myFace

Transforming Lives: Diagnosis and Management of Syndromic Craniosynostosis

myFace is pleased to collaborate with the American Cleft Palate Craniofacial Association (ACPA) to present this educational program

ACPA
American Cleft Palate Craniofacial Association

Generous funding for this program provided by The Milbank Foundation

Monday, February 13, 2023

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Welcome and Introductions

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Executive Director
myFace

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Presentation

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Associate Dean of Surgery and Hospital Affairs, Oral and Maxillofacial Surgery
Ostrow School of Dentistry of USC
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Children's Hospital Los Angeles (CHLA)
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Disclosures

- **Mark Urata, MD, DDS**, no disclosures

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Syndromic Craniosynostosis

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Syndromic Craniosynostosis

- Occur with other anomalies or birth defects in well-defined patterns that make up clinically recognized syndromes.
- Typically, more complex and require a multidisciplinary approach.
- Most commonly, bicoronal synostosis:

• Crouzon Syndrome	FGFR-2
• Apert Syndrome	FGFR-2
• Pfeiffer Syndrome	FGFR-2/1
• Saethre-Chotzen Syndrome	TWIST 1
• Muenke Syndrome	FGFR-3

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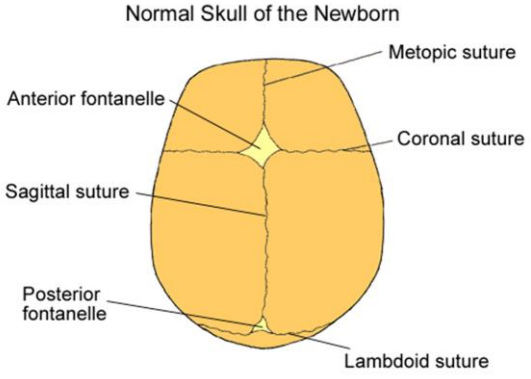
Bicoronal synostosis (Syndromic) Physical Findings

- Wide Skull (Brachycephalic)
- Tall Skull (Turricephalic)
- Palpebral fissure widened
- Supraorbital rim superiorly displaced
- Midface hypoplasia
- Orbits shallow
- Eyes proptotic
- Orbital hypertelorism



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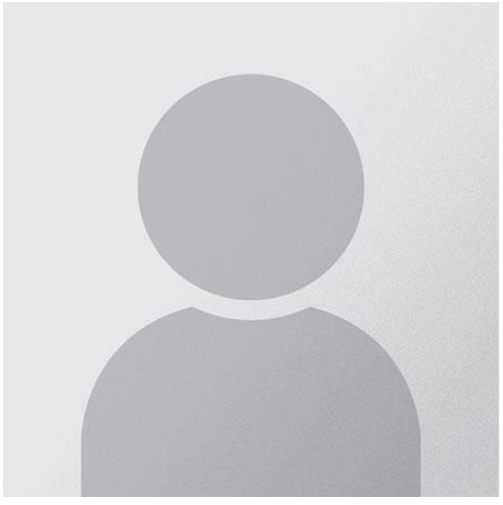

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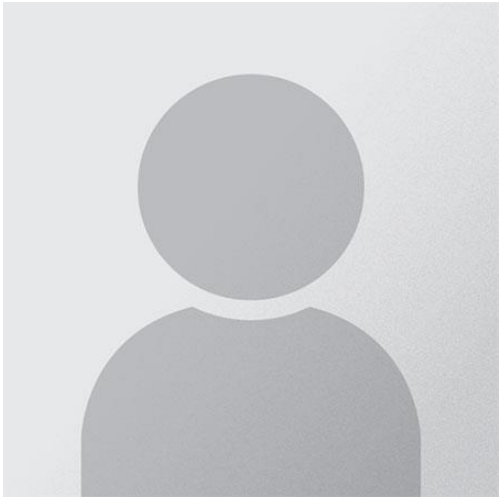

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Syndromic Craniosynostosis

- We operate for two reasons:
  - We want to ensure there is adequate room for the exponential growth of the brain (optic atrophy and blindness, developmental delay)
  - We want to achieve a naturalization of the appearance of the child
- Marathon (syndromic) versus Sprint (non-syndromic) – 20-year work of Art and Nature
- Focus on the Foundation Anticipating The Surgical Needs of the Future while visualizing the end result

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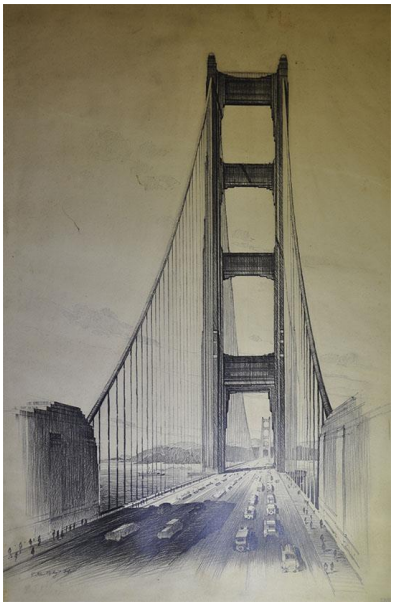

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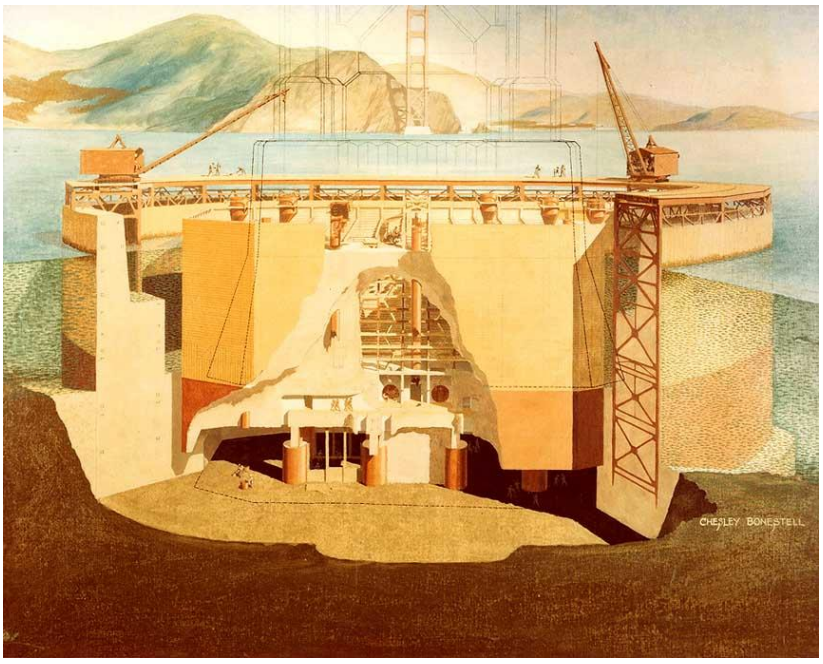

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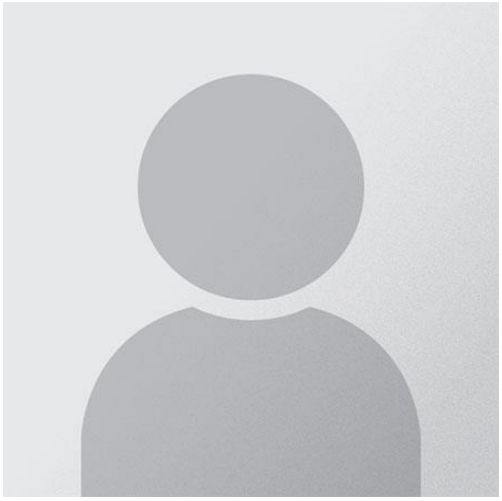

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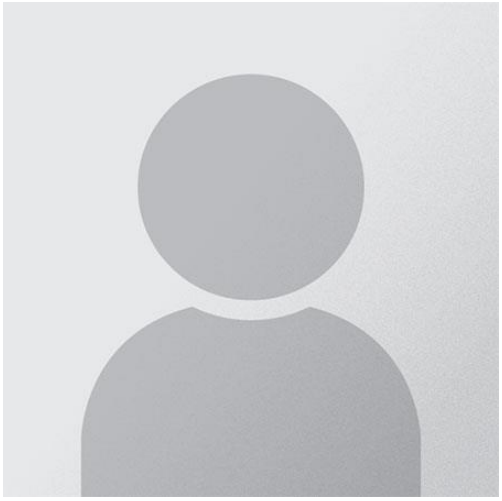

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Different Techniques

HEAD AND UPPER THIRD OF THE FACE
- FOA/CVR (Fronto-Orbital Advancement/Calvarial Vault Remodeling)
- Minimally invasive/endoscopic assisted suturectomy
- Posterior calvarial distraction
- Spring distraction

MIDFACE
- LeFort III advancement/distraction
- LeFort II advancement/distraction
- LeFort I advancement/distraction

LOWER THIRD OF THE FACE
- Mandibular distraction
- LeFort III/I/BSSO/genioplasty combos

EYE SOCKETS
- Box osteotomies
- Facial bipartition
- Monobloc advancement/distraction

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Factors that Must Be Considered

- Variations in those techniques
- Relapse
- Overcorrection
- Stability
- Psychosocial implications
- Risk of anesthesia
- Complications
- Skeletal maturity
- Teeth eruption
- Truth Telling

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Craniosynostosis Team

- Plastic Surgeon
- Social Worker
- Pediatrician (2)
- Craniofacial RN (2)
- Geneticist
- Psychologist
- Neurosurgeon
- Ophthalmologist

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Current Protocol

- CT scan fine cuts with 3D reconstruction
  - Evaluate skull shape
  - Look for Chiari Malformation
    - 70% Crouzon
    - 82% Pfeiffer
    - 100% Kleeblattschaedel
- Digital 3D Imaging (Mirror Software) (3D MD)
- Directed donor for blood transfusion

