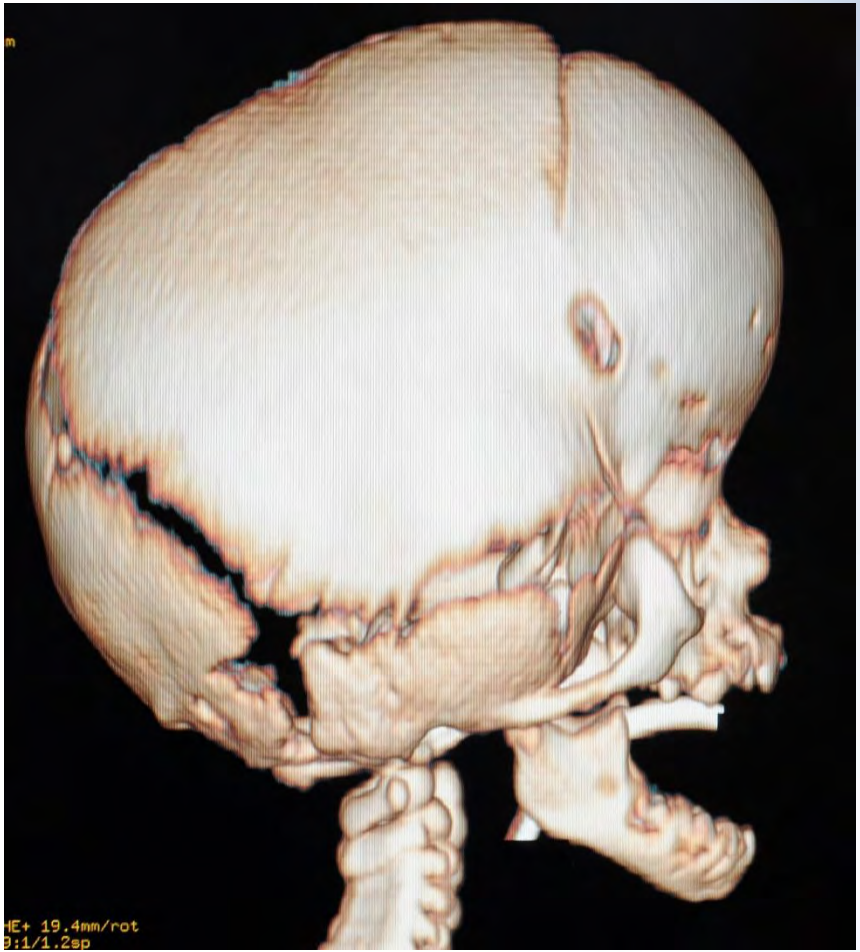
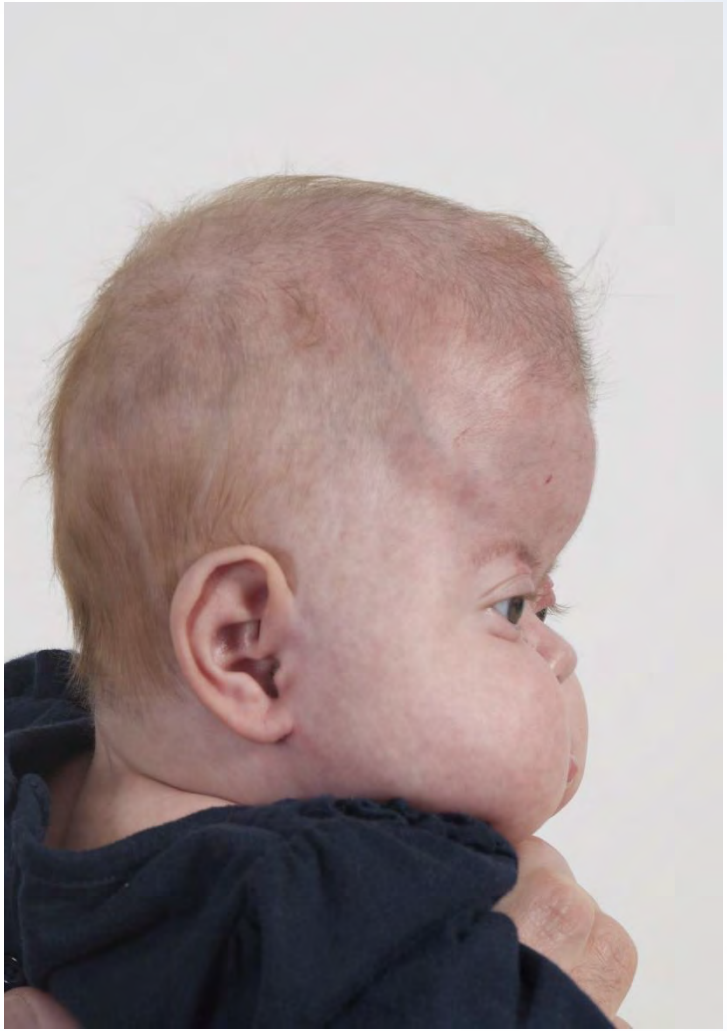
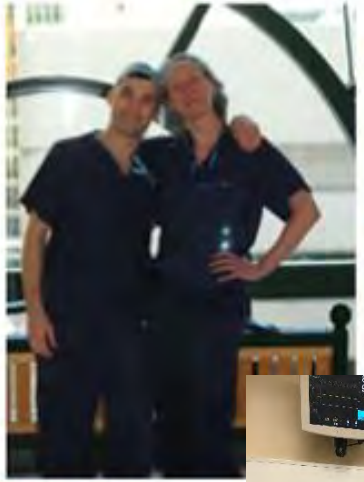








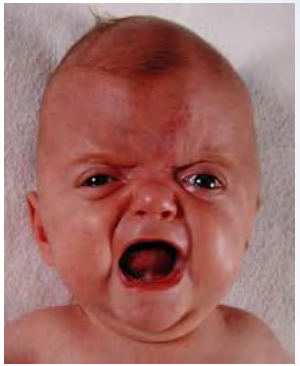


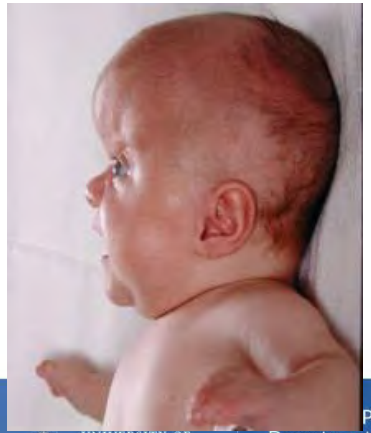


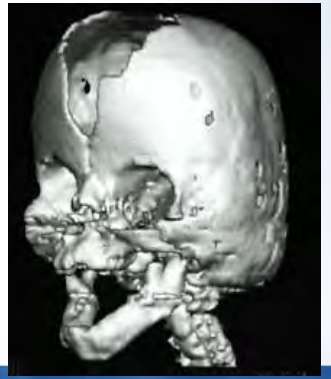



Syndromic Craniosynostosis

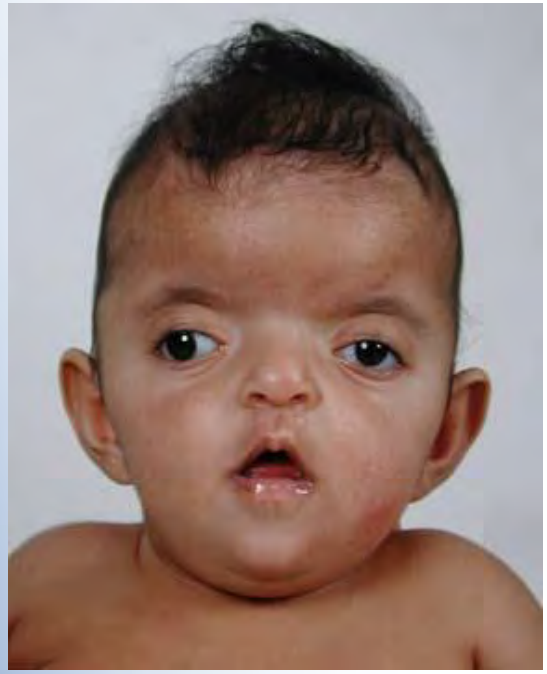
- Apert syndrome (1906)
  - 1 in 60,000 - 100,000 births
  - Autosomal dominant or sporadic (majority)
  - FGFR 2 mutations - Ser252Trp and Pro253Arg
  - Clinical features
    - Bicoronal synostosis
    - Midface hyoplasia
    - Hypertelorbitism
    - Complex acrocephalosyndactyly
    - Acne vulgaris
    - Cleft palate (30%)
    - Deafness (30%)
    - Hydrocephalus (5-10%) - venous drainage
    - Developmental delay
    - Cerebellar tonsillar herniation

