Hemangioma

Presented by: Cameron Freelove DDS

Introduction

- Also known as infantile hemangioma
- Most common benign tumor of infancy
- Affects 5-10% of infants
- Appear within the first few weeks of life
- Marked by early proliferation, then spontaneous involution

Etiology

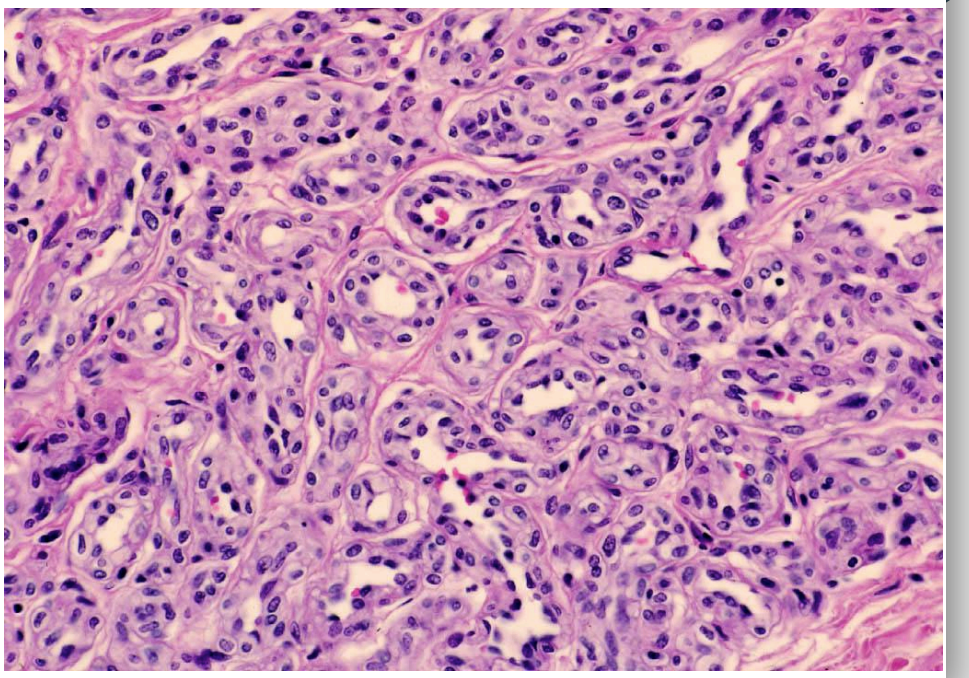
- Currently, not really known, however there are theories
- Some studies show that localized soft tissue hypoxia along with increased estrogen after birth may be a cause.
- Other theories link causes to placental tissue, endothelial progenitor cells, and mesenchymal stem cells.
- Little evidence linking hemangioma to hereditary influences, although there is a study suggesting it may be linked to an autosomal dominant trait.

Epidemiology

- In white infants, 3% present at birth, 10% present at one year of age.
- Incidence is 22-30% for preterm infants that weigh less than 1kg.
- 10-12 times more likely in white infants than in black or asian infants.
- 3:1 ratio among females to males

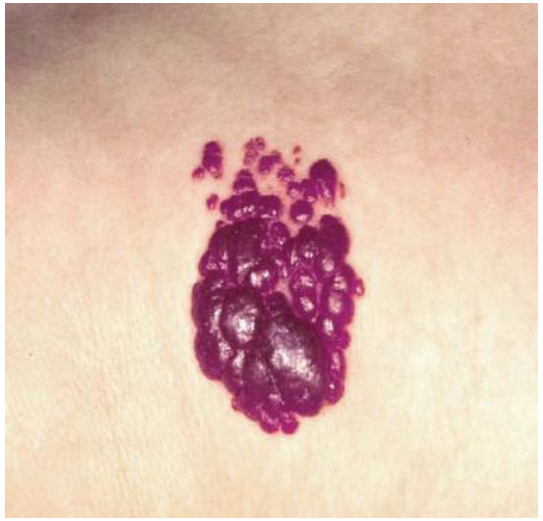
Pathophysiology

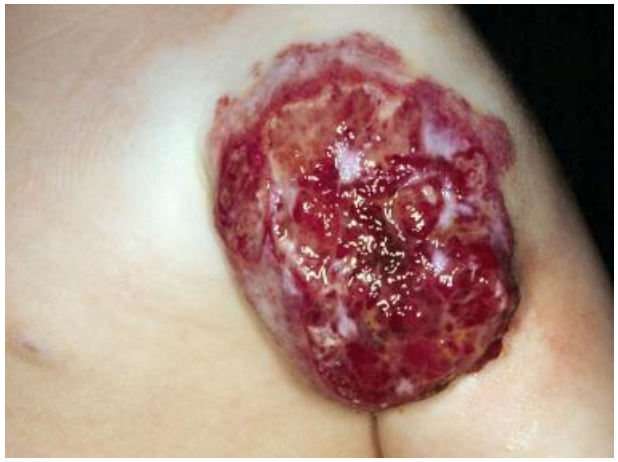
- Hemangiomas consist of proliferating endothelial cells.
- These endothelial cells eventually form vascular channels.
- Angiogenic peptides, such as beta-fibroblast growth factor, vascular endothelial growth factor (VEGF), and proliferating cell nuclear antigen cause proliferation of these endothelial cells.
- This causes hemangioma.

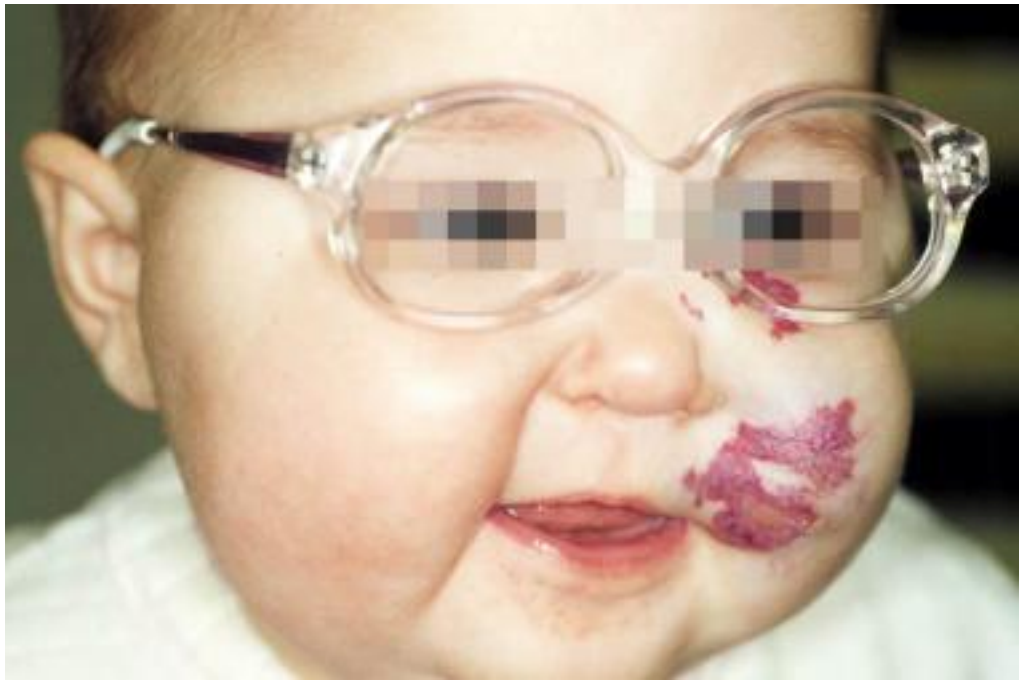


Clinical Presentation

- Early lesions start as blanching of the skin, moving on to fine telangiectasias, and finally to a red/purple macule or papule
- As hemangioma ages, it may have dome shaped, bosselated, plaquelike, or tumoral textures.
- Color depends on depth of hemangioma
- Consistency is firm, rubbery
- Most are within .5-5cm, but can reach up to 20cm in diameter
- Eighty percent of infantile hemangiomas are focal and solitary. Sixty percent of cutaneous hemangiomas occur on the head and neck, 25% on the trunk, and 15% on the extremities