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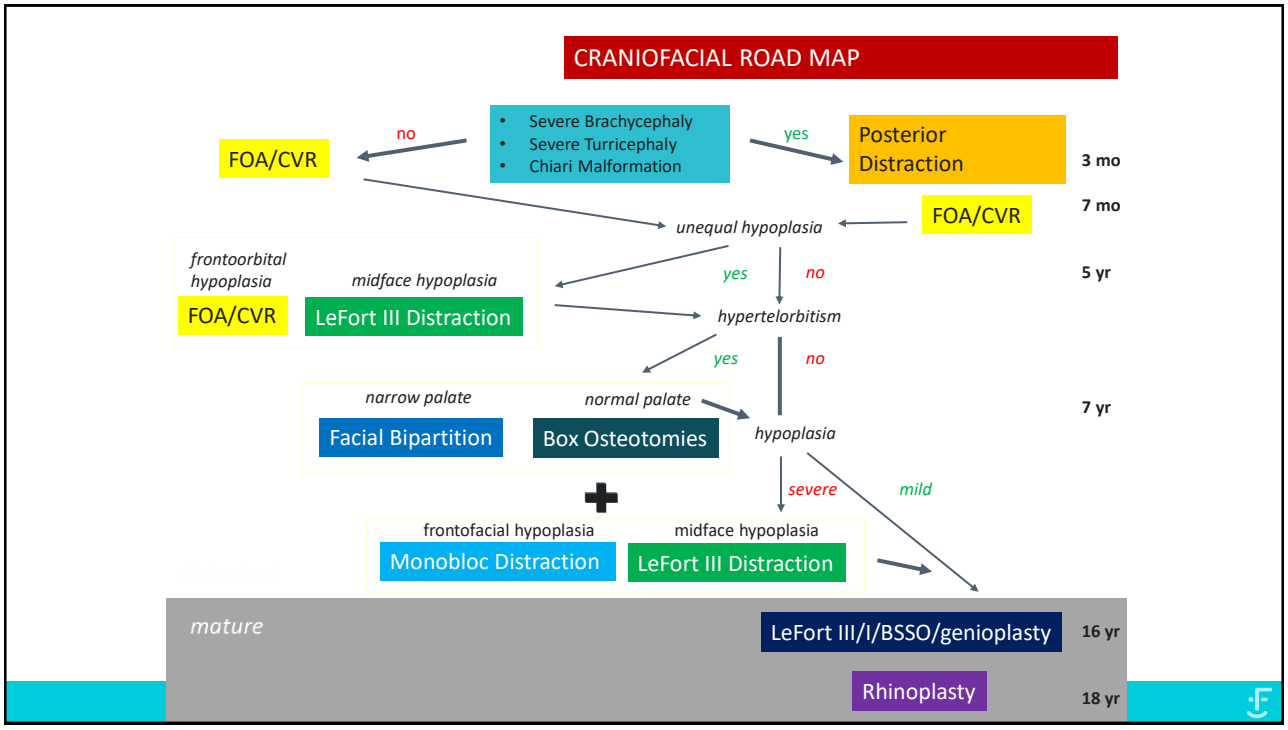
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Severe Turricephaly
Chiari Malformation
no
yes
FOA/CVR
Posterior Distraction
3 mo
7 mo
FOA/CVR
unequal hypoplasia
frontoorbital hypoplasia
midface hypoplasia
yes
no
5 yr
FOA/CVR
LeFort III Distraction
hypertelorbitism
yes
no
narrow palate
normal palate
7 yr
Facial Bipartition
Box Osteotomies
hypoplasia
severe
mild
frontofacial hypoplasia
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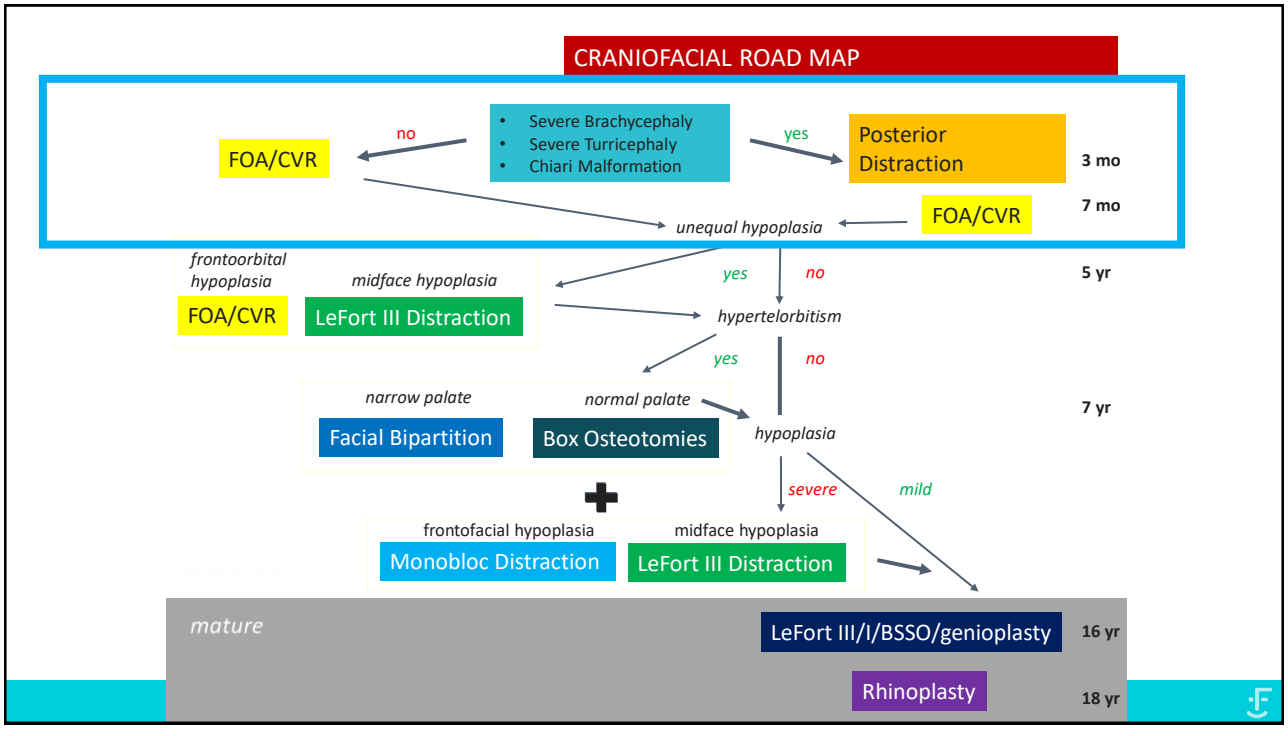
Monobloc Distraction
LeFort III Distraction
mature
LeFort III/I/BSSO/genioplasty
16 yr
Rhinoplasty
18 yr

23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CRANIOFACIAL ROAD MAP

FOA/CVR

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- Severe Turricephaly
- Chiari Malformation

yes

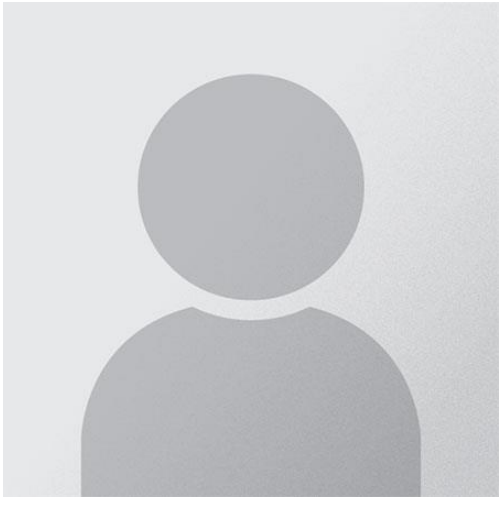
Posterior Distraction

FOA/CVR

3 mo

7 mo

25



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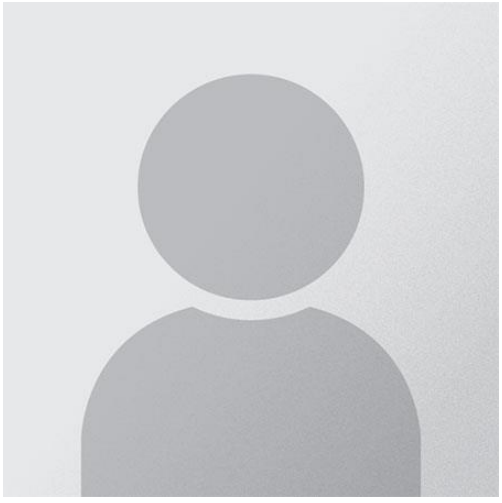

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CRANIOFACIAL ROAD MAP

FOA/CVR

no

- Severe Brachycephaly
- Severe Turricephaly
- Chiari Malformation

yes

Posterior Distraction

FOA/CVR

3 mo

7 mo

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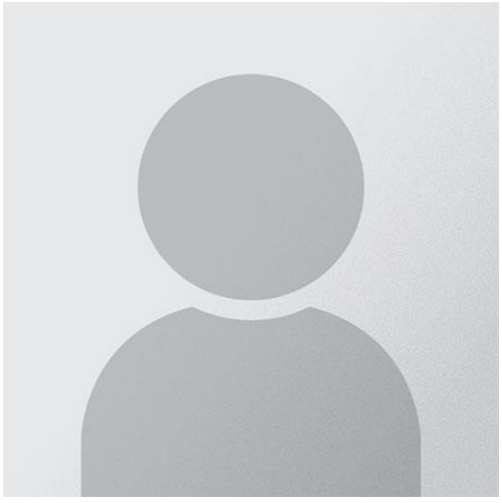