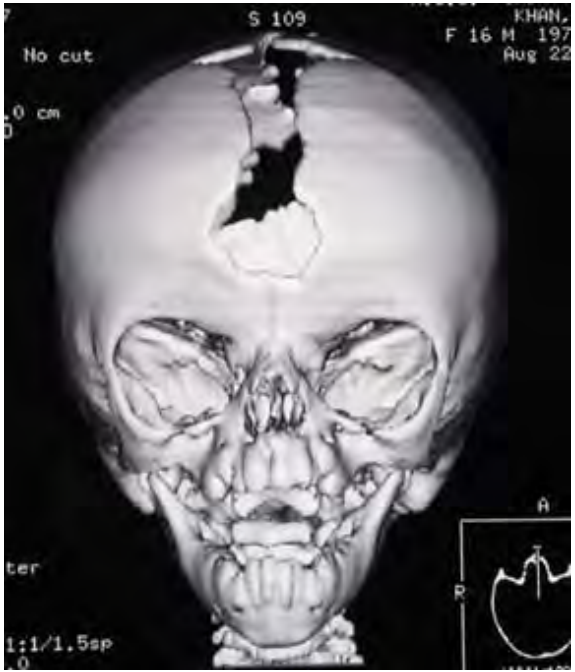


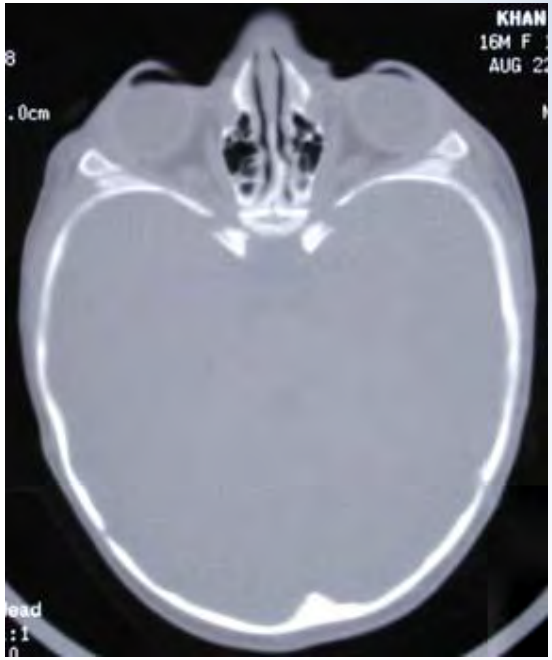




Apert syndrome







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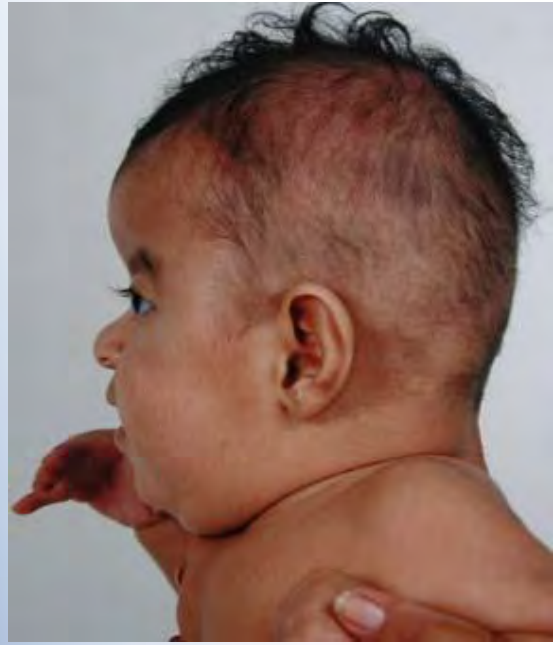
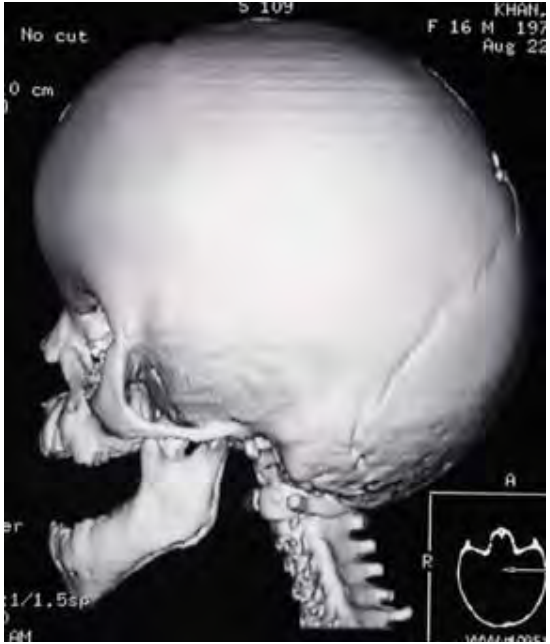
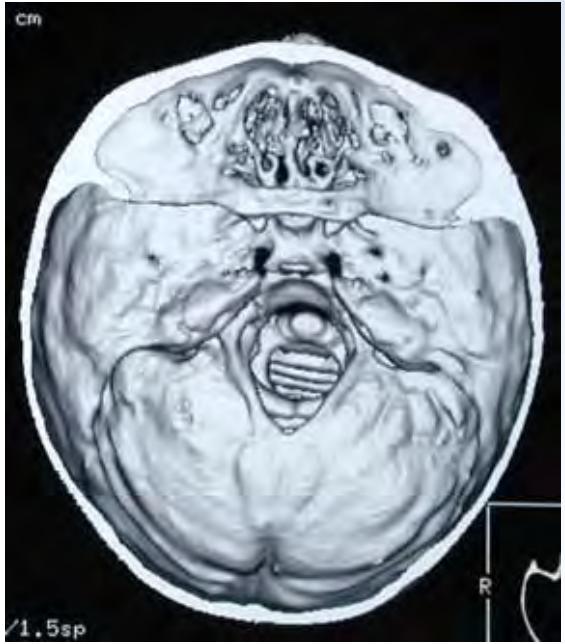
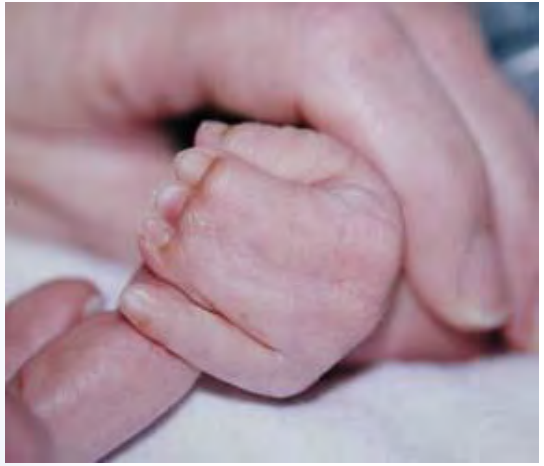
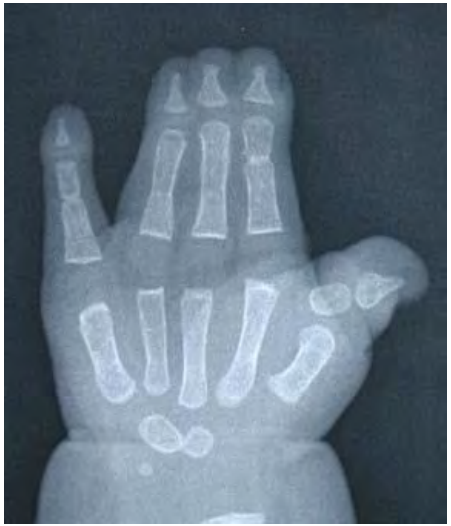