Diagnosis

- Combination of history and physical exam.
- Most develop weeks after birth, so if growth is there at birth, it most likely is not a hemangioma
- Superficial hemangiomas are bright red, those deeper in subcutaneous tissues are more purple.
- Rapid growth in first 4-8 weeks with slower growth over next 6-9 months
- Most growth done in 3 months
- For some deeper hemangiomas, ultrasound or MRI can help diagnose

Treatment/Management

- Most do not require any intervention and will involute over the course of years. Regression usually starts at about 5 years of age.
- Corticosteriods have been used in the past.
- Interferon-a and vincristine are other medications for hemangioma due to their anti-angiogenic properties.
- 1st line of defense - Beta blockers, specifically Propranolol
  - Vasoconstriction, downregulation of VEGF, and apoptosis.
- Surgery is rarely indicated, but can be used when medical approaches fail.

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PIERRE ROBIN SEQUENCE

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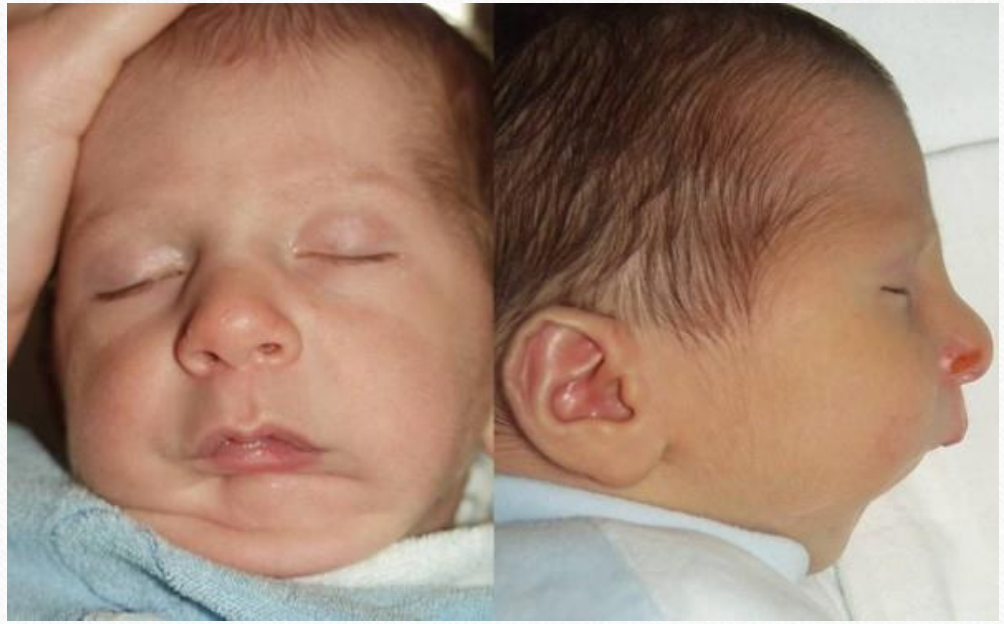
Introduction

- Previously referred to as Pierre Robin Syndrome.
- Congenital anomaly in development of craniofacial structures
- Characterized by Micrognathia and Glossoptosis
- Resulting in upper airway obstruction, stridor, cyanotic episodes and feeding difficulties.
- The incidence is approximately 1 in 85 000 children.
- Can occur in isolation or with other but more often it is associated with other syndromes. Stickler syndrome is the most commonly associated diagnosis followed by velo-cardio-facial syndrome.

Background

- Lannelongue and Menard first described Pierre Robin syndrome in 1891
- In 1926, Pierre Robin published the case of an infant with the complete syndrome.
- Until 1974, the triad was known as Pierre Robin syndrome; however, the term syndrome is now reserved for those errors of morphogenesis with the simultaneous presence of multiple anomalies caused by a single etiology.