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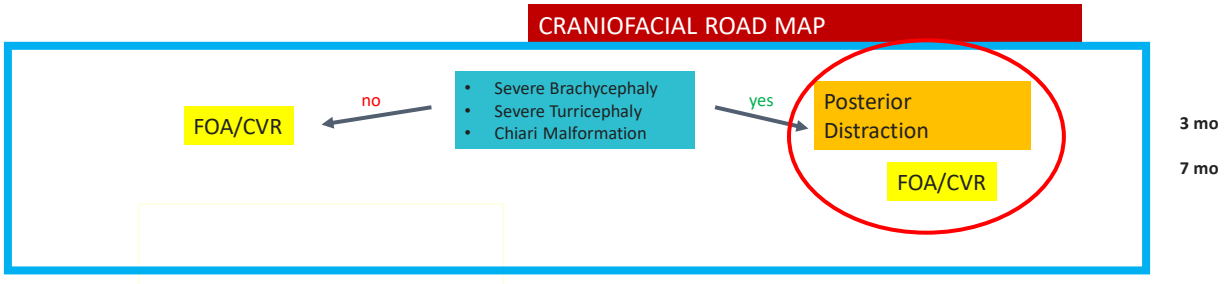

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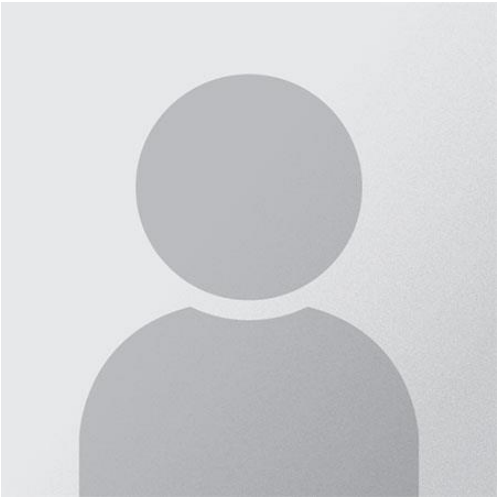

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Posterior Vault Expansion (2-3 months of age)

- Distractors (slow stretch over 2-3 weeks)
- Calvarial vault remodeling
- Spring (slow stretch over several weeks)

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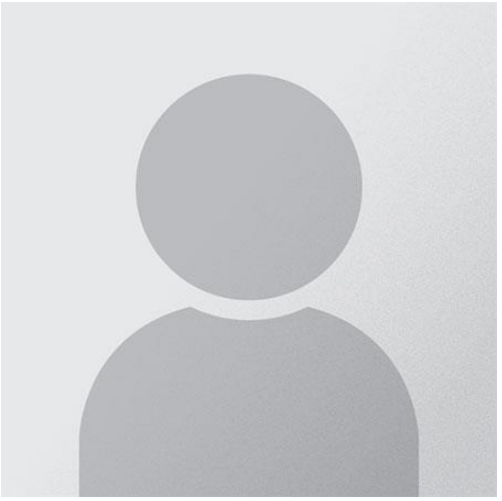

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Fronto-orbital Advancement
Calvarial Vault Remodeling
(7-12 months of age)

- Remove posterior distractors
- Move forehead forward
- Decrease Height
- Dissolvable screws and plates

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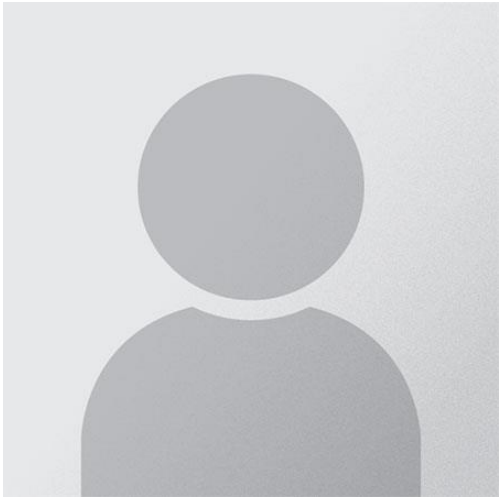

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Post-Operative Care

- 1 night ICU
- 2-3 nights on floor
- Discharge home:
  - Oral antibiotics
  - Pain medication
- Measurements for helmet 1 week after discharge
- Helmet delivery 1 week after that
- Helmet therapy for 6 months to 1 year post-operatively

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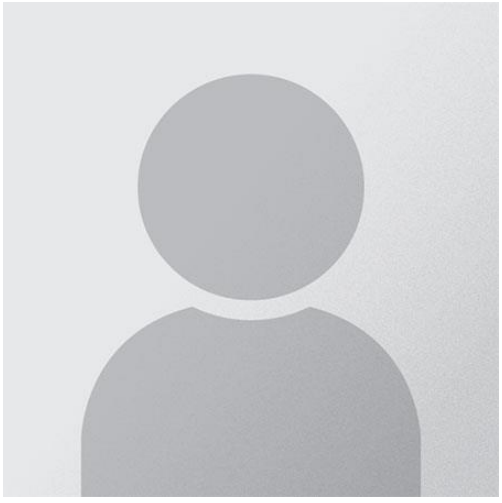

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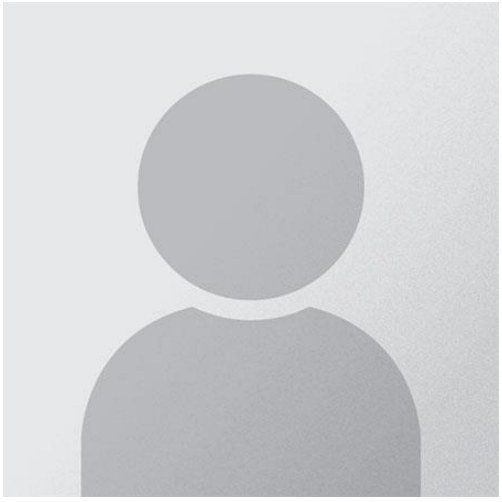

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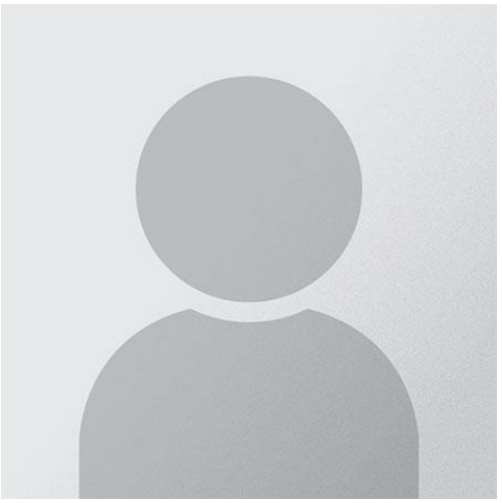

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Follow Up Appointments

• 1 week after discharge	PS/NS
• 2 week after discharge	PS/NS
• 1 month after discharge	PS/NS
• 3 month after discharge	PS
• 6 month after discharge	PS
• 1 year after discharge	PS/NS
• Every year	PS/CF

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CROUZON SYNDROME

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CROUZON SYNDROME

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APERT SYNDROME

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APERT SYNDROME

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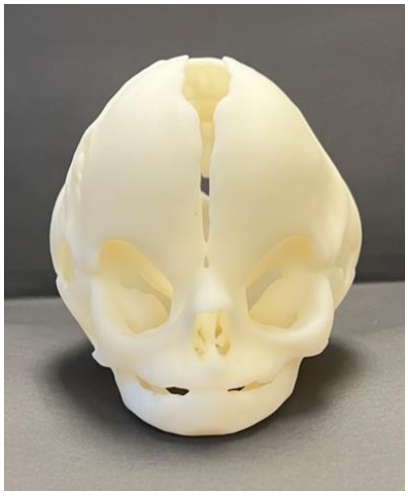
PFEIFFER SYNDROME



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PFEIFFER SYNDROME

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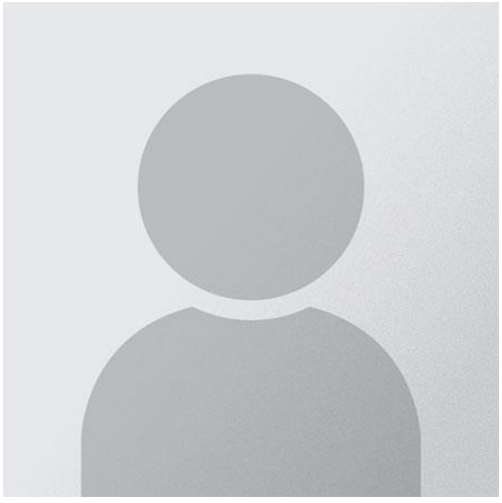

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unequal hypoplasia ← FOA/CVR — 7 mo

yes → *frontoorbital hypoplasia* / *midface hypoplasia* → FOA/CVR / LeFort III Distraction — 5 yr

no → *hypertelorbitism*

yes → *narrow palate* → Facial Bipartition

normal palate → Box Osteotomies — 7 yr

no → *hypoplasia*

severe → frontofacial hypoplasia → Monobloc Distraction

mild → midface hypoplasia → LeFort III Distraction

mature — LeFort III/I/BSSO/genioplasty — 16 yr

Rhinoplasty — 18 yr

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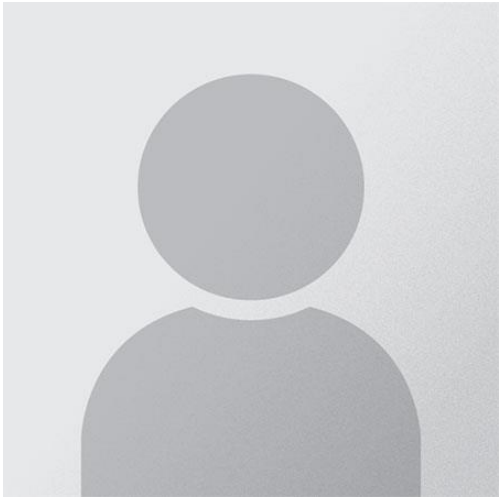