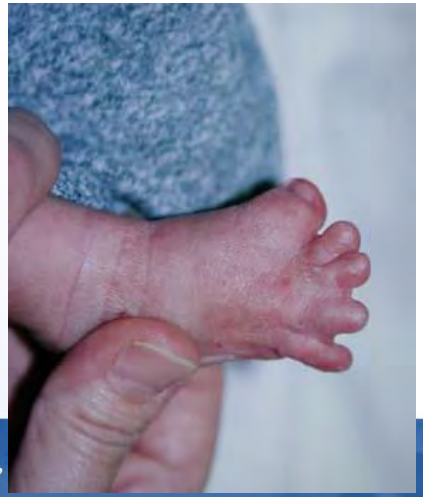


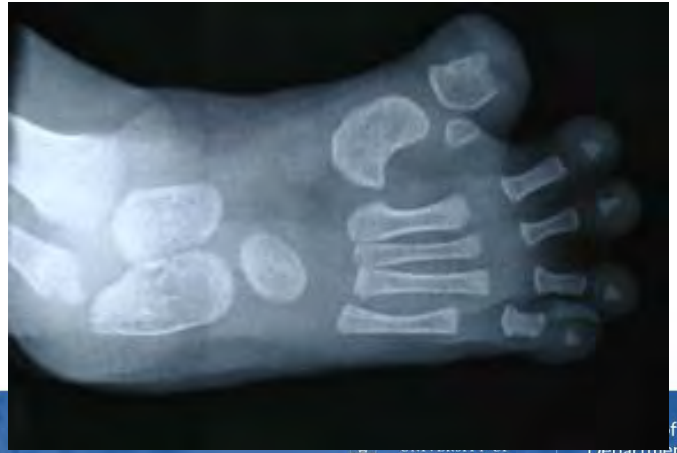


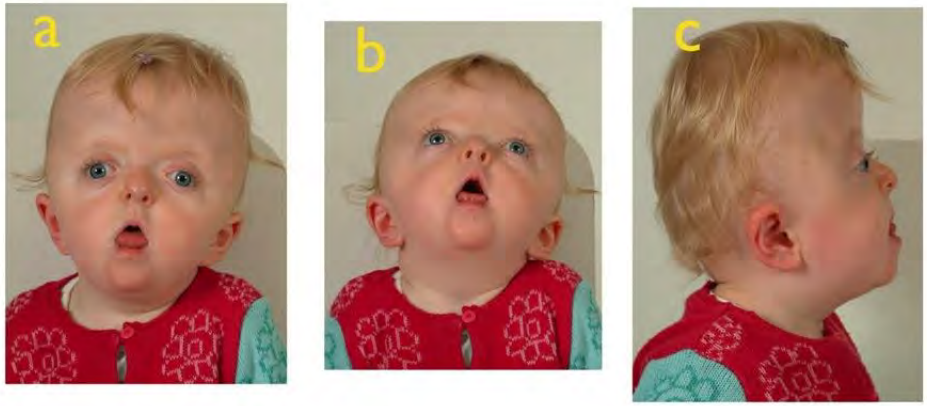
Apert syndrome







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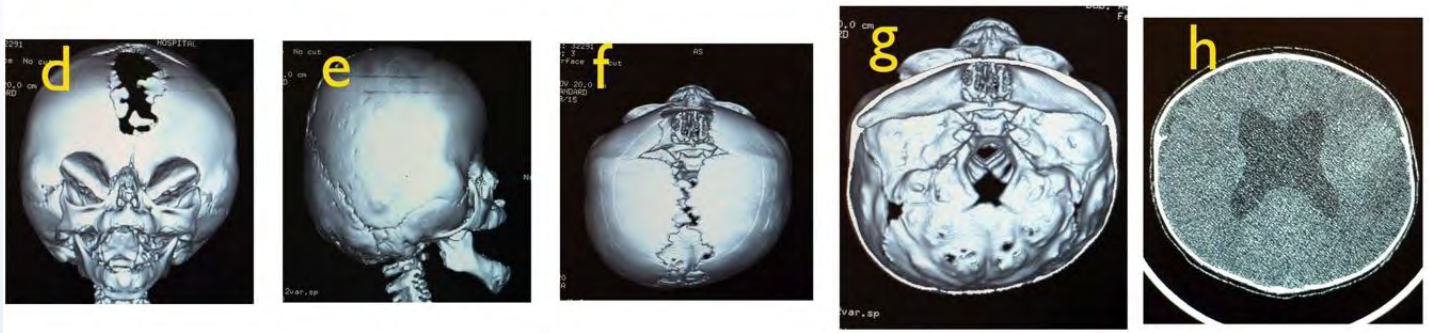
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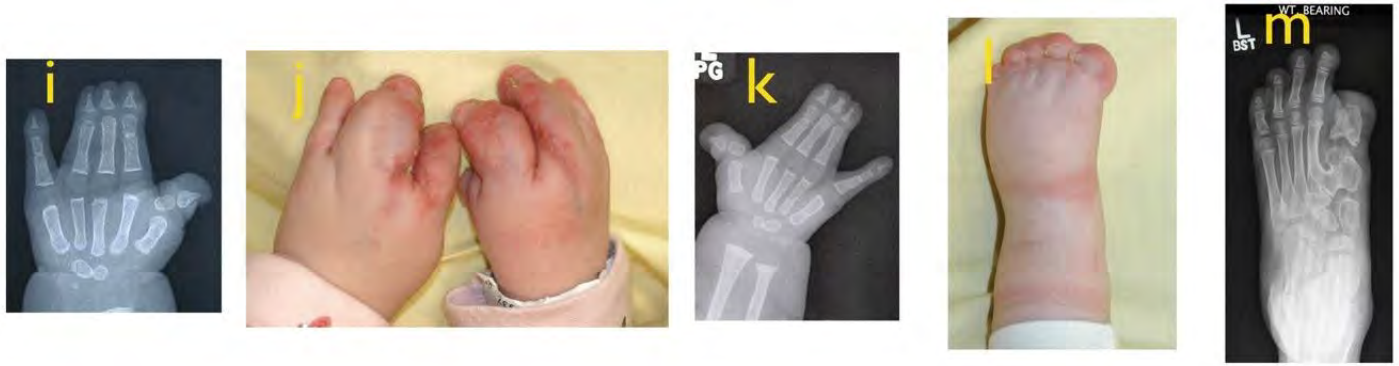
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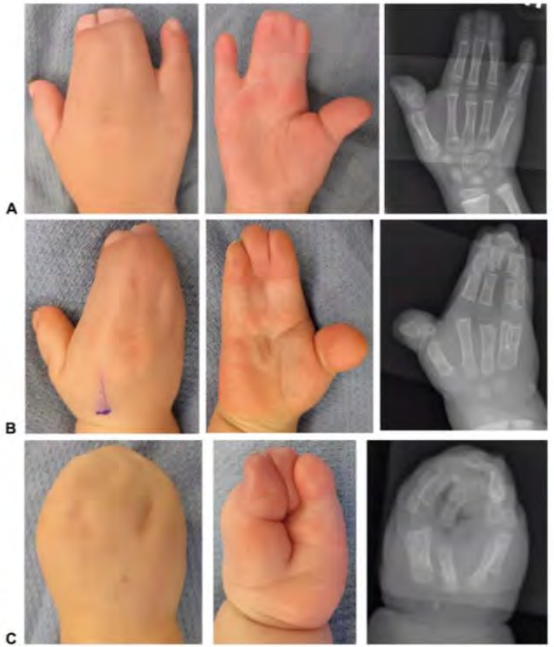
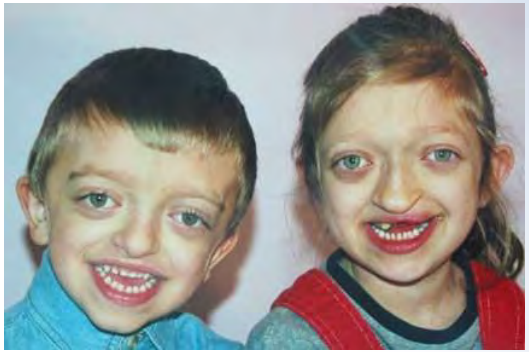
APERT HAND FEATURES

	Type I (*"spade"*)	Type II (*"mitten"*)	Type III (*"rosebud"*)
First webspace	Simple Syndactyly	Simple Syndactyly	Complex Syndactyly
Middle three fingers	Side-to-side fusion with flat palm	Fusion of fingertops forming a concave palm	Tight fusion of all digits with one conjoined nail
Fourth webspace	Simple and incomplete syndactyly	Simple and complete syndactyly	Simple and complete syndactyly

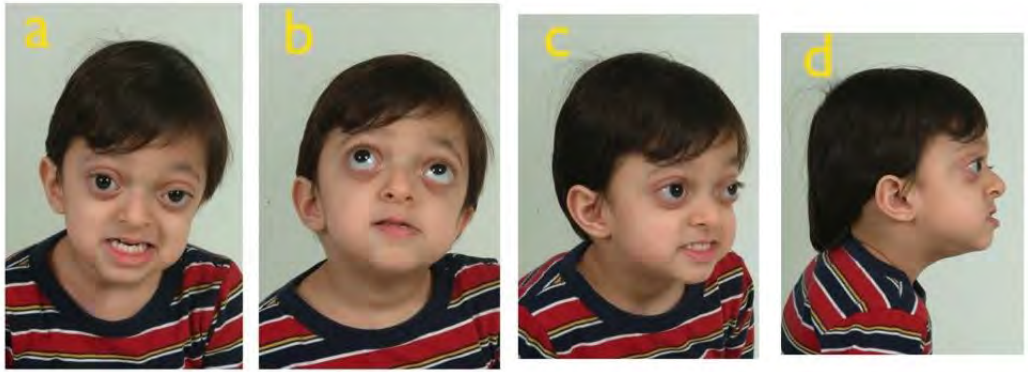


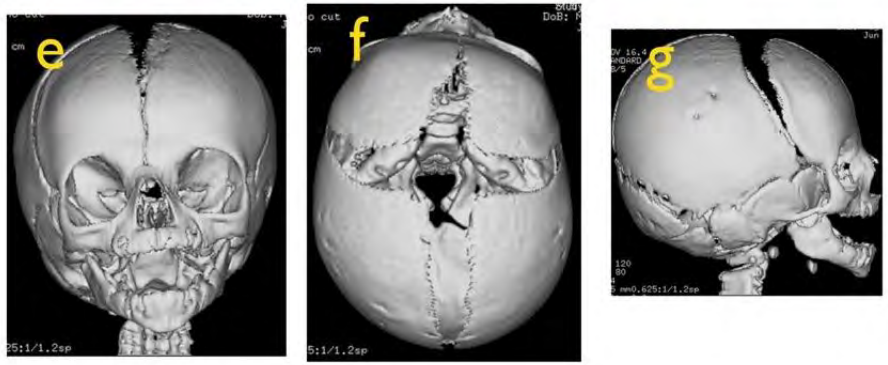
"bad hands, good airway and face" and "good hands, bad airway and face"
- GOSH

Syndromic Craniosynostosis

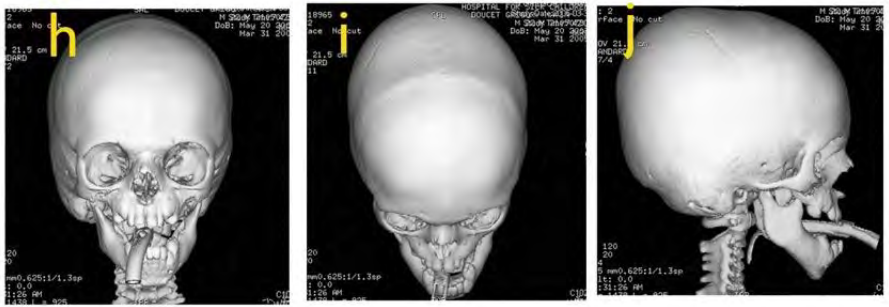


- Crouzon syndrome (1912)
  - 4 key characteristics
    - Exorbitism
    - Retromaxillism
    - Inframaxillism
    - Paradoxical retrogenia
  - 1 in 25,000 births
  - Autosomal dominant with near complete penetrance and variable expressivity
  - FGFR 2 mutation
  - FGFR 3 mutation - Crouzon's with acanthosis nigricans
  - Clinical features
    - Normal intelligence
    - Craniosynostosis (sagittal, metopic, early versus late bicoronal)
    - Hypertelorbitism
    - Midface retrusion





Age 3 months

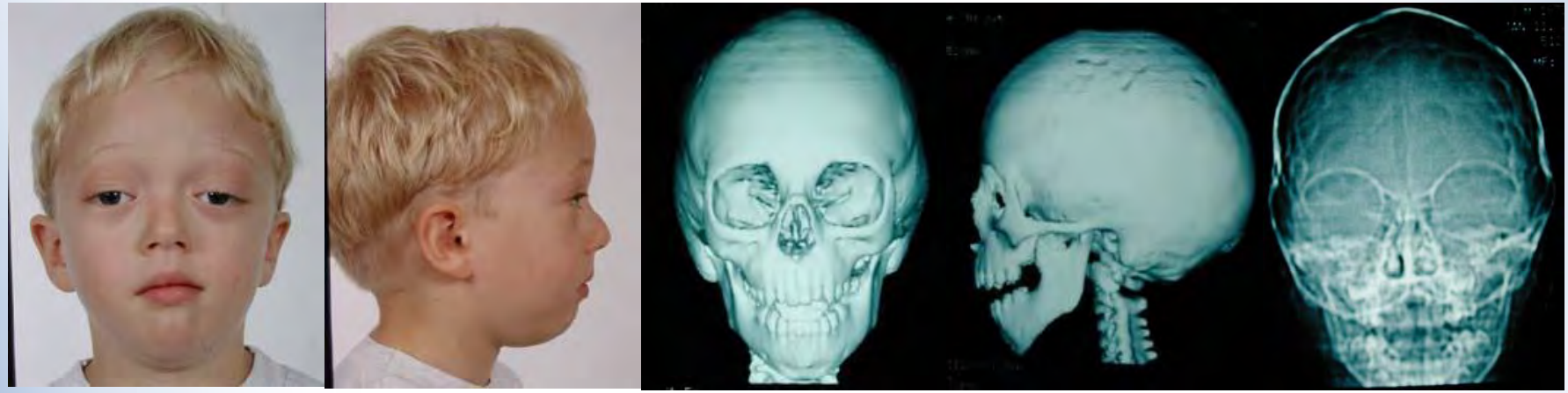


Age 3 years

Crouzon Syndrome

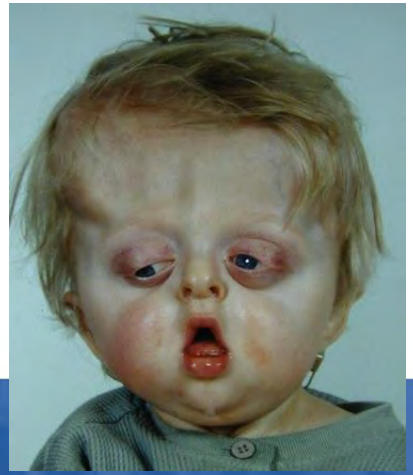
Normocephaly

headaches, elevated ICP



Syndromic Craniosynostosis: Pfeiffer syndrome

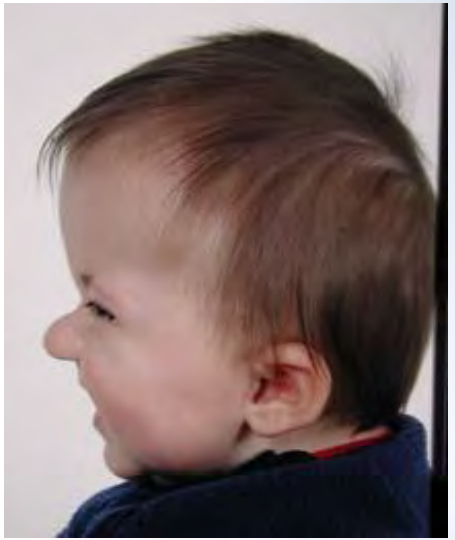
- Brachycephaly with midface deficiency
  - Pfeiffer's syndrome (1963)
  - Autosomal dominant with complete penetrance
  - FGFR1and FGFR2 gene mutations
  - Variable expressivity in phenotype
  - Craniosynostosis, orbital dystopia, midface hypoplasia, broad and medial deviated thumbs and great toes, partial syndactyly