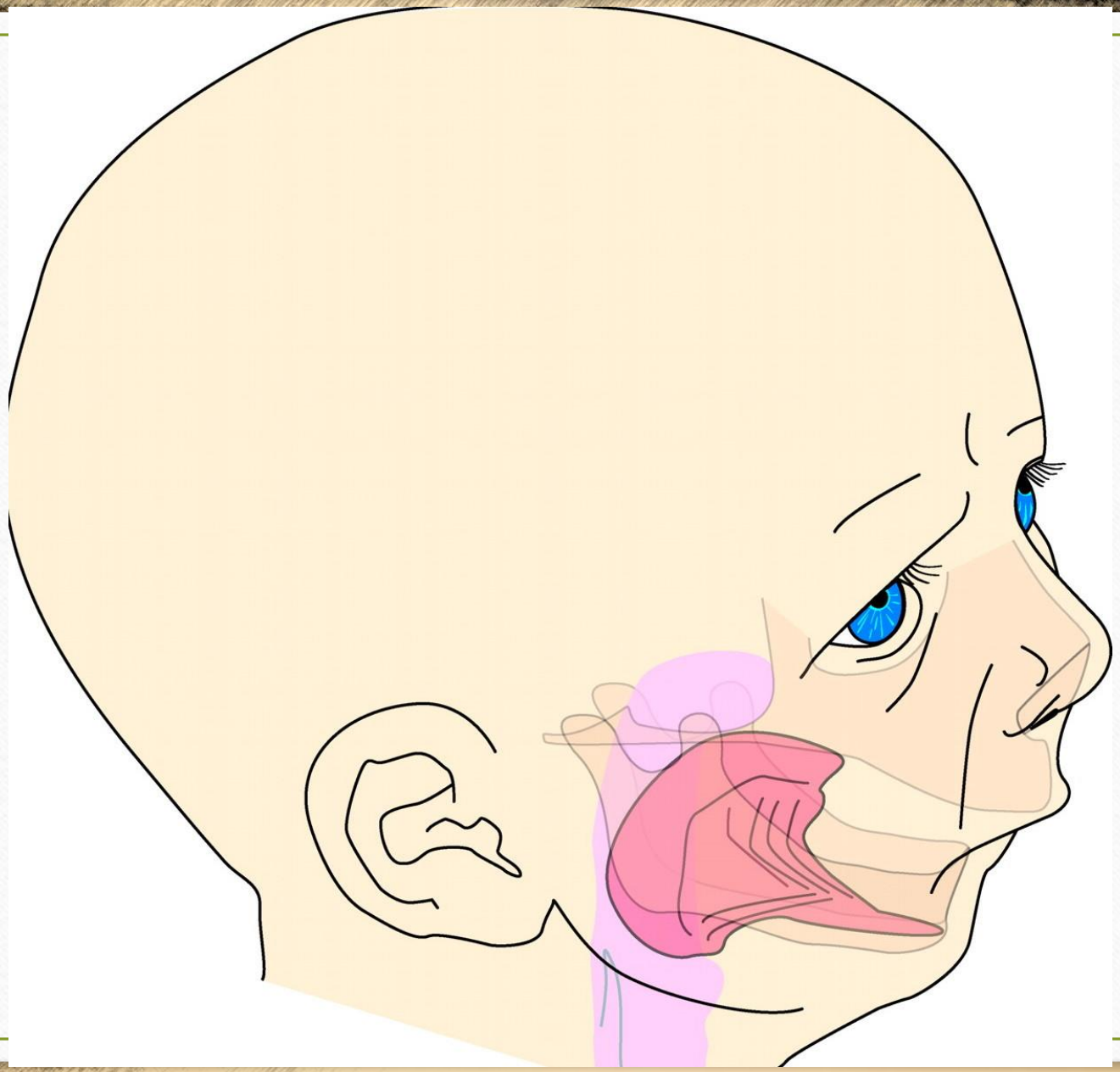


Etiology

- Support for a genetic basis is evidenced by a high incidence of twins with PRS.
- Jakobsen et al suggests that non-syndromic PRS is associated with SOX9 and KCNJ2 dysregulation, both on chromosome 17. SOX9 gene codes for a transcription factor, SOX9 protein which is pivotal for skeletal development. KCNJ2 on the other hand belongs to a large family of genes that produce potassium channels which are active in skeletal and cardiac muscles.
- Izumi et al evaluated two cohorts of patients clinically diagnosed with PRS using fluorescence in situ hybridization (FISH) and array comparative genomic hybridization (CGH). This study concluded that 40% of PRS was isolated and 60% were associated with another syndrome, most commonly Stickler and velocardiofacial syndromes.

Pathogenesis

- Three pathophysiological theories exist to explain the occurrence of Pierre Robin sequence.
- **The mechanical theory**: most accepted. Main event: mandibular hypoplasia occurring between 7^{th} and 9^{th} week of gestation. This disrupts normal palate formation. Theory explains why cleft of palate occurs in absence of clefting in lip. Oligohydramnios could play a role in the etiology since the lack of amniotic fluid could cause deformation of the chin and subsequent impaction of the tongue between the palatal shelves



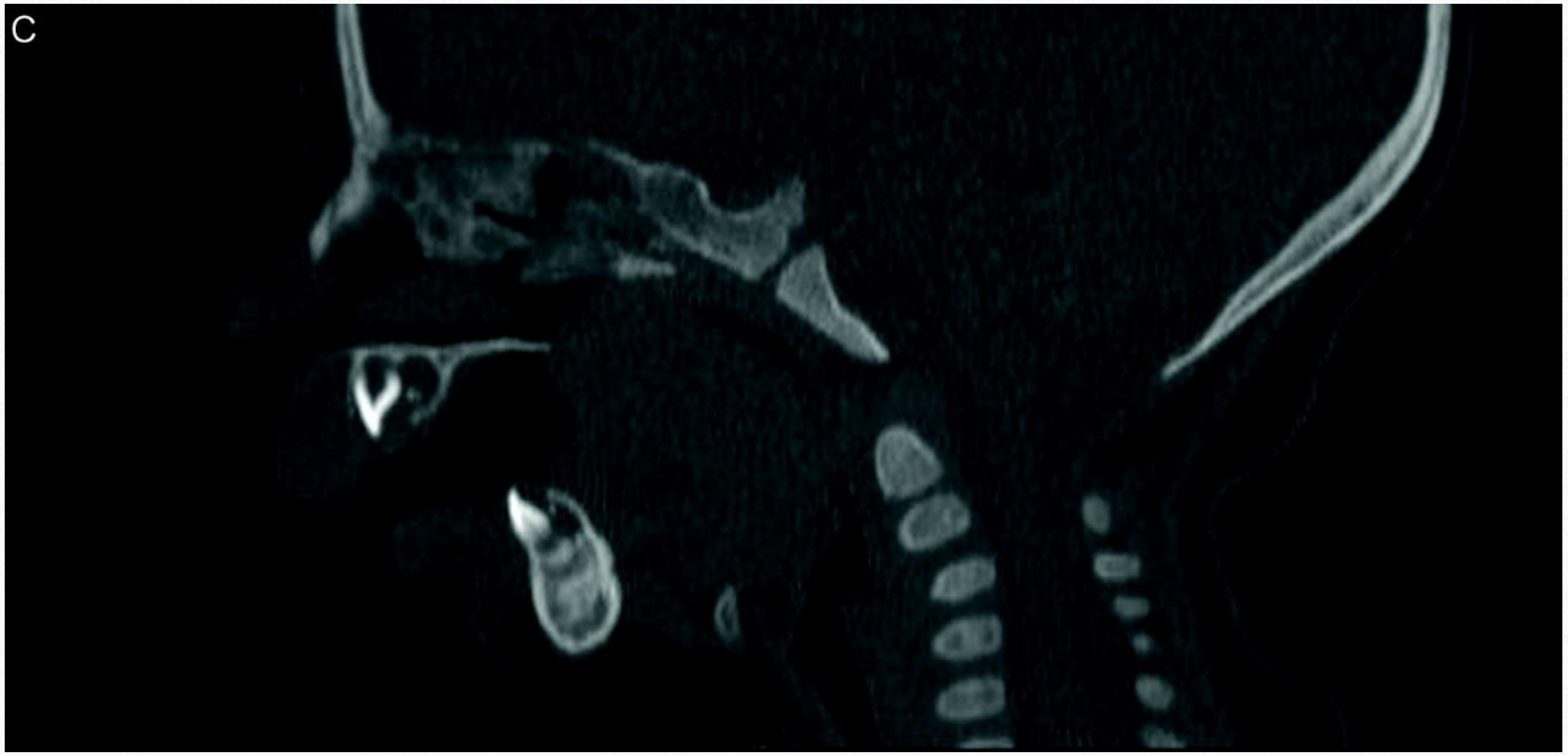
Pathogenesis

- **The neurological maturation theory:**
- A delay in neurological maturation has been noted on electromyography of the tongue musculature, the pharyngeal pillars, and the palate, as has a delay in hypoglossal nerve conduction. The spontaneous correction of the majority of cases with age supports this theory.
- **The rhombencephalic dysneurulation theory.**