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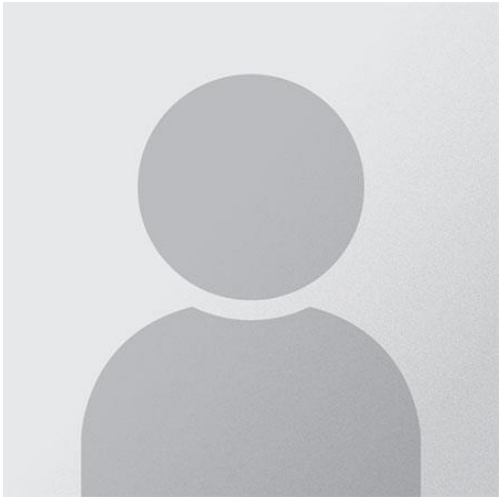

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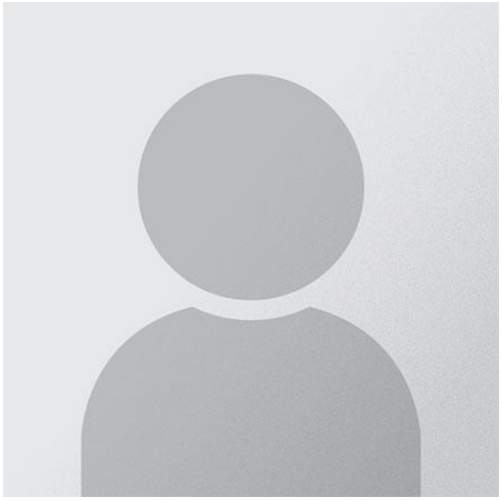

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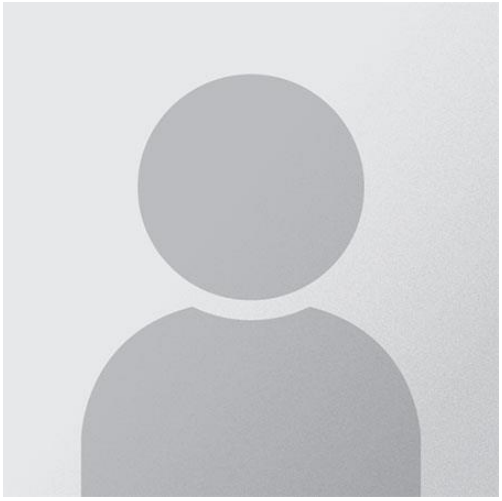

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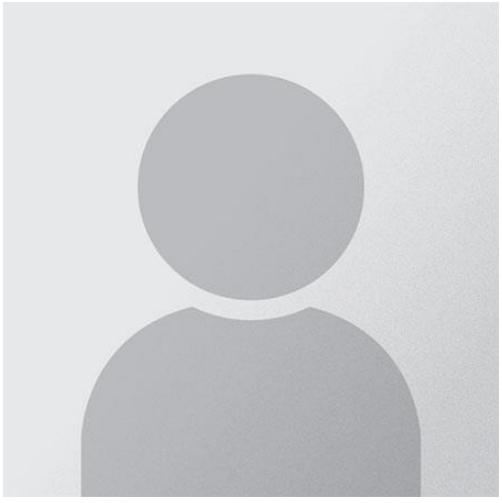

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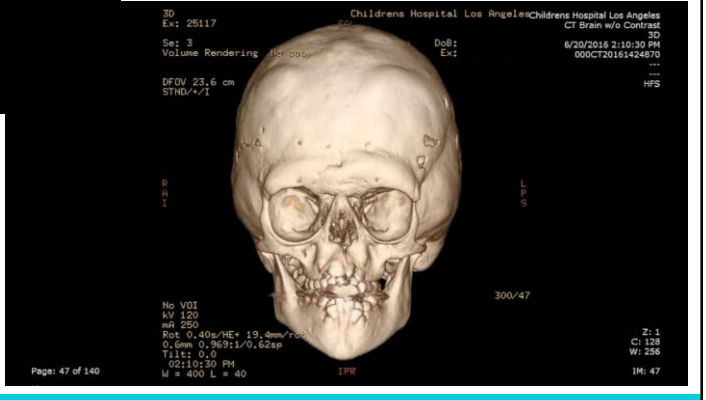

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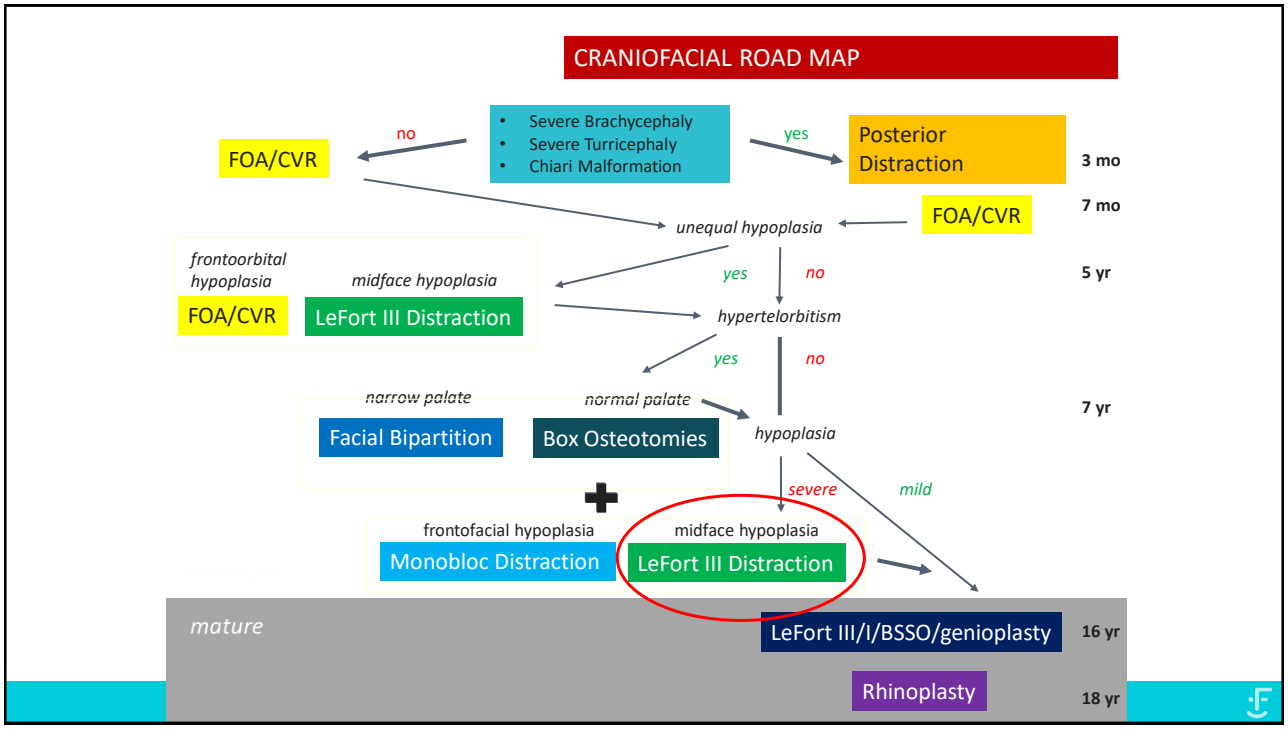

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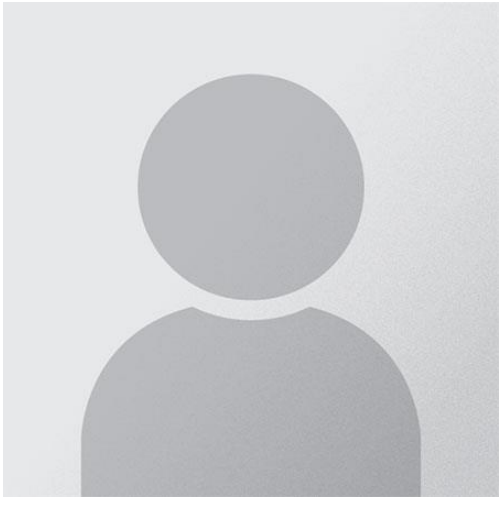

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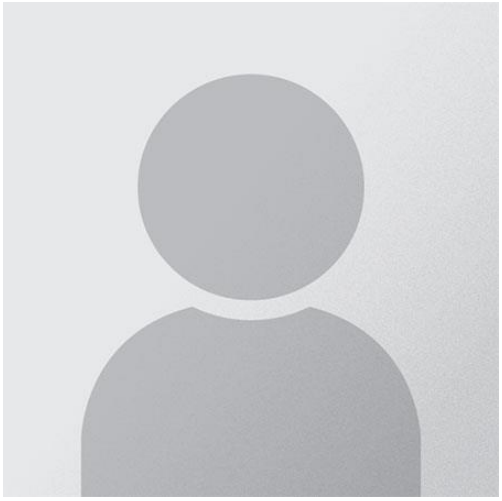

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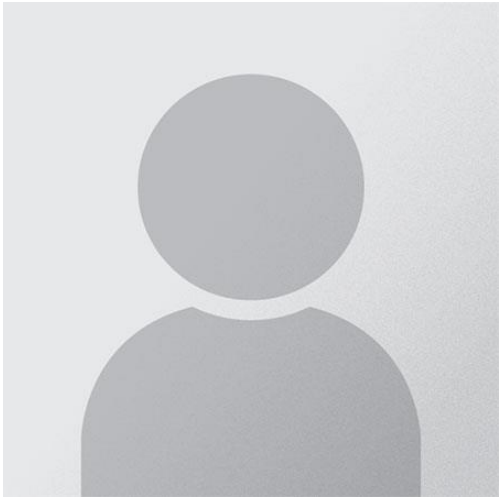