Syndromic Craniosynostosis

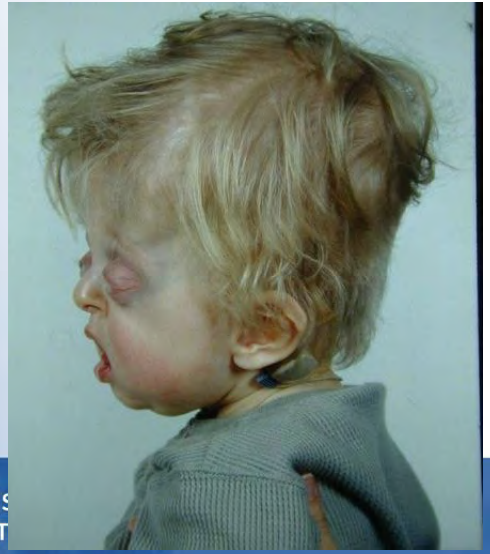
Pfeiffer Syndrome: Type I (Cohen. 1993)

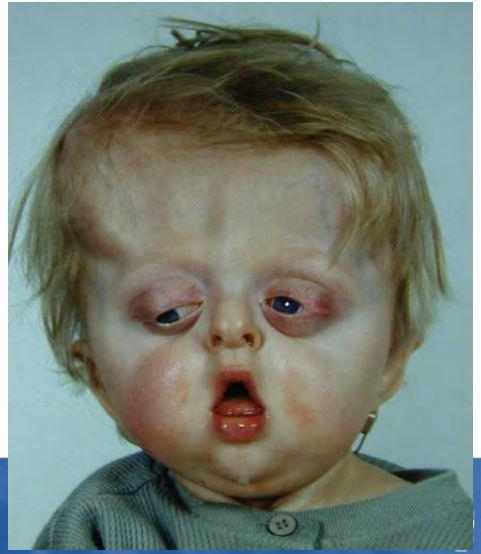
- Near normal IQ
- Bicoronal synostosis, variable midface hypoplasia, minimal ocular proptosis
- Hydrocephalus absent

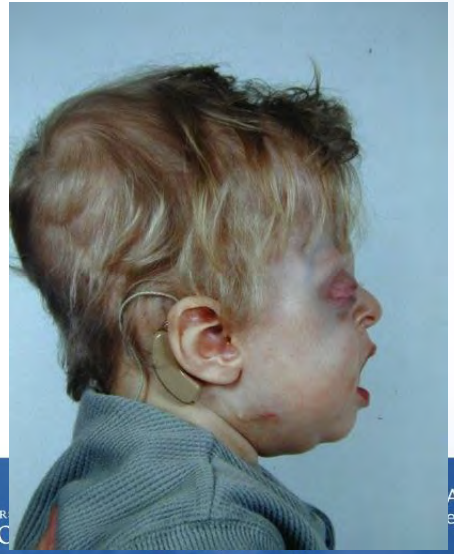


Syndromic Craniosynostosis

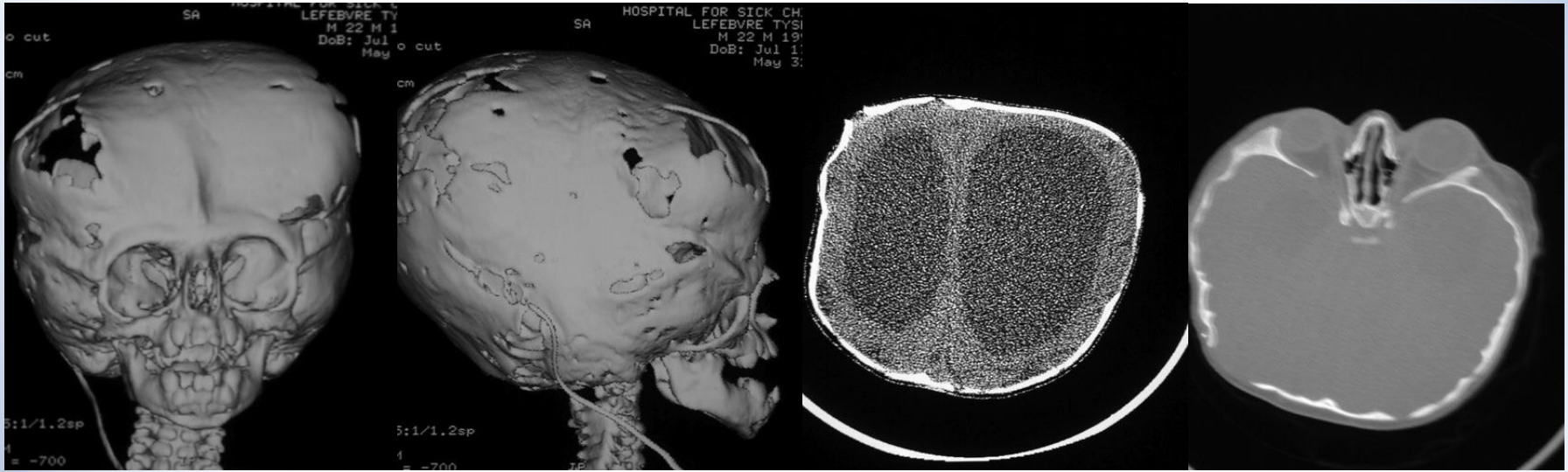
- Pfeiffer Syndrome: Type II (cloverleaf type) (Cohen. 1993)
  - Cloverleaf skull
  - Severe midface deficiency and exorbitism
  - Hydrocephalus
  - Elbow ankylosis, broad thumbs and great toes
  - Multiple other anomalies
  - Limited life span

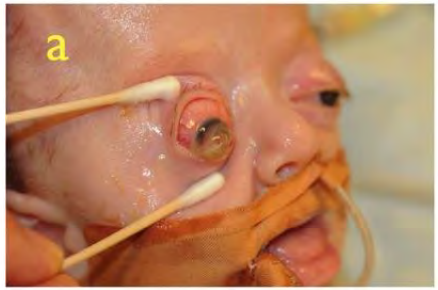






Pfeiffer Syndrome: Type II (cloverleaf type)

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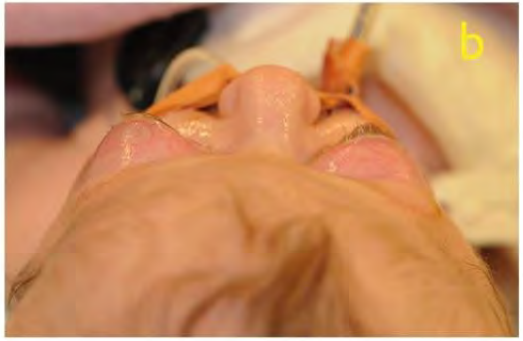
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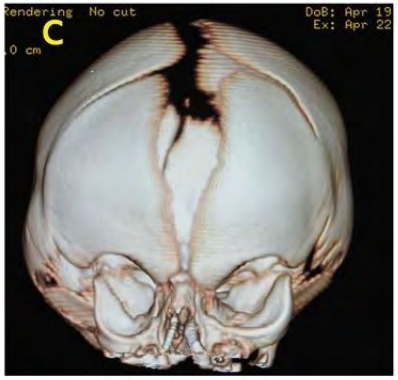
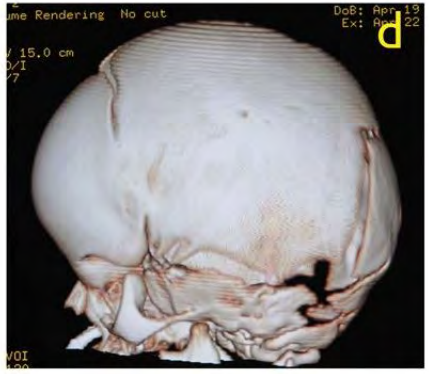