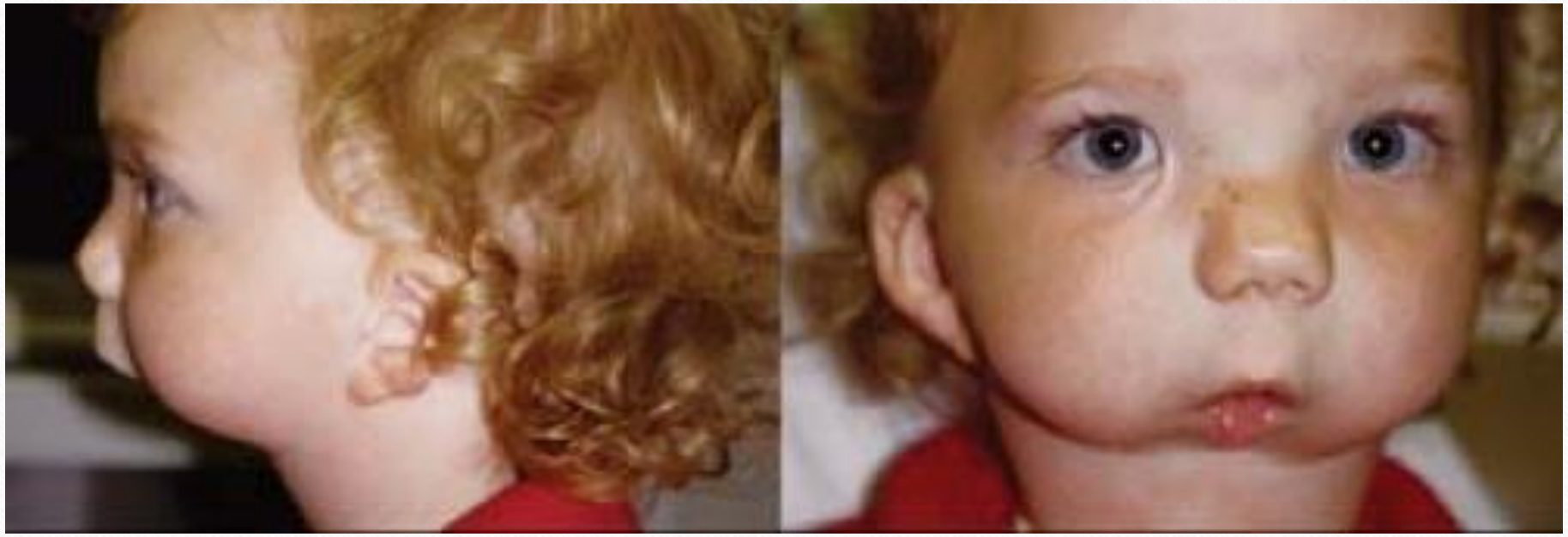
Clinical features

- **Microretrognathia** is immediately identified at birth and is a defining feature of the diagnosis
- Hypoplastic mandibles are small in both the vertical and horizontal dimensions.
- Apart from the micrognathia, Randall described the notable finding of retrogenia, or posterior displacement of the chin, to characterize the initiating anomaly in this sequence
- **Glossoptosis,** defined as an abnormal posterior placement of the tongue, is the second characteristic feature of PRS.