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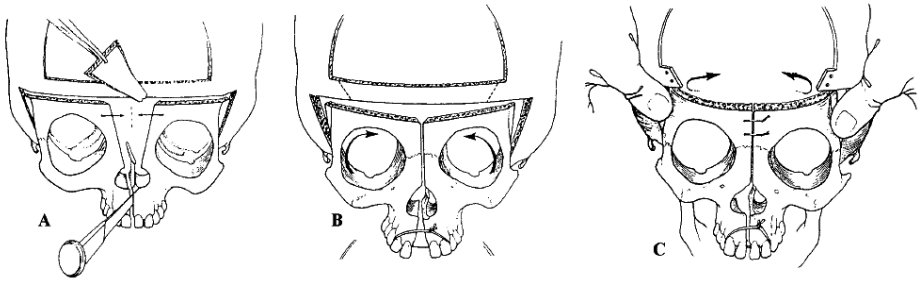
Monobloc Facial Bipartition



- Frontofacial deficiency
- Hypertelorbitism

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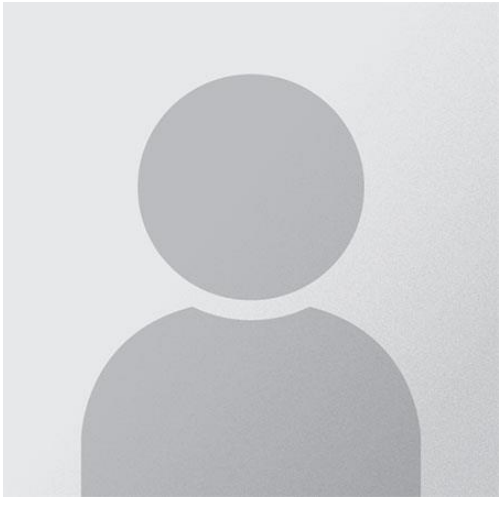

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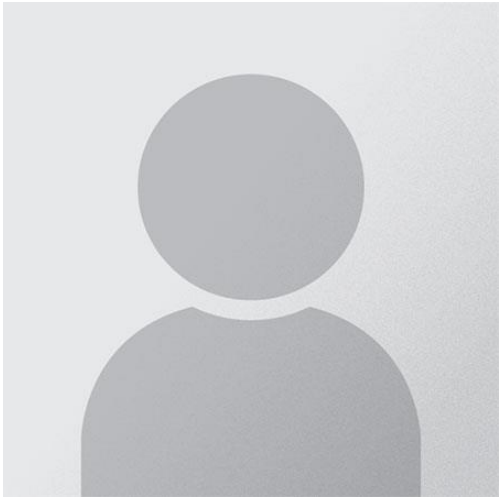

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hypertelorbitism

yes → narrow palate → Facial Bipartition

no → normal palate → Box Osteotomies — 7 yr

hypoplasia

severe → frontofacial hypoplasia → Monobloc Distraction

mild → midface hypoplasia → LeFort III Distraction

mature

LeFort III/I/BSSO/genioplasty — 16 yr

Rhinoplasty — 18 yr

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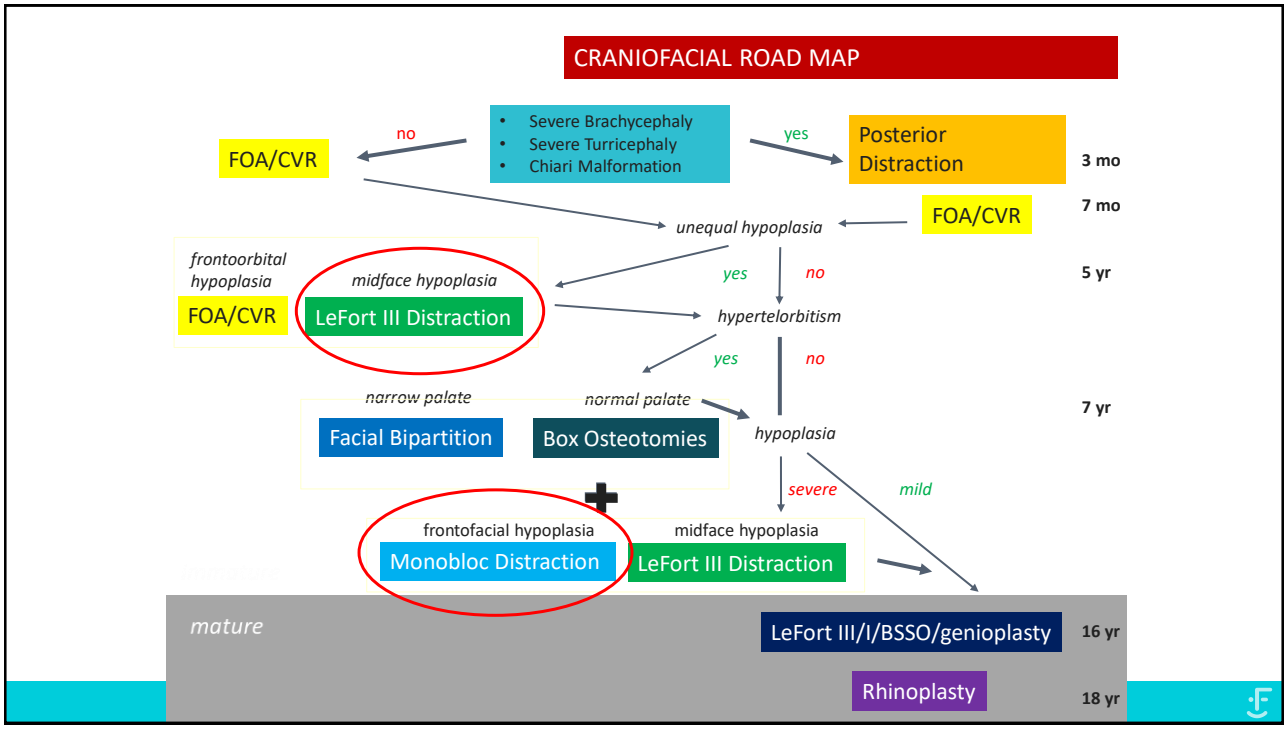

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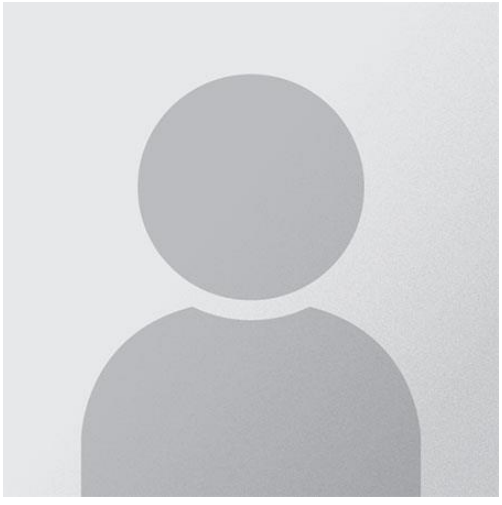

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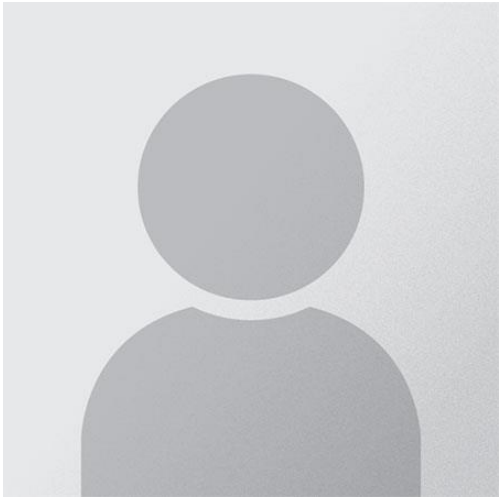

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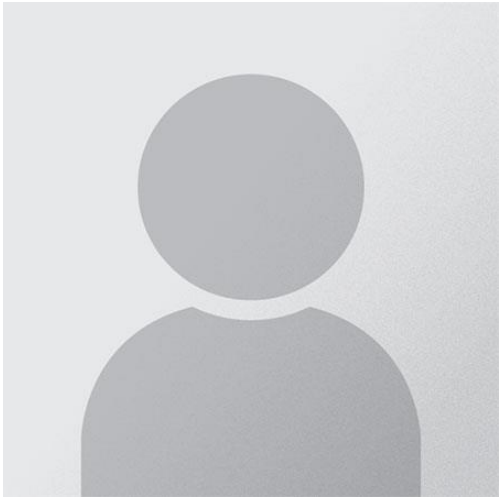

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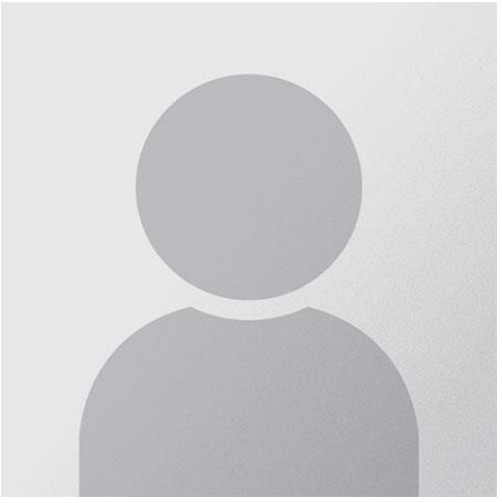

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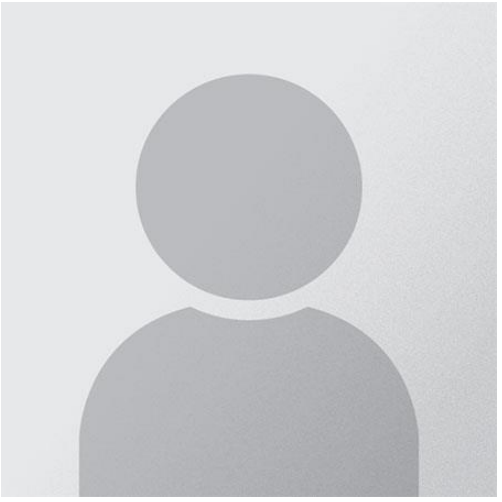