Rendering No cut
DoB: Apr 19
Ex: Apr 22
c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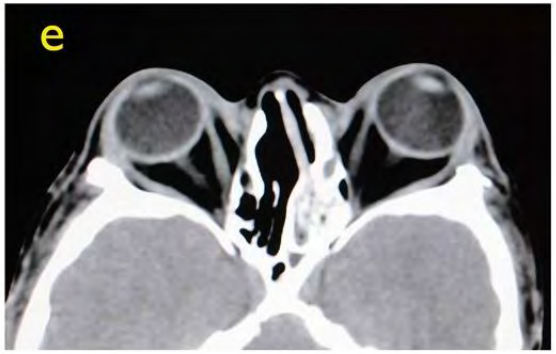
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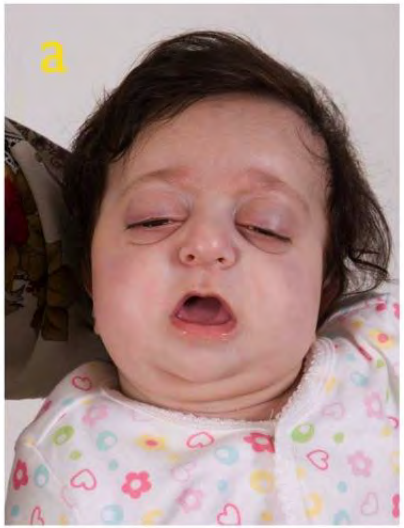
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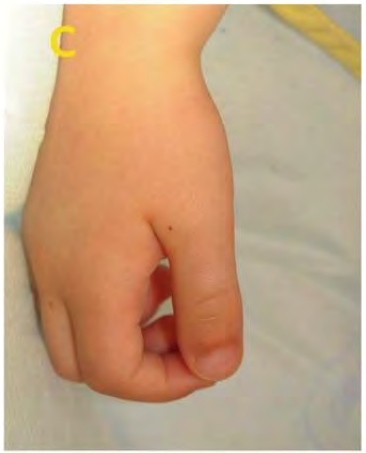
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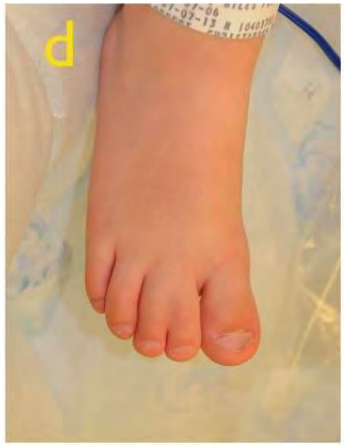
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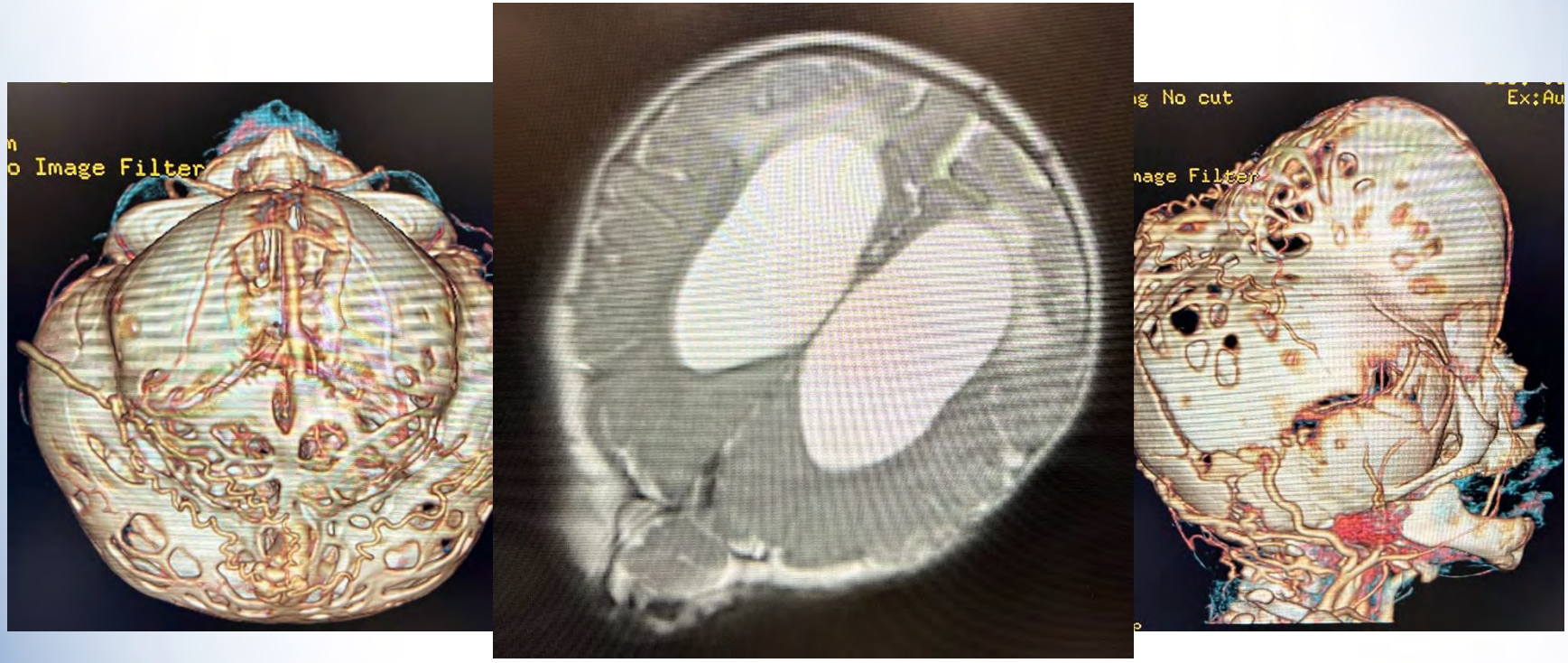
Pfeiffer Syndrome: Type II (cloverleaf type)

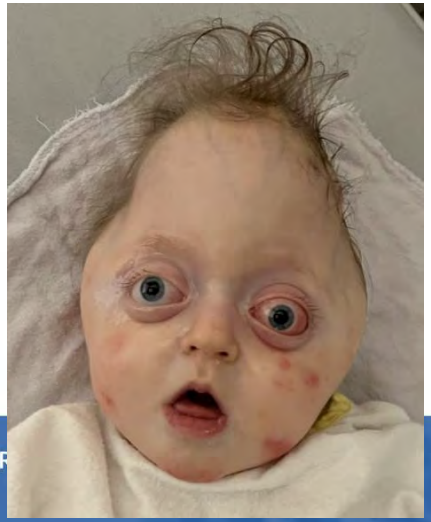


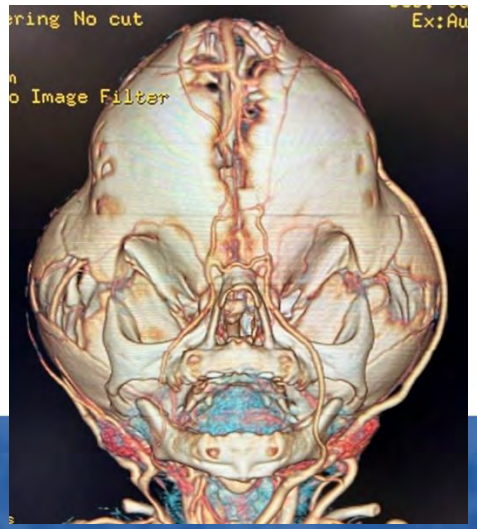
Syndromic Craniosynostosis

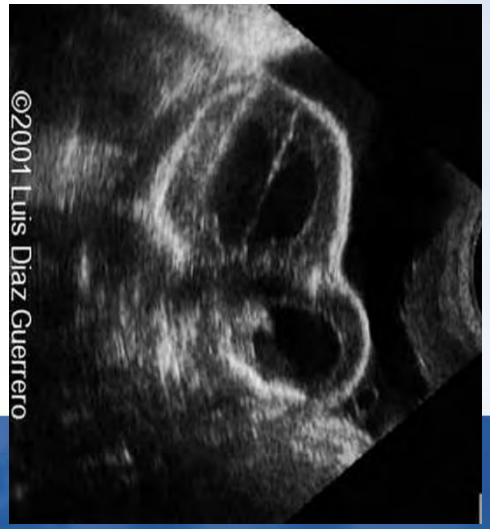
- Pfeiffer Syndrome: Type III (non-cloverleaf type)
  - Same as Type II but no cloverleaf skull
  - Visceral abnormalities
  - Severe neurologic compromise
  - Short life span

Syndromic Craniosynostosis

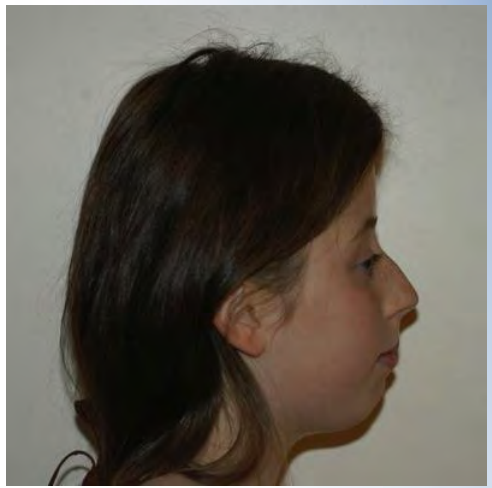
- Kleeblattschädel or cloverleaf skull deformity
  - Not a syndrome per se
  - bicoronal and bilateral lambdoid synostosis with delayed fusion of the sagittal and squamosal sutures
  - May be associated with any of the craniofacial dysostosis syndromes
  - Associated with high morbidity and mortality