Lateral Cephalogram showing the mandibular microretrognathia



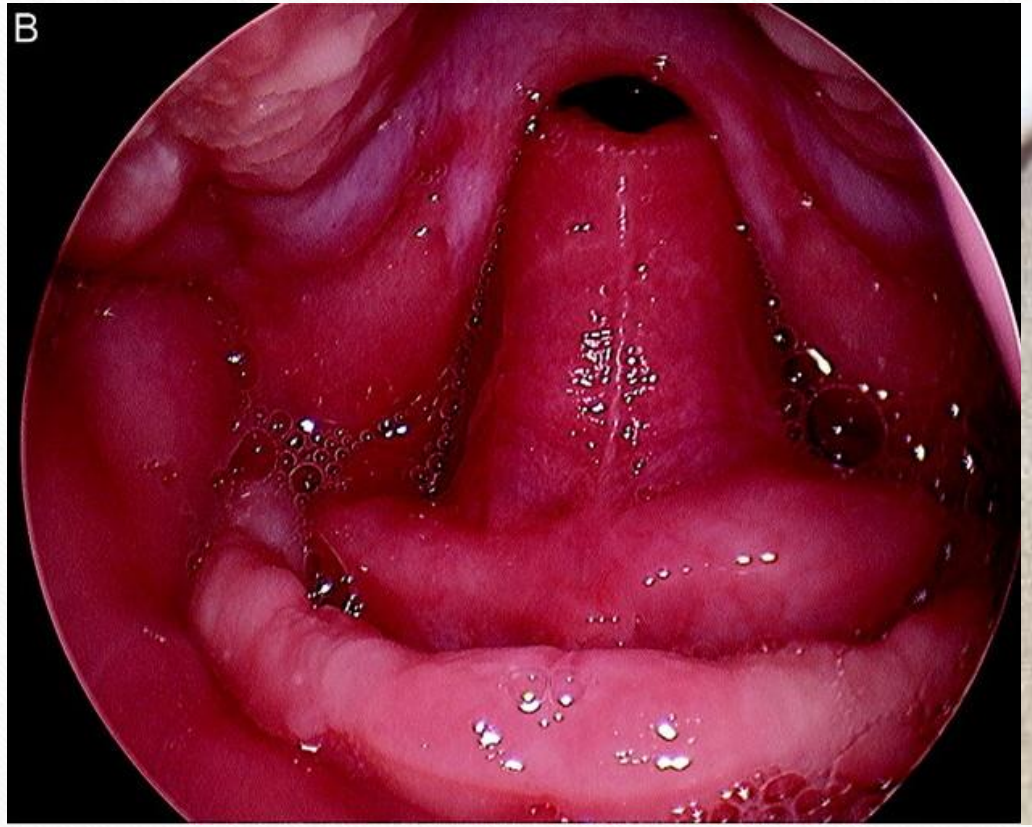
Severe mandibular microretrognathia



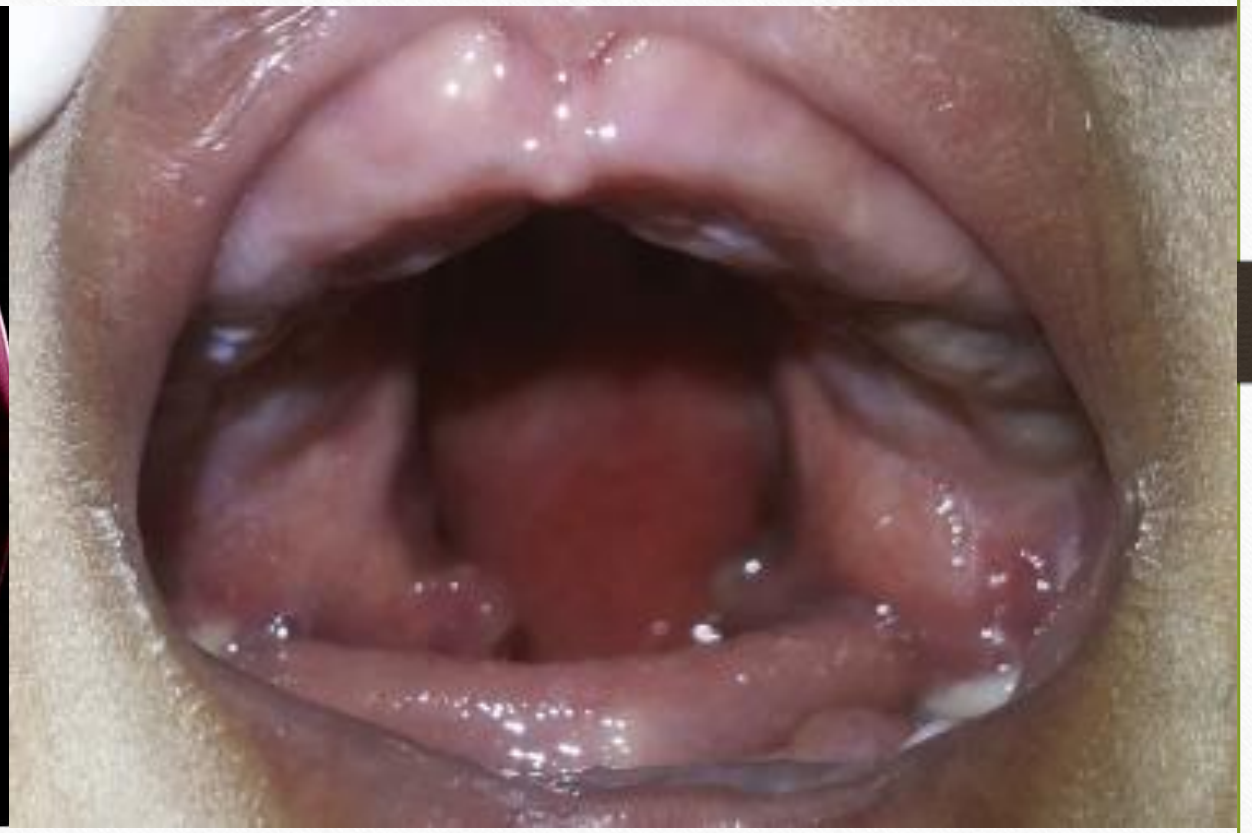
Clinical features

- Although the tongue is typically noted to be of normal size, the hypoplastic mandible provides less volume in the oral cavity and forces the tongue to fit into a smaller space, which further serves to exacerbate the blockage of the posterior pharynx.
- **Third defining feature: unclear.** Either palatal defect or airway obstruction.
- Airway Obstruction: may be at the level of tongue base. In order to combat the obstruction, high volume of energy is expended to continue breathing manifested by suprasternal retractions and the use of accessory muscles of respiration.

Glossoptosis



U shaped clefting of palate



Clinical features

- Feeding difficulties are common as infants struggle to breathe during eating. Gastroesophageal reflux and aspiration are common sequelae of this process.
- The associated cleft prevents the formation of negative intraoral pressure, which is required to suck milk from the breast or bottle; the micrognathia and glossoptosis further impede mechanical sucking.

Systemic manifestations

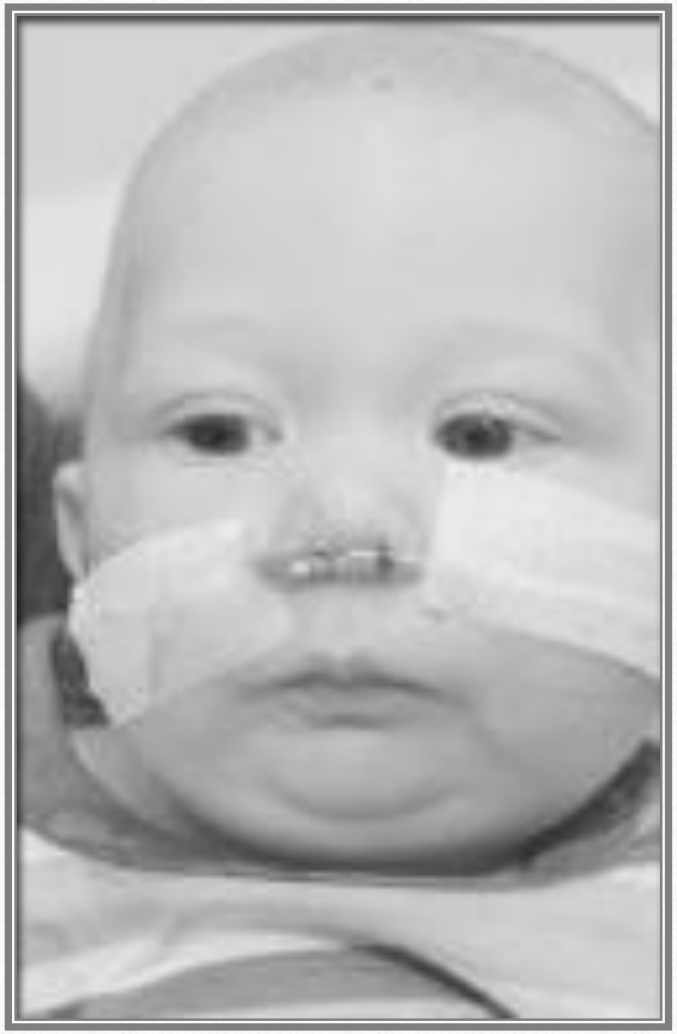
- Anomalies involving the **musculoskeletal system** are the most frequent systemic anomalies (noted in 70-80% of cases). They include syndactyly, dysplastic phalanges, polydactyly, hyperextensible joints etc.
- **Central nervous system (CNS)** defects such as language delay, epilepsy, neurodevelopmental delay, hypotonia, and hydrocephalus may occur. The incidence of CNS defects is around 50%.
- **Cardiovascular findings** such as benign murmurs, pulmonary stenosis, patent ductus arteriosus, pulmonary hypertension etc. Their prevalence varies in the literature from 5-58%.

Management
Non Surgical

Isolated PRS patients usually respond more favorably to the conservative approach.

Prone or lateral positioning will solve the airway obstruction in ~70% of cases of PRS.

With appropriate positions, many of these children will also be able to feed normally and no further treatment is necessary.



Management

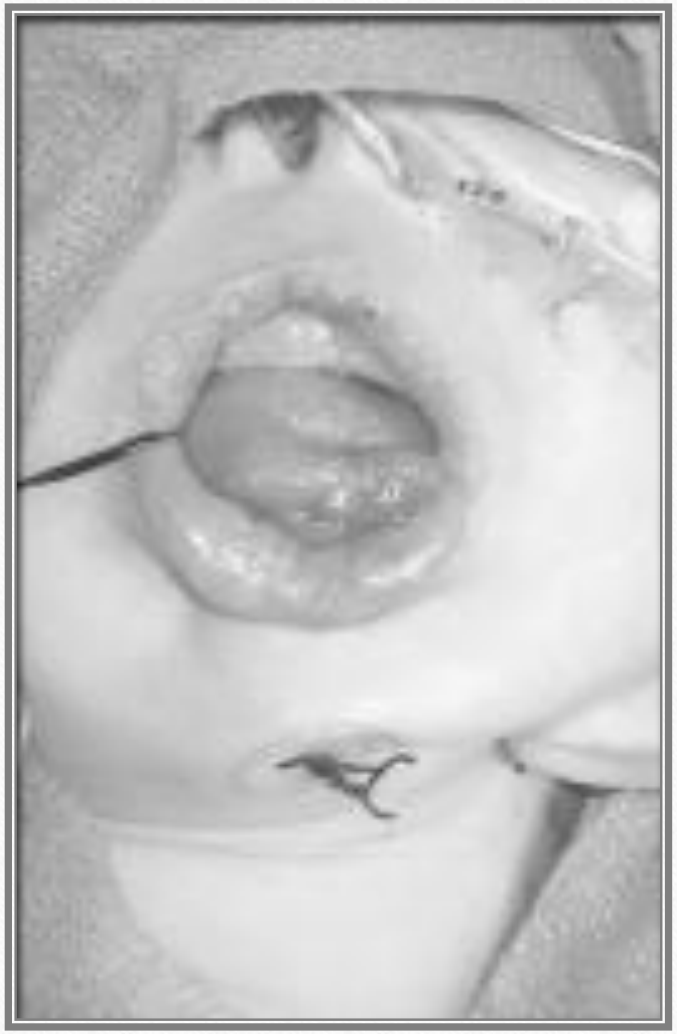
Surgical

Tongue–Lip Adhesion

The procedure serves to correct the problem of glossoptosis by pulling the base of the tongue forward and suturing it to the lower lip.

This is the method of choice when the obstruction is at the level of tongue base and cannot be corrected by positioning alone.