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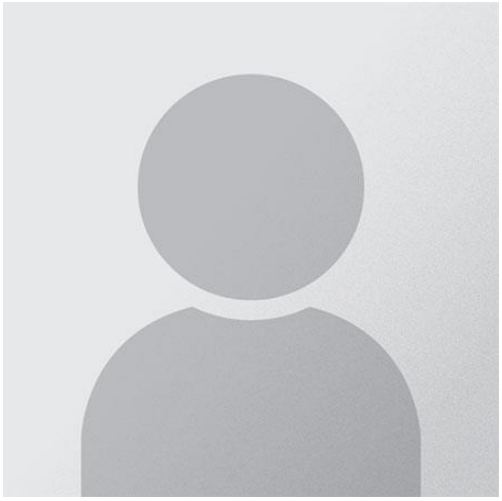

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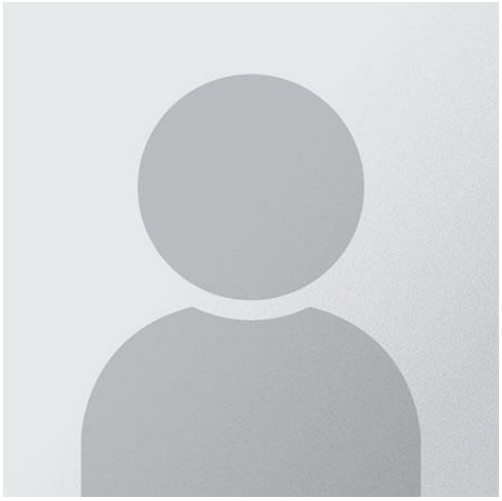

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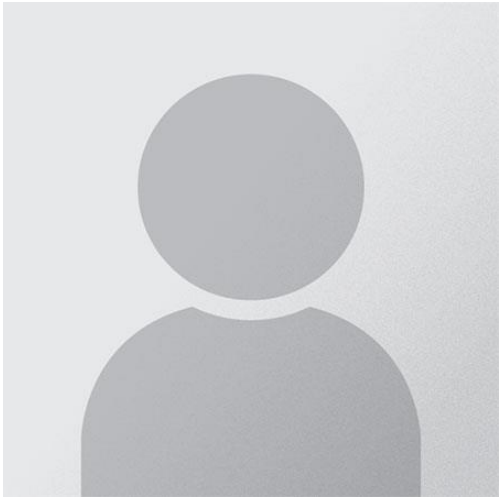

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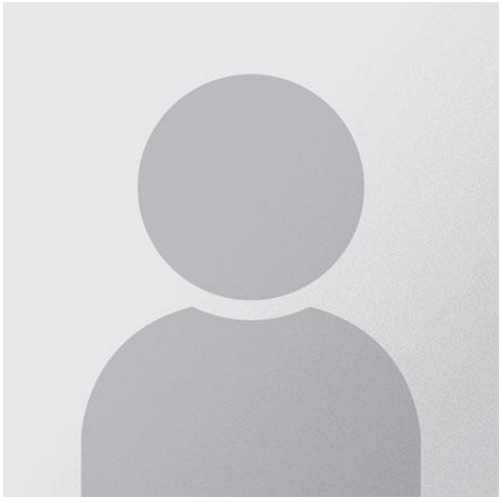

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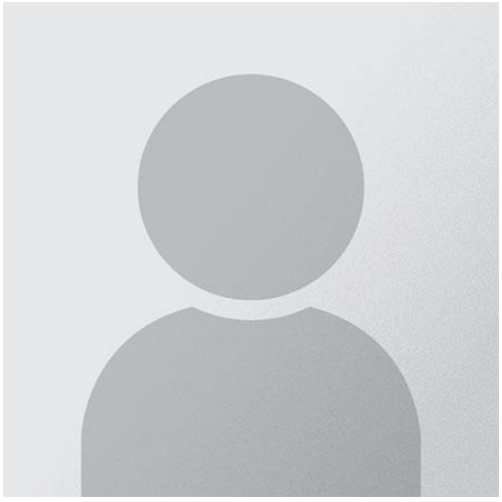

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CRANIOFACIAL ROAD MAP

- Severe Brachycephaly
- Severe Turricephaly
- Chiari Malformation

no → FOA/CVR

yes → Posterior Distraction — 3 mo

unequal hypoplasia ← FOA/CVR — 7 mo

frontoorbital hypoplasia / midface hypoplasia
FOA/CVR | LeFort III Distraction — 5 yr

hypertelorbitism
yes → frontoorbital hypoplasia / midface hypoplasia
no → hypoplasia

narrow palate / normal palate
Facial Bipartition | Box Osteotomies — 7 yr

hypoplasia
severe → frontofacial hypoplasia / midface hypoplasia
Monobloc Distraction | LeFort III Distraction
mild → mature

mature
LeFort III/I/BSSO/genioplasty — 16 yr
Rhinoplasty — 18 yr

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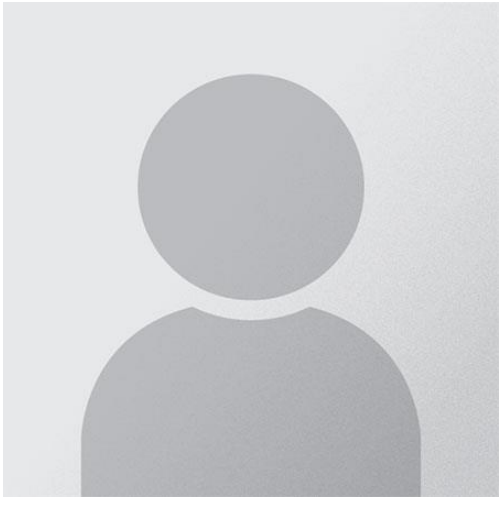

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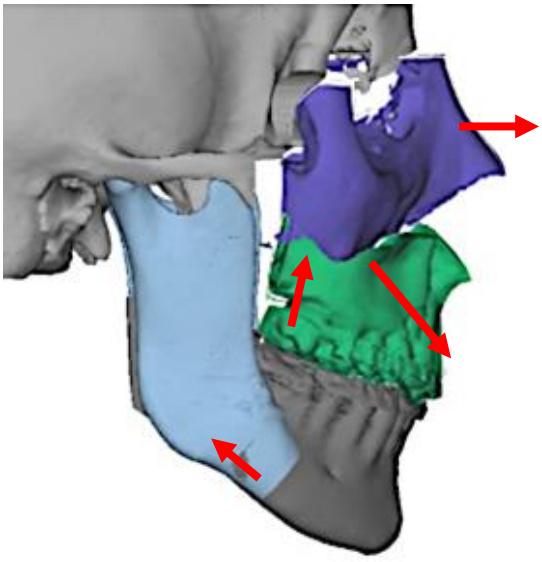

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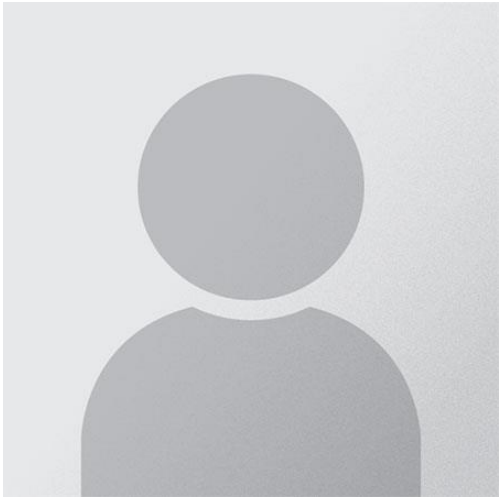

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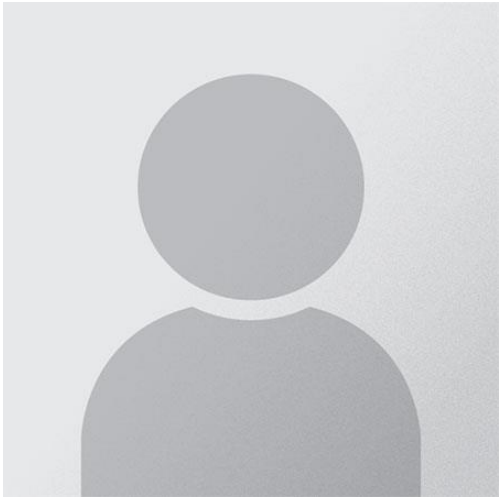

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Post-operative occlusion in orthodontic range of correction

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Our Masterpiece

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Presentation

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Division Head; Chief, Division of Plastic and Maxillofacial Surgery
Children's Hospital Los Angeles (CHLA)
Audrey Skirball Kenis Endowed Chair
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Disclosures

- **Alessia Johns, PhD, ABPP**, no disclosures

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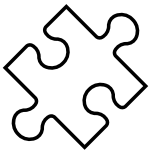
Neurodevelopmental Outcomes

- Broadly refers to overall functioning
- Focus on cognitive or intellectual functioning (IQ)
- Other important areas are generally separate, such as:
  - academic achievement
  - adaptive functioning
  - psychosocial adjustment
- Health-related quality of life can also be helpful to look at global functioning



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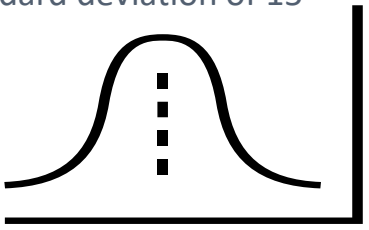
Assessment

- Early Development Assessment
  - Cognitive Functioning
  - Receptive Language
  - Expressive Language
  - Fine Motor Skills
  - Gross Motor Skills
- Child to Adult Assessment
  - Full Scale IQ
  - Verbal Comprehension
  - Visual Spatial/Perceptual Reasoning
  - Fluid Reasoning
  - Working Memory
  - Processing Speed

- Standardized tests developed with reliability and validity processes
- Raw scores have meaning *only* in comparison to the norm sample
- Standard scores fit within a normal curve with a mean of 100 and standard deviation of 15