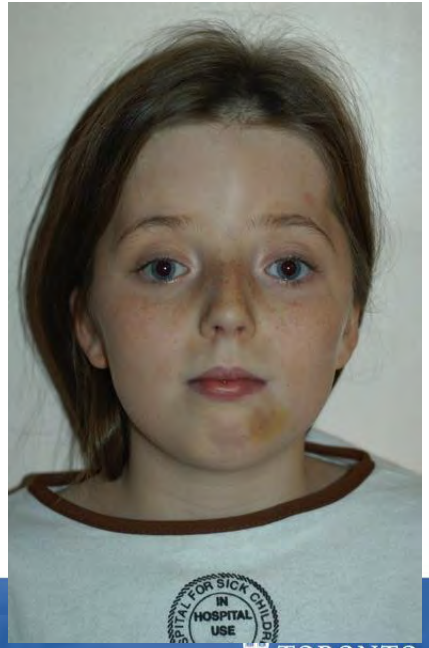


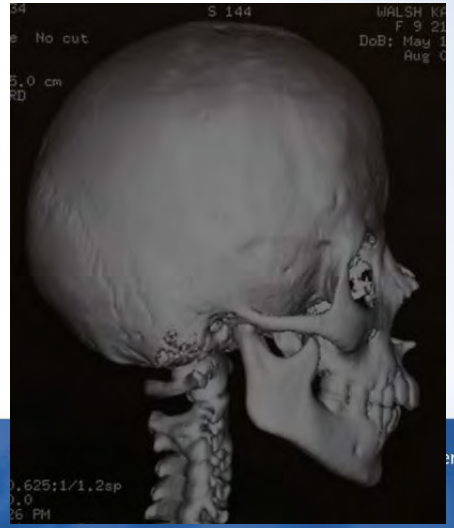


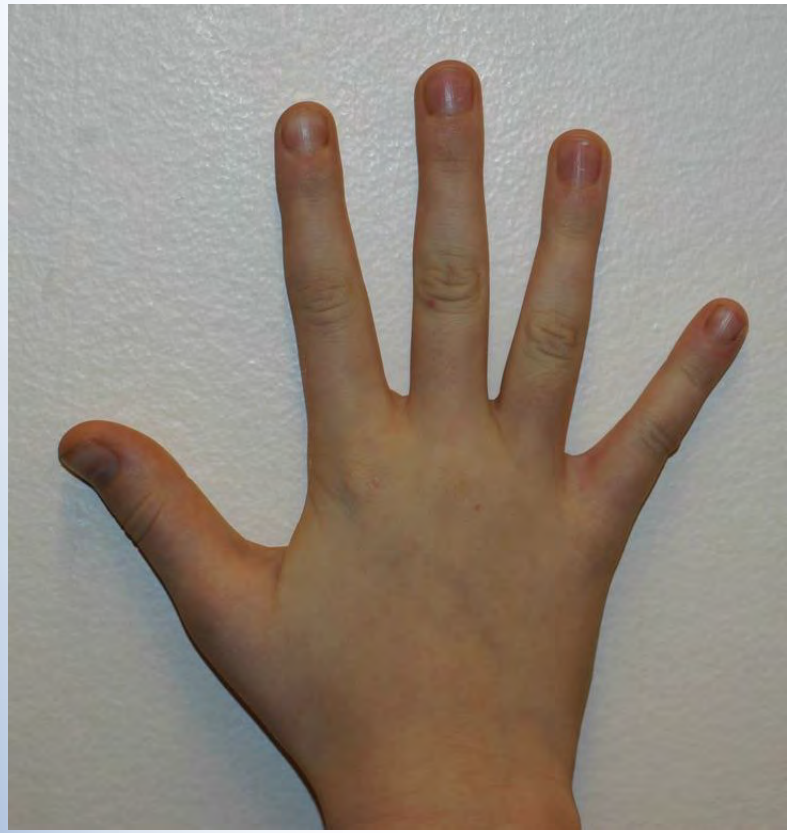
Syndromic Craniosynostosis

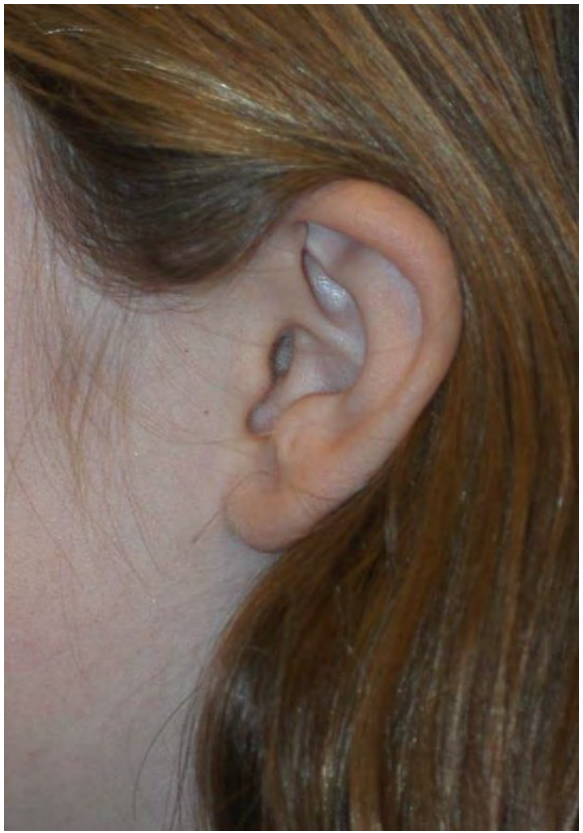


- Saethre-Chotzen syndrome (1931)
  - Autosomal dominant with high penetrance and variable expression
  - Normal intelligence
  - TWIST gene
  - Clinical features
    - Bicoronal synostosis
    - Ptosis
    - Facial asymmetry
    - Partial simple syndactyly
    - Ear anomaly







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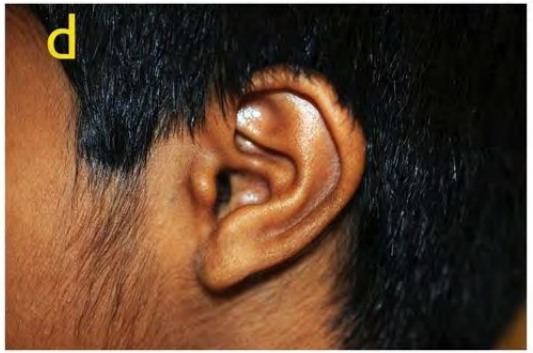
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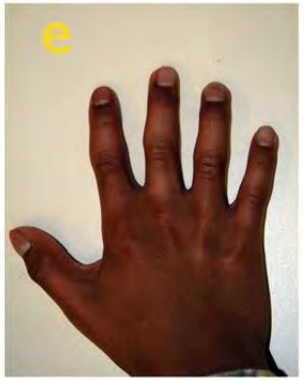
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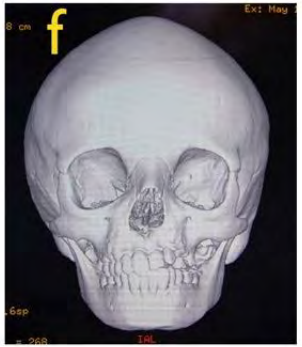
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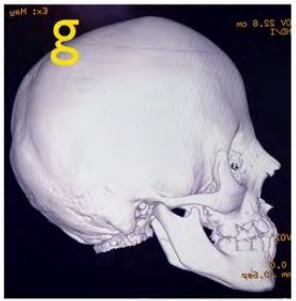
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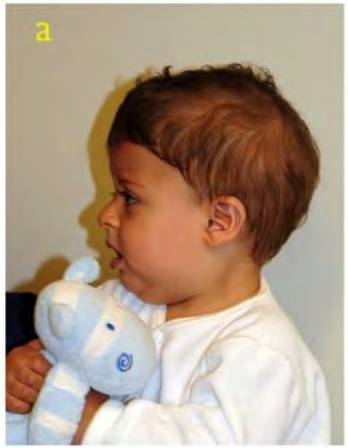
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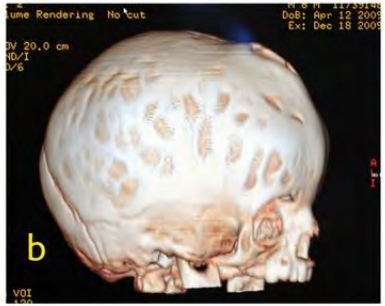
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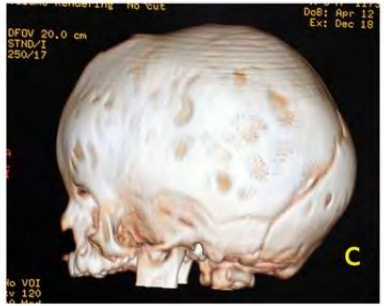
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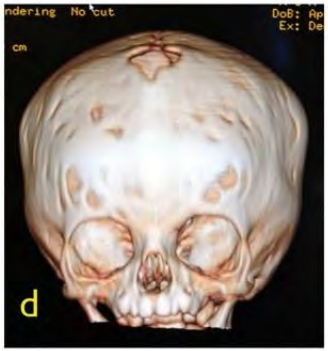
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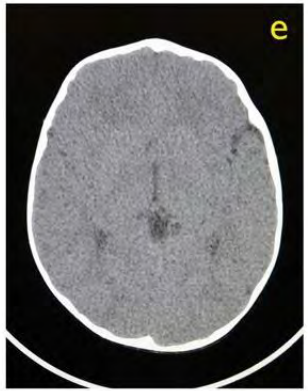
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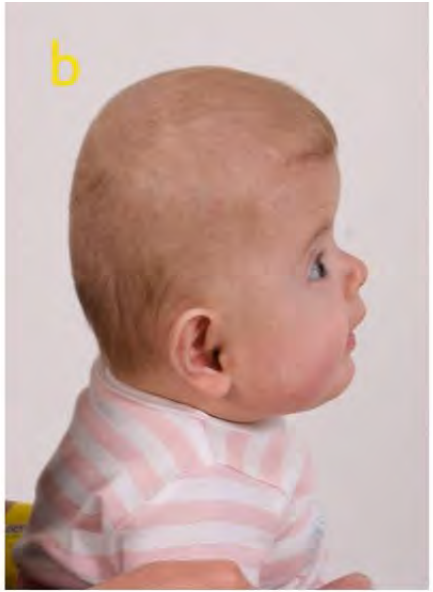
Muenke syndrome

- incidence variable, affects up to 20% of coronal synostosis patients, autosomal dominant
- refers to those patients with the proline 250 to arginine amino acid substitution in FGFR3 and has a highly variable phenotype .
  - uni or bicoronal craniosynostosis
  - brachydactyly, carpal and tarsal fusions
  - sensorineural hearing loss
  - Klippel-Feil cervical anomaly
  - variable degrees of mental impairment


a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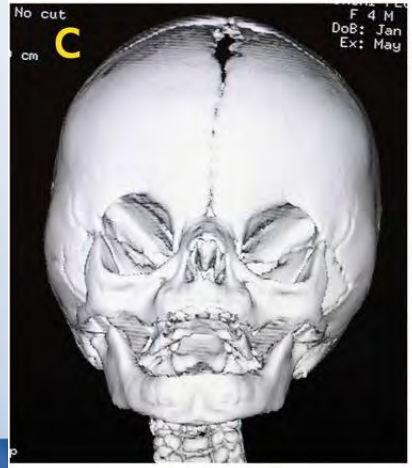
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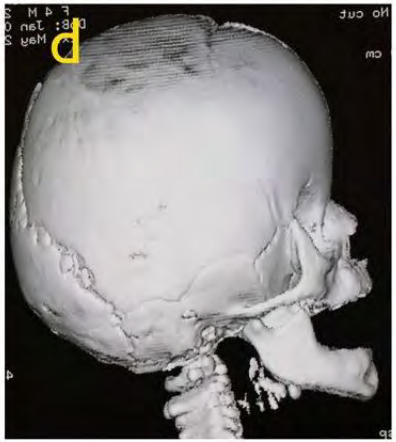
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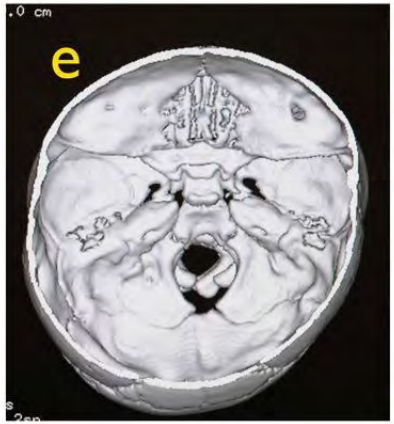
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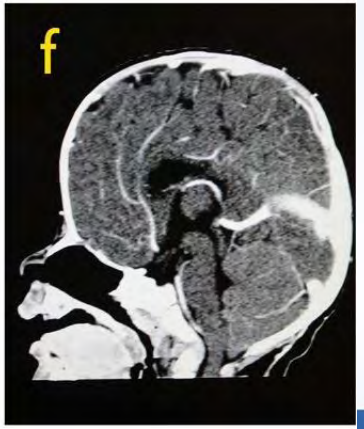
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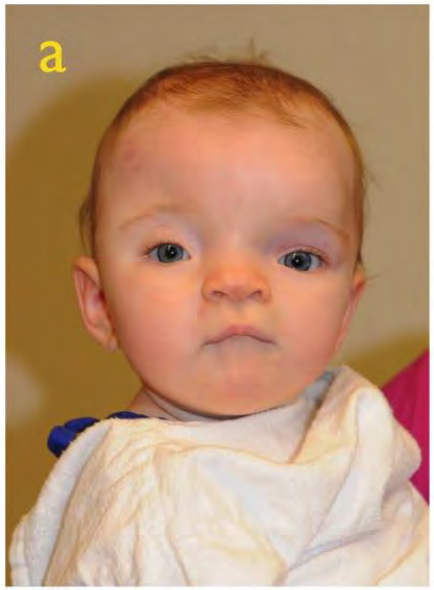
Craniofrontonasal dysplasia