Once healed, this mucosal attachment serves to tether the tongue anteriorly until the infant develops a more stable airway with growth. TLA can only be performed on infants who have not developed any lower teeth, as they could otherwise bite through the repair inadvertently. After adequate growth has occurred, the TLA must be released with a second procedure.

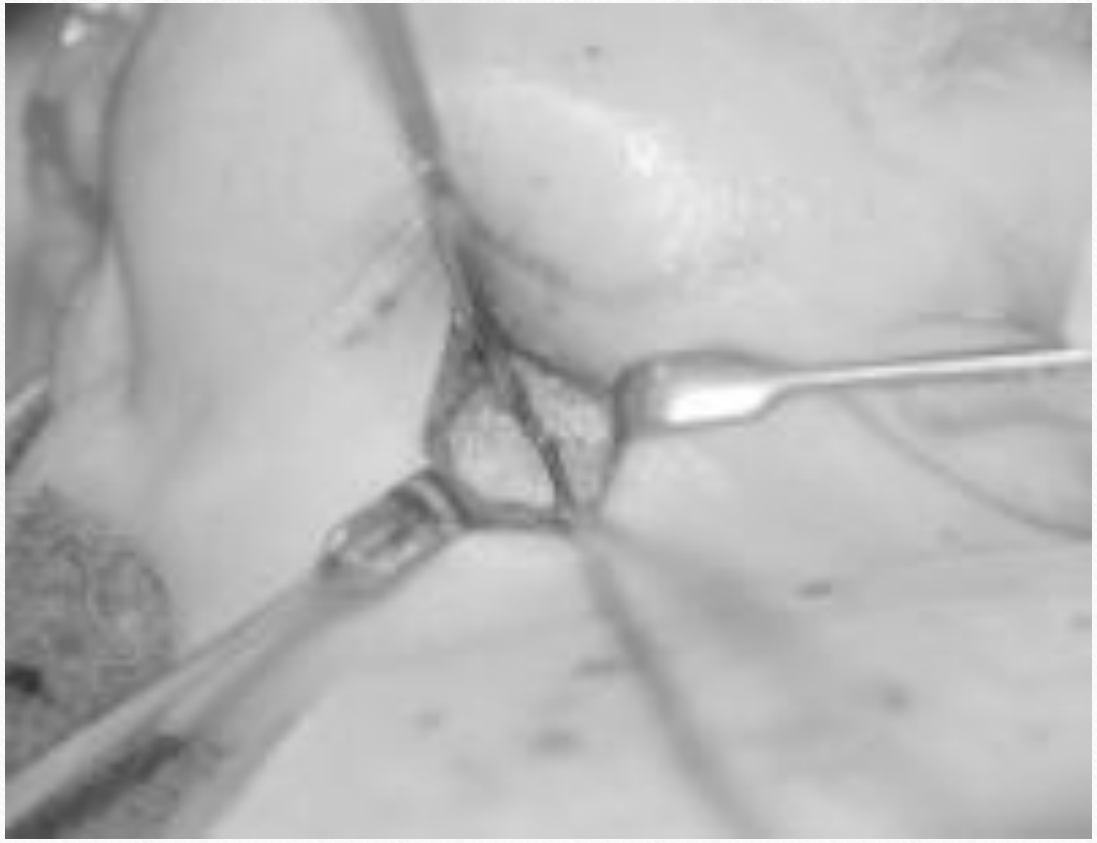


Management
Surgical

Distraction Osteogenesis of the Mandible

- Definitive technique to address the issues associated with PRS by relieving airway obstruction, improving facial cosmetics, and correcting malocclusion.
- As the mandible is projected forward, the tongue is also pulled anteriorly through its muscular attachments on the lingual surface of the mandible. Thus, this technique reverses the sequence of PRS by correcting the micrognathia, which in turn improves the glossoptosis, thereby relieving the obstruction of the airway.
- Surgeons vary in the length of the latency period before distraction is initiated. This period can range from 24 hours to 7 days, though most surgeons wait 2 to 3 days before activating distraction

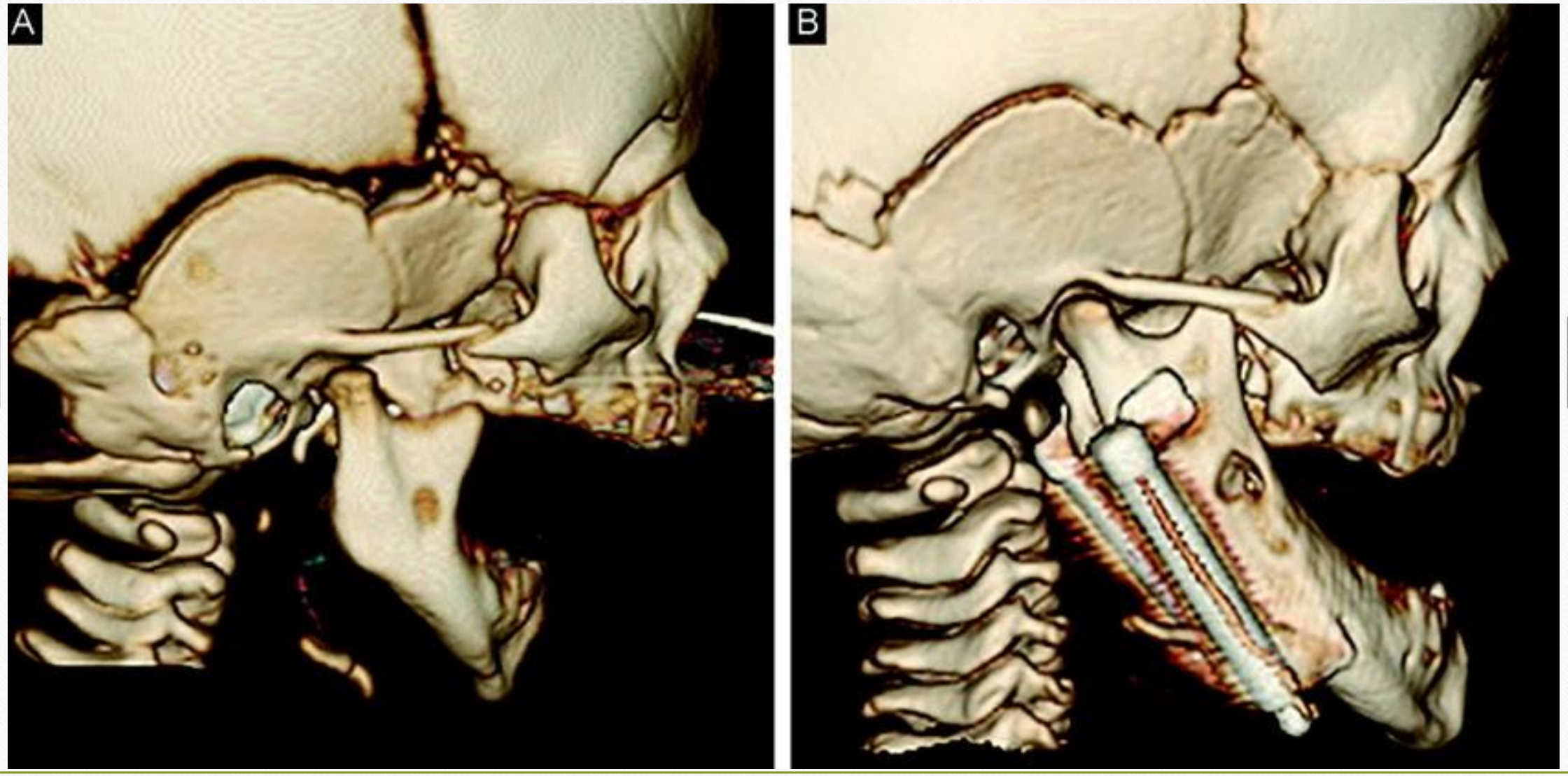
Distraction Osteogenesis



270 degree osteotomy cut

Placement of internal univector distractor

Distraction Osteogenesis



Pros and Cons of distraction osteogenesis

- The major advantage of DO is the avoidance of tracheotomy for airway management. Moreover, in several reported series, those patients who underwent tracheotomy prior to DO were able to be successfully decannulated after DO was complete