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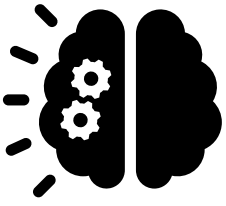
Environmental Factors Impact Development

- Prenatal Risk Factors
- Premature Birth
- Family Socioeconomic Status
- Nutrition



- Environmental Toxins
- Traumatic Stress
- Developmental Stimulation
- Caregiver-Child Relationship

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Possible Syndromic Craniosynostosis-Related Health Factors Impact Development

- Vision concerns
- Hearing loss
- Syndactyly
- Airway Obstruction
- Obstructive Sleep Apnea
- Cleft Palate
- Genetic Variations
- Hydrocephalus
- Structural Brain Differences

e.g., de Jong et al., 2010; Wang et al., 2016

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Increased Intracranial Pressure (ICP) Example

= 45 (19 syndromic)[a]

- 68% had elevated ICP with direct measurement using a fiberoptic probe
- Only 4% had papilledema on eye exam, which is used as a marker of ICP
- 96% had decreased ICP after surgery
- No differences by surgery type
- Children younger than 12 months were less likely to have elevated ICP

[a]Judy et al., 2018

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Increased Intracranial Pressure (ICP) Examples

= 24[b]

- 83% of patients with Apert syndrome had elevated ICP pre-treatment with average onset at age 18 months
- Of those patients, 35% had a recurrence of ICP 3.4 years post-treatment
- A third ICP recurrence was seen in 15%

= 49[b]

- 61% of patients with Crouzon syndrome had elevated ICP at an average onset at age 1.4 years
- Of those patients, 47% had a recurrence of ICP 1.4 years post-treatment
- A third ICP recurrence was seen in 10% 3.2 years post-treatment

[a]Marucchi et al., 2008; [b]Abu-Sittah et al., 2016

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Limitations in Our Knowledge

- Low incidence of syndromes translates to small sample sizes in research, which leads to multiple barriers in interpreting results.
- Many different types of measures are used over time and globally that are then difficult to compare and pool findings meaningfully.
- Contextual factors likely to impact cognitive development are often not accounted for in research.
- More recent genetic variations linked to syndromes with variable outcomes are not consistently available or described across studies.

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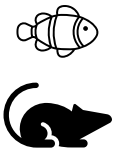
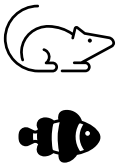
Examples of Advances through Animal Models



- With a Crouzon syndrome with acanthosis nigricans model, Cornille et al. (2022) identified and reversed a pathway of memory impairment with alterations in the hippocampus rather than in skull formation.



- Using a Saethre-Chotzen syndrome model, Yu et al. (2021) confirmed and then reversed increased intracranial pressure and neurocognitive anomalies.

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Syndromic Craniosynostosis as a Group

= 76[a]

- Total group average IQ was 97±22
- 30% had an IQ < 85
- 30% had ADHD

= 101[b]

- 23% had Intellectual Disability, which was 15.7 times more likely compared to 876 general population peers
- 4 times more likely than peers to have Autism Spectrum Disorder
- Diagnoses were generally made at 3 to 4 years

[a]Maliepaard et al., 2O14; [b]Junaid et al., 2022

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Syndromic Craniosynostosis Quality of Life

= 87-110 parents; 29 adolescents[a]

- Parents rated their children's quality of life lower than general population norms in 80% of areas, except for "change in health" and "family cohesion"
- Parents with lower socioeconomic status reported lower psychosocial quality of life in their children
- Adolescents were no different than general population norms in self-reported quality of life, except for lower "general health" and "family cohesion"
- Parents rated their own health-related quality of life lower for 67% of areas, including their own "general health" perceptions

[a]Bannink et al., 2010

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Syndromic Craniosynostosis Quality of Life

= 20[a]

- Individuals with Apert and Crouzon syndrome had no areas of quality of life in the below average range
- "Good" range for quality of life in 88% of areas, including:
  - self-esteem
  - body image
  - personal relationships
  - social support
  - ability to work
  - thinking, learning, and concentration

= 40[b]

- Higher quality of life ratings than the UK population for:
  - Physical
  - Psychological
  - Environmental
- No different for social quality of life than UK population
- No relationship between quality of life and MD ratings of appearance

[a]Raposo-Amaral et al., 2014; [b]Lloyd et al., 2016

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Muenke Syndrome (1:30,000)

 = 13[a]

- Group average IQ was 95±16
- 39% had an IQ < 85

 = 25 parents[b]

- Greatest areas of parental concern in health-related quality of life were:
  - 44% speech
  - 40% vision
  - 29% emotion

 = 4[c]

- 25% average range (IQ > 90)
- 50% well below average range (IQ 70-80)
- 25% had Intellectual Disability (IQ < 70)
- 75% completed secondary school
  - two with special education support

[a]Maliepaard et al., 2014; [b]de Jong et al., 2012; [c]Flapper et al., 2009;

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Saethre-Chotzen Syndrome (1:25,000-50,000)

 = 14[a]

- Group average IQ was 100±27
- 21% had an IQ < 85

 = 18 parents[b]

- Greatest areas of parental concern in health-related quality of life were:
  - 69% vision
  - 29% speech

= 30[c]

- *TWIST1*-confirmed
- 80% had hearing loss (~ conductive)
- 17% had preoperative ICP and 17% had postoperative ICP by direct measurement
- 43% had language difficulties < 16th percentile vs 6.5% in general population
  - 34% without vs 75% with whole gene deletion
  - 50% who had ICP
  - Drop in longitudinal scores coinciding with onset of ICP

[a]Maliepaard et al., 2014; [b]de Jong et al., 2012; [c]Kilcoyne et al., 2019

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Crouzon Syndrome (1:62,500)

 = 11[a]

- 43% had brain differences
- 9% had Intellectual Disability (IQ < 70)
- No association between test scores with surgical status, age at surgery, brain differences, or socioeconomic factors

 = 10[b]

- 60% had brain differences
- 37% had Intellectual Disability (IQ < 70)
- Language skills and academics were below average
- No association between test scores with surgical status, age at surgery, or brain differences

[a]Yacubian-Fernandes et al., 2007; [b]Maximino et al. 2017

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Crouzon Syndrome

 = 11[a]

- 55% average range (IQ > 90)
- 18% below average range (IQ 80-90)
- 18% well below average range (IQ 70-80)
- 9% Intellectual Disability (IQ < 70)
- 91% completed secondary school
- 21% completed university

 = 39 parents[b]

- Greatest areas of parental concern in health-related quality of life were:
  - 50% vision
  - 44% speech
  - 39% cognition

[a]Flapper et al., 2009; [b]de Jong et al., 2012

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Apert Syndrome (1:65,000-100,000)

= 18[a]

- 56% had a brain difference identified on MRI
- 22% had Intellectual Disability (IQ < 70)
- No association between test scores with surgical status, age at surgery, or brain differences, but did find relationship between socioeconomic factors

= 8[b]

- 63% had brain differences on MRI
- 37% had Intellectual Disability (IQ < 70)
- Language skills and academics were below average
- No association between test scores with surgical status, age at surgery, or structural brain differences

[a]Yacubian-Fernandes et al., 2005; [b]Maximino et al. 2017

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Apert Syndrome

= 28[a]

- 43% IQ > 70
- 57% had Intellectual Disability
  - 29% IQ was 50-69
  - 29% IQ was 35-49
- 100% delayed language development
  - 18% later achieved typical speech skills
- 25% special education programs
- 39% general education with assistance
- 29% went to post-secondary training
- 36% employed

[a]David et al., 2016

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Apert Syndrome

 = 29[a]

- 48% had an IQ > 70
- 52% had Intellectual Disability
  - 31% had an IQ 50-70
  - 14% had an IQ 35-49
  - 7% had an IQ < 35
- No relationship between IQ with surgical approach or timing

 = 20 parents[b]

- Greatest areas of parental concern in health-related quality of life were:
  - 100% dexterity
  - 80% speech
  - 75% cognition
  - 50% vision

[a]Patton et al., 1988; [b]de Jong et al., 2012

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Pfeiffer Syndrome (1:100,000)

 = 42[a]

- Multiple classification systems proposed, including mild to severe functional impact
- The same genetic findings can present with a wide range of severity
- 17% had mild functional impact (14% with ADHD)
- 21% had moderate functional impact (44% delays in development; 22% with ADHD)
  - Both groups had one individual with a cloverleaf skull and one without craniosynostosis
- 62% had severe functional impact (50% elevated ICP; significant health issues and early mortality)
  - 15% had cloverleaf skull and 12% without craniosynostosis

[a]Greig et al., 2013

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Pfeiffer Syndrome

= 3[a]

- W290C variant in *FGFR2*
- varying typical surgical protocol was associated with average cognitive development
  - strip craniectomy release in first month of life with VP shunt as needed placed
  - posterior vault distraction around 6 to 9 months
  - either monobloc distraction or fronto-orbital advancement at 12 to 24 months
  - additional suboccipital decompression as needed

= 5[b]

- 40% average range (IQ > 90)
- 20% well below average range (70–80)
- 40% Intellectual Disability (IQ < 70)
- 40% completed secondary school (one with a 1:1 aide and high level of assistance)
- 20% completed university

[a]Wegner et al., 2019; [b]Flapper et al., 2009

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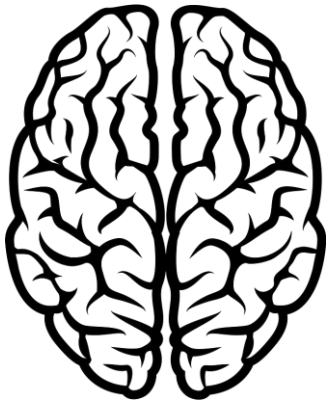
Implications



- We are still learning about the impact of syndromic craniosynostosis on development.
- ICP is a significant developmental risk factor that is likely occurring more often than noninvasive screening identifies.
- ICP tends to have an increased incidence after age 12 months.
- Beyond general timing, clear associations with specific surgical decisions are not reported and may be largely due to small groups of study participants.
- Management of ICP alone does not address multifactorial risk for developmental concerns.