- Rare, familial X-linked syndrome
- coronal synostosis (brachycephaly or plagiocephaly), hypertelorbitism (frequently asymmetric), and extracranial limb anomalies
- females predominating and males being less severely affected
- Mutations in the EFNB1 gene in Xq12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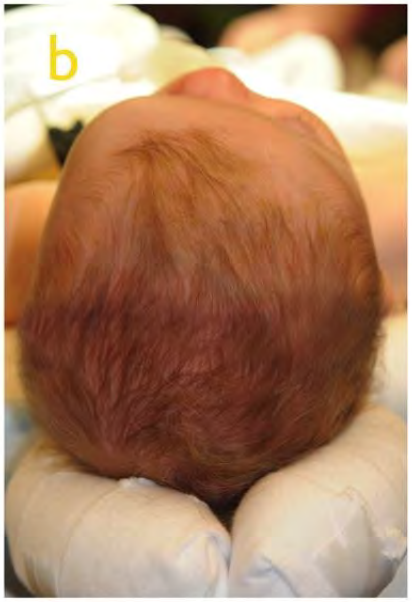
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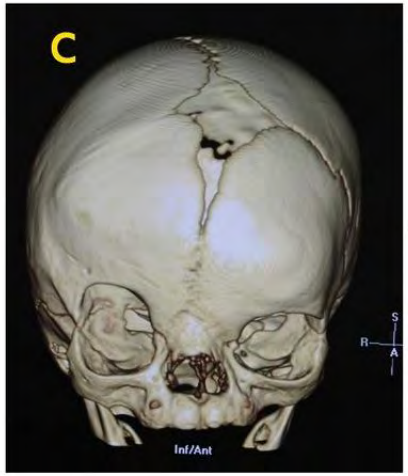
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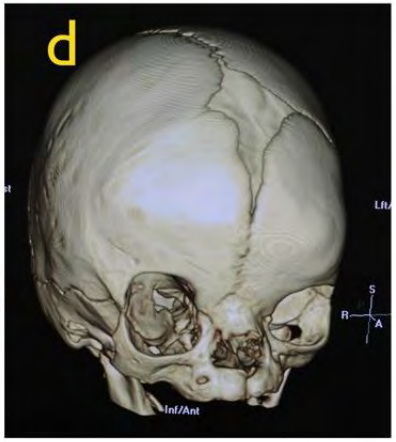
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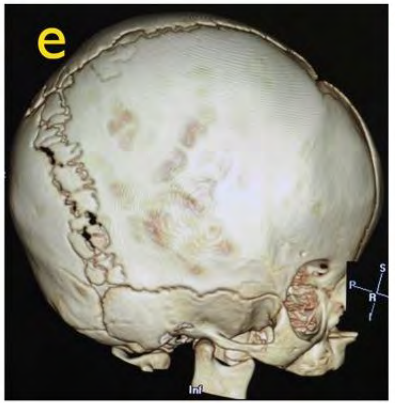
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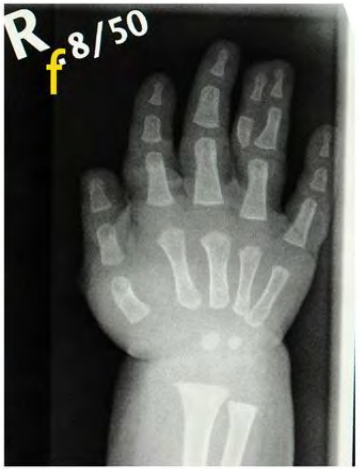
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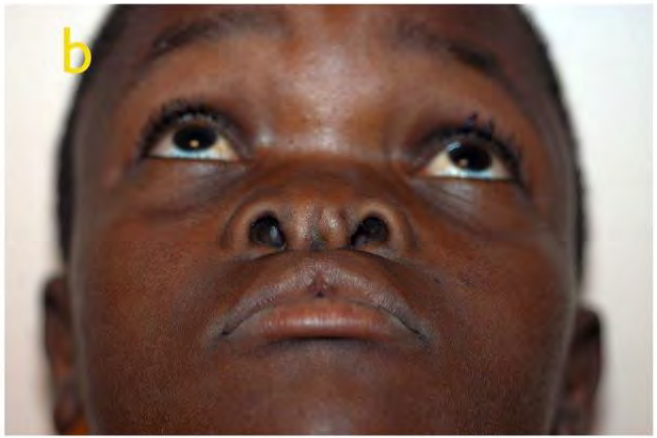
Frontonasal dysplasia

- median cleft syndrome, represents a variety of rare disorders affecting primarily the face and head
- causes of frontonasal dysplasia are unknown but recent evidence has pointed towards the role of genes encoding ciliary proteins on cranial neural crest cells and Hedgehog signaling
- Frontonasal dysplasia occurs twice as often in males as in females, and is associated with increased parental age
- Clinical features
  - hypertelorism
  - bifid nose
  - clefting of the lip and palate
  - anterior encephalocele
  - abnormal hairline
  - mild to moderate mental retardation


a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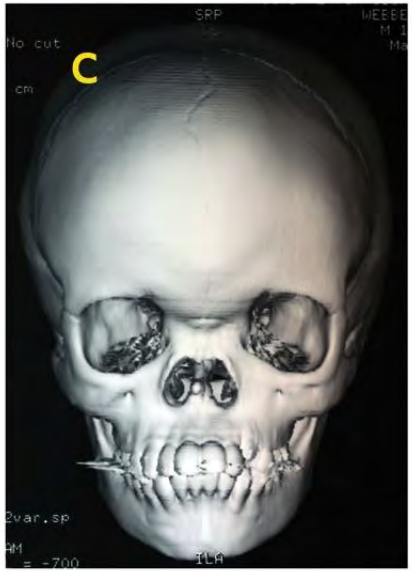
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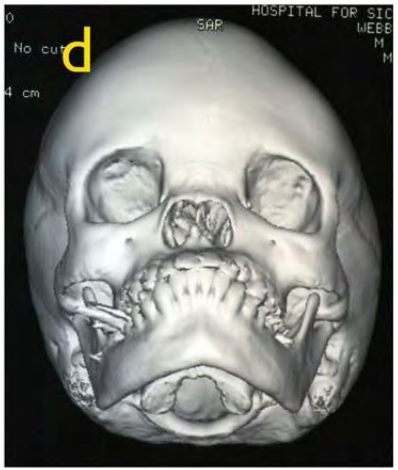
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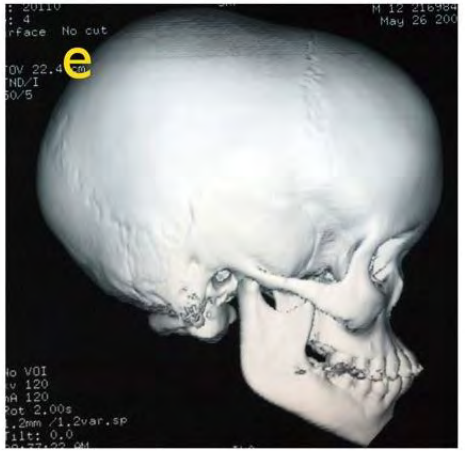
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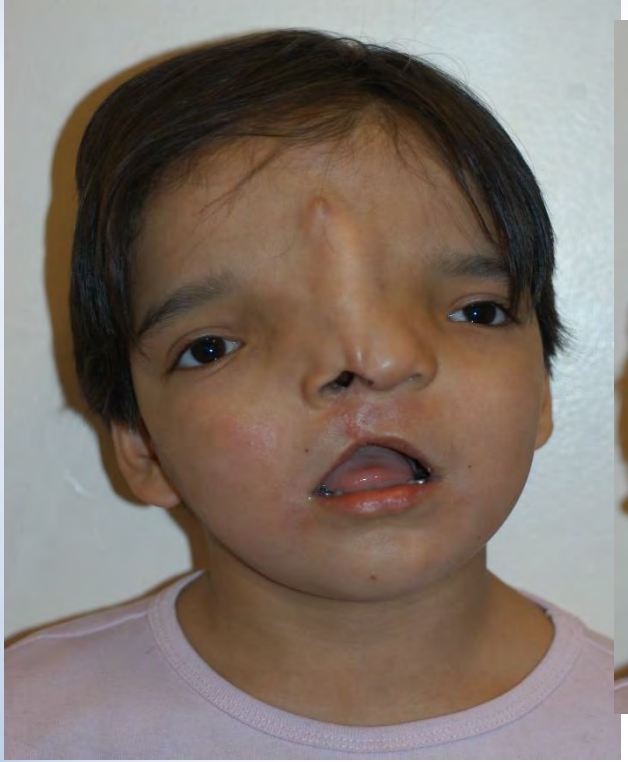