- Complications of distraction osteogenesis: pin site infections, inferior alveolar neurapraxia, injury to the tooth roots, unacceptable scarring, resorption/ankylosis at the temporomandibular joint, malunion, and failure of distraction due to incomplete osteotomies, early consolidation, or device failure

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Monasterio, Fernando Ortiz MD; Drucker, Mónica MD; Molina, Fernando MD; Ysunza, Antonio MD

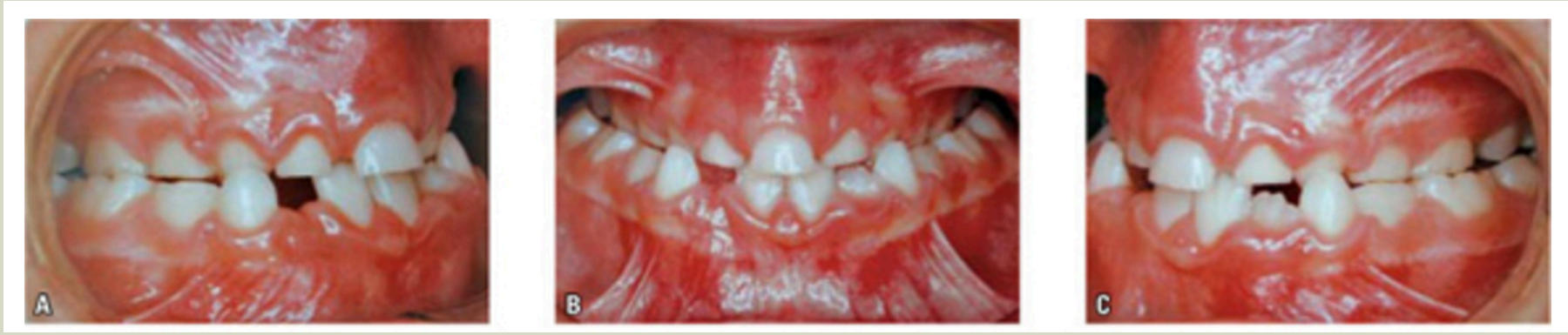
THANK YOU.

SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR (SMMCI) SYNDROME

Janice Lee

SMMCI

- Complex disorder consisting of multiple, mainly midline defects together with other midline structures of the body of development resulting from an unknown factor(s) or events occurring between the 35th and 38th days *in utero* together with other midline structures of the body [3]

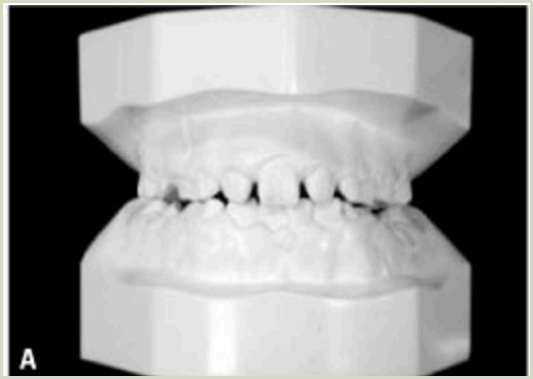


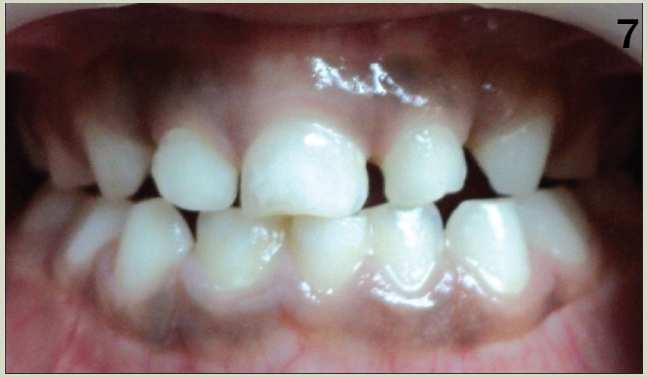
SMMCI

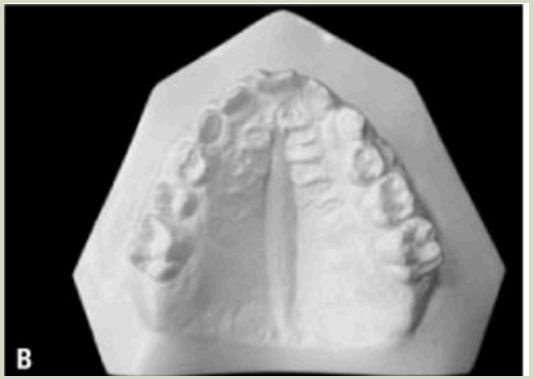
- Maxillary dental laminae normally fuse in midline between days 38-40 *in utero*
  - Left and right dental laminae fuse prematurely in the midline
    - Prevents normal formation of the central incisor tooth germs, intervening bone, and soft tissue
- Normal lateral growth of the maxillae and orbits, together with the other midline structures in the region slows or ceases
- Etiology is uncertain[3]

SMMCI TOOTH

- **Solitary**: tooth present exists as the only central incisor tooth in the maxilla
- **Median**: tooth present precisely in the midline of the maxillary alveolus
- **Maxillary**: tooth occurs only in the maxilla and not in the mandible
- **Central incisor**: a central incisor tooth, although of unusual crown form, and is not a supernumerary tooth (mesiodens) [3]

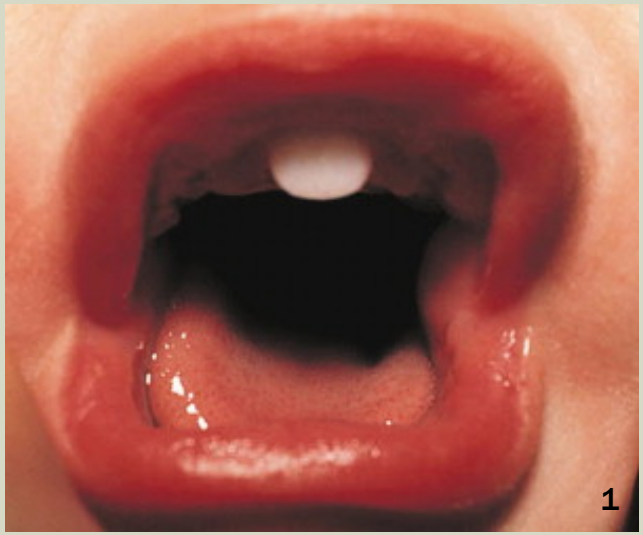






SMMCI TOOTH

- Symmetric
- Develops and erupts in the midline of the maxillary dental arch in both primary and permanent dentitions
- May appear as isolated trait, but other clinical manifestations present majority of the time [3]