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Implications

- There is wide variation in development outcomes within most diagnoses, even with identical genetic findings.
- Early and ongoing screening is instrumental in identifying individual needs and any additional diagnoses, such as ADHD or a learning disorder.
- Assessment results and recommendations can help ensure individuals receive appropriate interventions.
- In addition to patients' concerns, caregivers often experience stress and need their own support.

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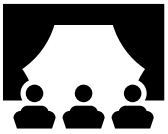
Family Role



- Be strong advocates across settings.
- Help support adherence to hearing aids and/or glasses.
- Link to state and local developmental support programs.
- Ensure Individualized Education Program (IEP) services ages 3-22.
- Be in frequent communication with teachers and interventionists.
- Support a wide variety of extracurricular activities.
- Include social opportunities with both general peer groups as well as seeking peers with similar diagnoses.
- Proactively coach children for how to respond to possible teasing and address concerns quickly if they occur.

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Looking Forward

- Significant support is available for individuals with Intellectual Disability and their families.
- Individuals with syndromic craniosynostosis rate their own quality of life similarly or higher than general population peers.
- Many adults with syndromic craniosynostosis have completed post-secondary education, are employed, and have families.
- Medical, surgical, developmental, school, and psychosocial supports are continuingly evolving with improved outcomes seen.

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References

Abu-Sittah GS, et al. Raised intracranial pressure in Crouzon syndrome: Incidence, causes, and management. *J Neurosurg Pediatr.* 2016;17:469-475.

Bannink N, et al. Health-related quality of life in children and adolescents with syndromic craniosynostosis. *J Plast Reconst Aesthet Surg.* 2010;63:1972-1981.

Cornille M, et al. *FGFR3* overactivation in the brain is responsible for memory impairments in Crouzon syndrome mouse model. *J Exp Med.* 2022;219:e20201879.

David DJ, et al. Apert syndrome: Outcomes from the Australian craniofacial unit's birth to maturity management protocol. *J Craniofac Surg.* 2016;27:1125-1134.

de Jong T, et al. Long-term functional outcome in 167 patients with syndromic craniosynostosis: Defining a syndrome-specific risk profile. *J Plast Reconst Aesthet Surg.* 2010:63:1635-1641.

de Jong T, et al. Health-related problems and quality of life in patients with syndromic and complex craniosynostosis. *Childs Ner Syst.* 2012;28:879-882.

Faasse M, et al. Guideline on treatment and management of craniosynostosis: Patient and family version. *J Craniofac Surg*. 2023;34:418-433.

Ferguson KT, et al. The physical environment and child development: An international review. *Int J Psychol.* 2013;48:437-468.

Flapper WJ, et al. Intellectual outcomes following protocol management in Crouzon, Pfeiffer, and Muenke syndromes. *J Craniofac Surg.* 2009;20:1252-1255.

Greig AVH, et al. Pfeifer syndrome: Analysis of a clinical series and development of a classification system. *J Craniofac Surg*. 2013;24:204-215.

Judy BF, et al. Intraoperative intracranial pressure monitoring in the pediatric craniosynostosis population. *J Neurosurg Pediatr.* 2018;22:475-480.

Junaid M, et al. Association between craniofacial anomalies, intellectual disability and autism spectrum disorder: Western Australian population-based study. *Pediatr Research.* 2022;92:1795-1804.

Kilcoyne S, et al. Language development, hearing loss, and intracranial hypertension in children with *TWIST1*-confirmed Saethre Chotzen Syndrome. *J Craniofac Surg.* 2019;30:1506-1511.

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References

Lloyd MS, et al. The quality of life in adult patients with syndromic craniosynostosis from their perspective. *J Craniofac Surg.* 2016;27:1510-1514.

Maliepaard M, et al. Intellectual, behavioral, and emotional functioning in children with syndromic craniosynostosis. *Pediatrics*. 2014;133:e1608-e1615.

Marucci DD, et al. Raised intracranial pressure in Apert syndrome. *Plast Reconstr Surg*. 2008;122:1162-1168.

Mathijssen IMJ, et al. Guideline for care of patients with diagnosis of craniosynostosis: Working group on craniosynostosis. *J Craniofac Surg.* 2015;26:1735-1807.

Maximino LP, et al. Syndromic craniosynostosis: Neuropsycholinguistic abilities and imaging analysis of the central nervous system. *Arq Neuropsiquiatr.* 2017;17:862-868.

Patton MA, Goodship J, Hayward R, Lansdown R. Intellectual development in Apert syndrome: a long term follow up of 29 patients. *J Med Genet.* 1988;25:164–167.

Reposo-Amaral CE, et al. Patient-reported quality of life in highest-functioning Apert and Crouzon syndromes: A comparative study. *Plast Reconstr Surg*. 2014:133:182e-191e.

Wang JC et al. Syndromic craniosynostosis. *Facial Plast Surg Clin N Am*. 2016;24:531-543.

Wenger TL, et al. A genotype-specific surgical approach for patients with Pfeiffer syndrome due to W290C pathogenic variant in *FGFR2* is associated with improved developmental outcomes and reduced mortality. *Genet Med.* 2019;21:471-476.

Yacubian-Fernandes, A, et al. Apert syndrome: Factors involved in cognitive development. *Arq Neuropsiquiatr.* 2005;63:963-968.

Yacubian-Fernandes, A, et al. Crouzon syndrome: Factors involved in neuropsychological development and quality of life. *Arq Neuropsiquiatr.* 2007;65:467-471.

Yu, M, et al. Cranial suture regeneration mitigates skull and neurocognitive defects in craniosynostosis. *Cell.* 2021;184:243-256.

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Resources

1. Faasse M, Mathijssen IMJ, & ERN CRANIO Working Group on Craniosynostosis. Guideline on treatment and management of craniosynostosis: Patient and Family Version. *J Craniofac Surg*. 2023;23:418-433. https://doi.org/10.1097/scs.0000000000009143
2. myFace's Guide to Craniofacial Conditions: https://www.myface.org/craniofacial-conditions/
3. American Cleft Palate-Craniofacial Association: https://acpacares.org/resource/educational-materials/
4. Children's Craniofacial Association informational booklets: https://ccakids.org/syndromes.html
5. Information about IEPs: https://www.handyhandouts.com/search.aspx?searchstr=IEPs+and+Testing

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Meet Jake and his family

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Questions & Answers

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Closing Remarks

Stephanie Paul
Executive Director
myFace

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myFace Offers a "Guide to Craniofacial Surgeries"

- myFace offers a FREE tool that provides in-depth information about some of the most common craniofacial surgeries, including
  - BSSO, FOA, Genioplasty, Le Fort I, Le Fort III, Monobloc
- Includes expert medical advice + firsthand accounts from patients and families
- Available at: www.myface.org/surgeries/

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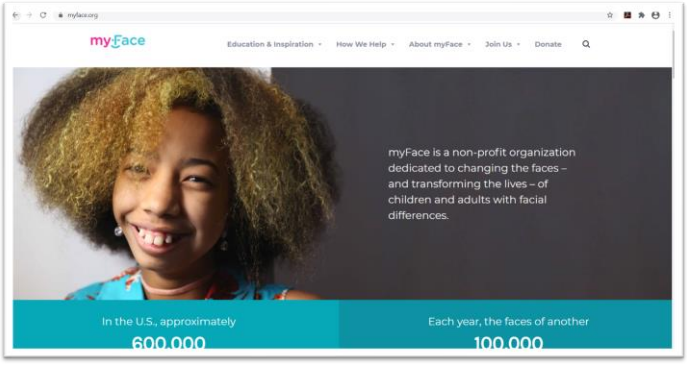
For additional information and resources visit **myFace.org**



Or email us at
info@myface.org

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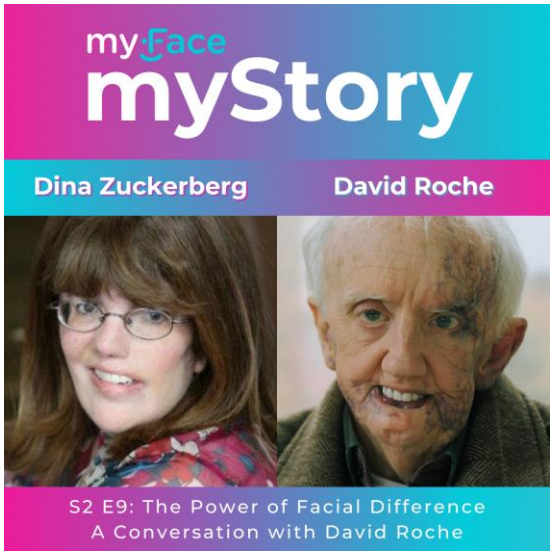
Upcoming Events at myFace

A new episode of our *myFace, myStory* podcast will debut on all podcast platforms + YouTube on

Wednesday, Feb. 15th

This month's episode will be a conversation with inspirational humorist David Roche to discuss his vascular malformation diagnosis and his lifelong journey to find self-acceptance and true love.

Register at:
www.myface.org/mystory/



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Upcoming Events at myFace

Join us on
Monday, May 15th at 6 PM ET
for musical performances, awards, and a benefit for the craniofacial community.

You can participate either in person in New York City, or online.
Further details will be available on the myFace website soon.

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Upcoming Events at myFace

Join us for the next webinar in our ***Transforming Lives*** **Webinar Series:**

Navigating Adolescence with a Craniofacial Condition: Strategies for Empowerment and Success – June 15, 2023

Meredith Albert, PhD
Pediatric Psychologist
Shriners Hospitals for Children, Chicago
Assistant Clinical Professor
University of Illinois at Chicago

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