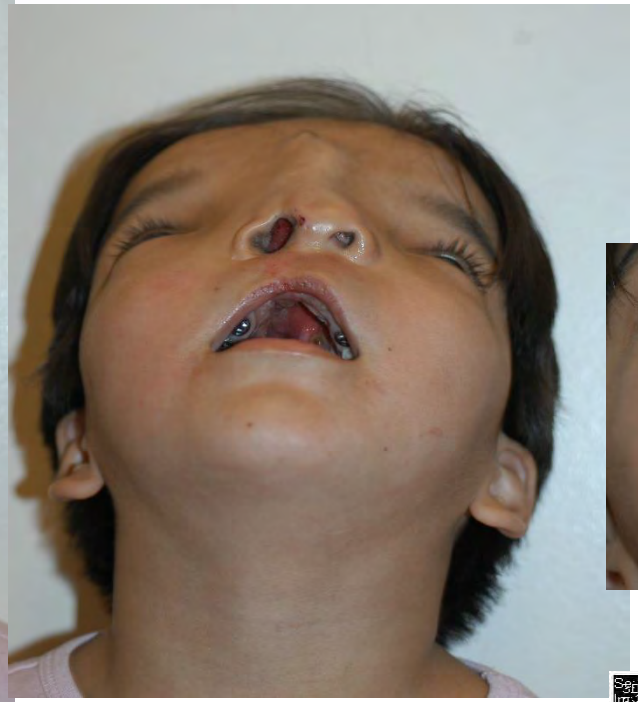

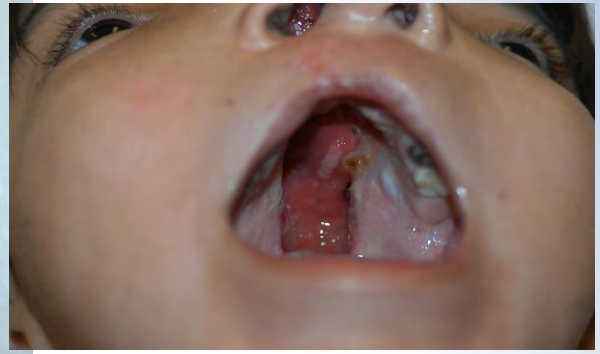

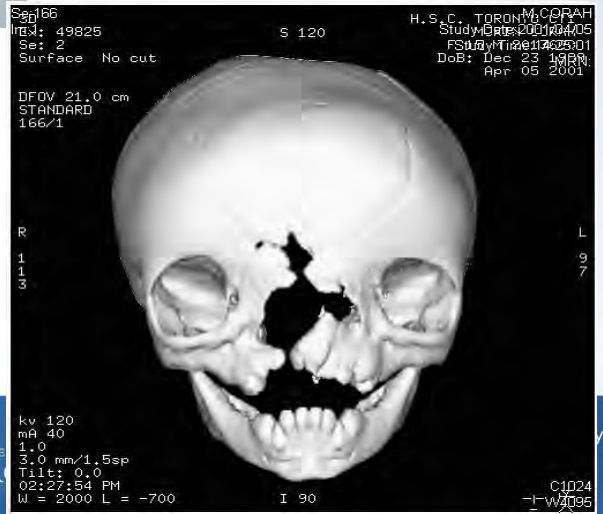
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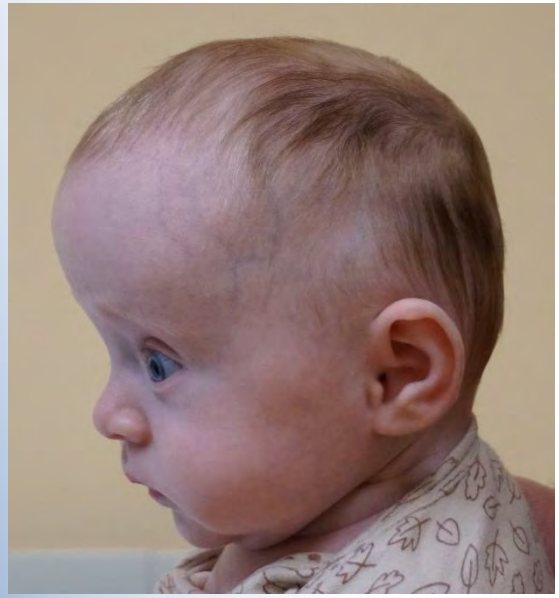
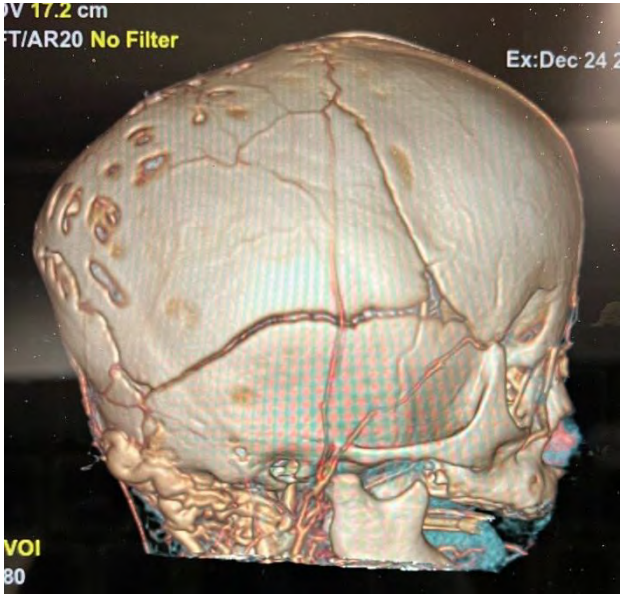
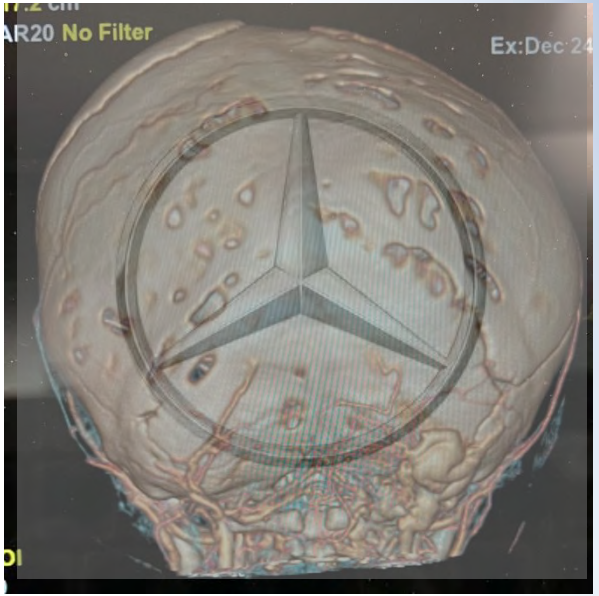
Differentiating Features of Common Syndromic Craniosynostosis

	Muenke Syndrome	Saethre-Chotzen Syndrome	Crouzon syndrome	Pfeiffer Syndrome	Apert Syndrome	Craniofrontonasal Dysplasia
Gene Mutation	FGFR3 (Pro250Arg)	TWIST1	FGFR2	FGFR1 FGFR2	FGFR2 (Ser252Trp or Pro253Arg)	EFNB1
Inheritance	Autosomal Dominant	Autosomal Dominant	Autosomal Dominant	Autosomal Dominant	Autosomal Dominant	X-linked Dominant
Incidence	1 in 10,000 to 1 in 30,000	1 in 25,000 to 1 in 50,000	1 in 65,000	1 in 100,000	1 in 70,000	1 in 100,000 to 1 in 120,000
Skull Morphology	Unicoronal or bicoronal synostosis	Unicoronal or bicoronal synostosis, metopic synostosis	Bicoronal synostosis, occasionally sagittal synostosis	Bicoronal synostosis, Kleeblattschädel (clover-leaf) with Type II	Bicoronal synostosis, large confluent anterior fontanelle	Unicoronal or bicoronal synostosis
Facial Features	Mild exorbitism, strabismus, thick straight hair, mild midface retrusion	Lid ptosis, low anterior hairline, midface hypoplasia, ear anomalies	Hypertelorism, exorbitism, midface retrusion	Hypertelorism, severe exorbitism, midface retrusion	Hypertelorism, small beak nose, exorbitism due downslanting palpebral fissures, narrow maxillary arch	Hypertelorism (asymmetric with unicoronal synostosis), broad nasal root, bifid nasal tip, occasional cleft lip/palate, dry frizzy hair
Limb Anomalies	Brachydactyly, symphalangism	Radioulnar synostosis, brachydactyly, simple syndactyly, hallux valgus with bifid distal phalanx	Usually none.	Broad thumbs and great toes, tarsal bone fusion, elbow ankylosis/synostosis, simple syndactyly.	Complex acrocephalosyndactyly involving hands and feet, tarsal fusions, radiohumeral fusion	Syndactyly, clinodactyly, grooved or split nails
Neurocognitive Impairment	Intellectual disability and/or developmental delay, autism, ADHD, low-frequency sensorineural hearing loss	Neuro development is usually normal, large genomic deletion may result in increased risk for intellectual challenges	Intellect normal. Conductive hearing loss	Intellect normal I Type I. Intellectual impairment and neurodevelopmental delay in Type II and III. Conductive hearing loss	Severe intellectual disability with neurodevelopmental delay, conductive hearing loss	Normal intellect in > 50%, 10% to 50% with developmental delay, mild learning difficulties, sensorineural hearing loss

Multi-suture synostosis

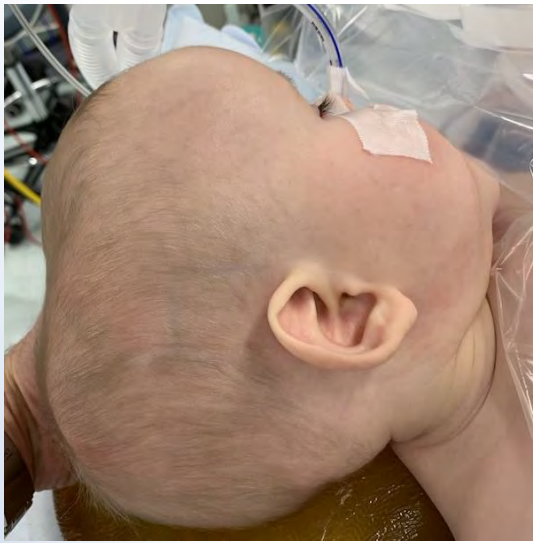


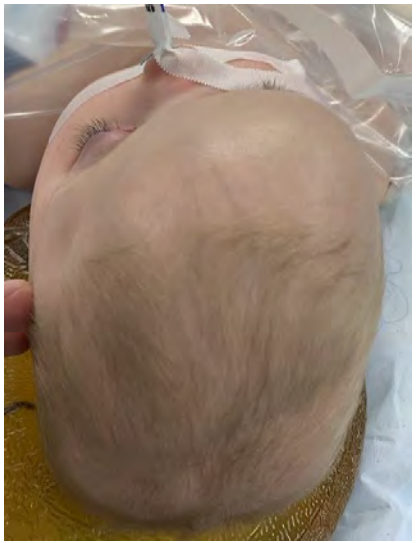


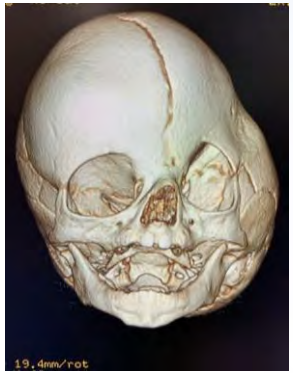


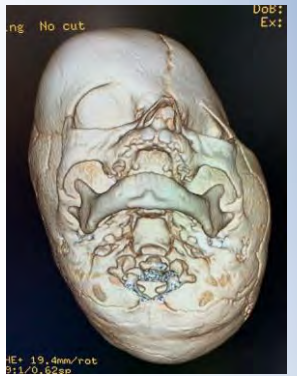
Mercedes Benz Synostosis

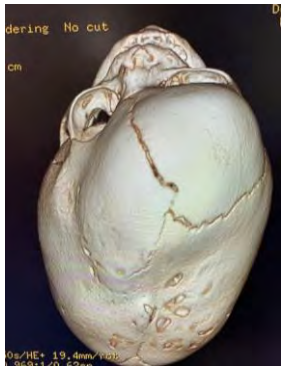
Multi-suture synostosis

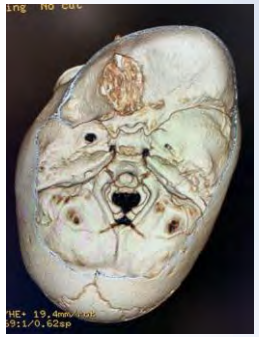


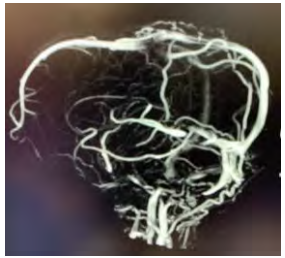


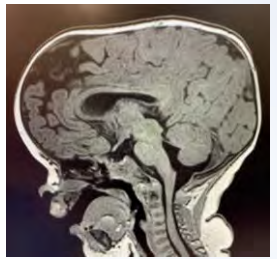












Sagittal and Left Uni-Coronal Synostosis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