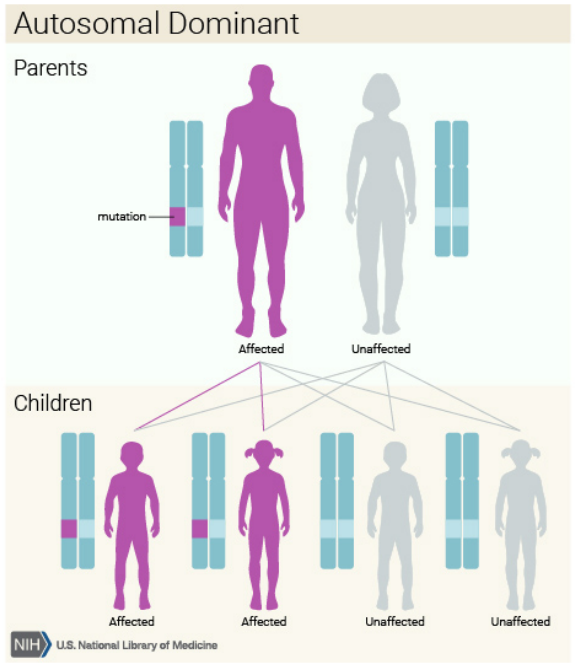


DIAGNOSIS

- Ultrasound at 18-22 weeks, or with genetic testing in familial cases via ultrasound scan
- Rarely made prenatally
- Diagnosis no later than 8 months of age on eruption of the primary maxillary incisor tooth
- Estimated to occur in 1:50,000 live births [3]

ETIOLOGY & INHERITANCE

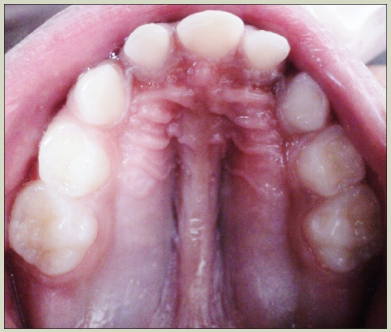
- Unknown [3]
- Suspected missense mutation in SHH at 7q36
- Heterogeneous condition [4]
- Autosomal dominant [8]
- Occurs in 1:50,000 live births [3]

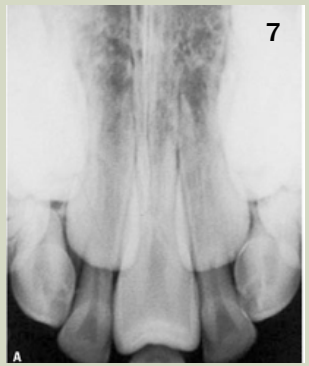


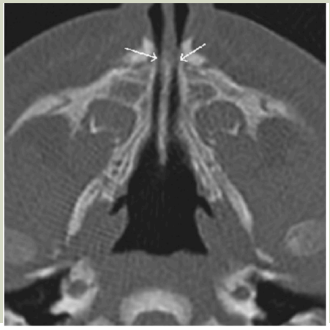
CLINICAL MANIFESTATIONS

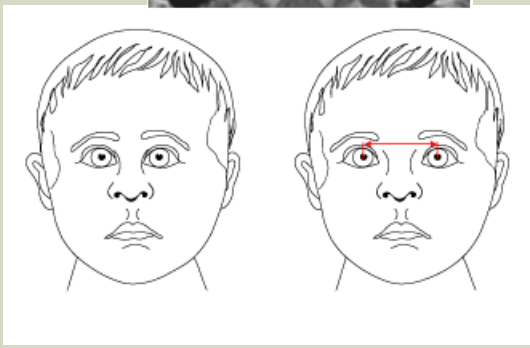
- Preterm birth and low birth weight
- Absent labial frenum
- Narrow nose
- Arch-shaped appearance of upper lip
- “V”-shaped palate with ridge along midpalatal suture
- Radiographically absent intermaxillary suture
- Congenital nasal airway obstruction
  - Choanal atresia
  - Midnasal stenosis
  - Congenital nasal pyriform aperture stenosis

  (CNPAS) in over 90% cases – nose may appear Hypoplastic and nostrils anteverted
- Short stature (50% of cases)
- Small head circumference
- Deviant sella turcica and pituitary gland morphology
- Hypotelorism
- Holoprosencephaly
- Intellectual disability of varying degrees
- Hypopituitarism
- Cleft lip and palate
- Congenital heart disease [3]



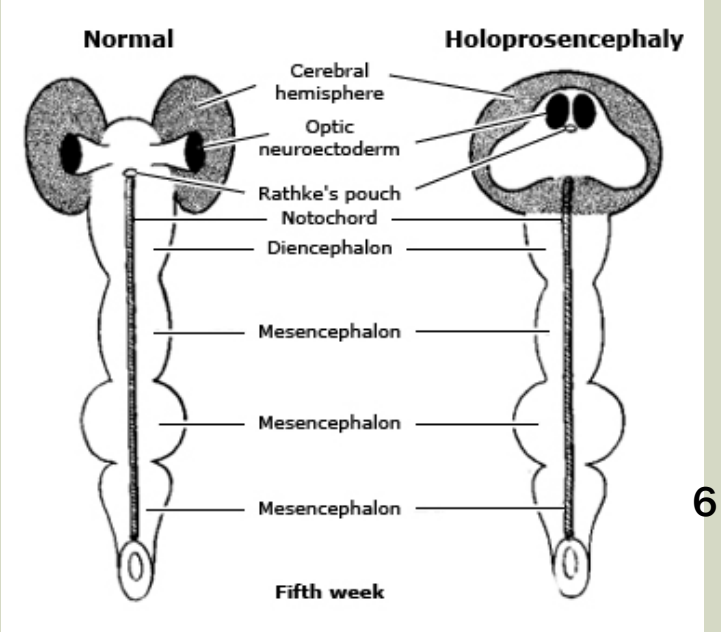






HOLOPROSENCEPHALY

- (HPE) is most described association with SMMCI
- A child with SMMCI should be examined for signs of holoprosencephaly
- When HPE is present, clinical expression is extremely variable ranging from alobar HPE and cyclopia, microforms of HPE
- Developmental field defect of the forebrain in which the cerebral hemispheres fail to separate into distinct halves [3]



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HOLOPROSENCEPHALY

- Cohen 2001
  - SMMCI should not be considered a microform of holoprosencephaly (HPE), but as either:
    - (1) an integral component of severe HPE;
    - (2) an anomaly that may occur in conditions unrelated to HPE;
    - (3) a solitary manifestation in some members of a dominantly affected family whose members have variable expressivity to HPE with incomplete penetrance; or
    - (4) (more rarely) as an isolated dominant trait with an SHH mutation [3]

ASSOCIATIONS

- Syndromes
  - CHARGE
  - VACTERL
  - VCF
  - Autosomal dominant holoprosencephaly
  - Ectodermal dysplasia
  - Duane retraction syndrome
- Chromosome abnormalities
  - Del(18p)
  - R(18)
  - Del(7q36qter)
  - 47XXX
  - Del(22q11.2) [3]

MANAGEMENT

- Depends on individual anomalies present
  - Emergency surgery for choanal stenosis
  - Plastic surgery for congenital pyriform aperture stenosis
  - Growth hormone therapy
- SMMCI tooth can be treated with orthodontic and/or restorative care for esthetic purposes [3]

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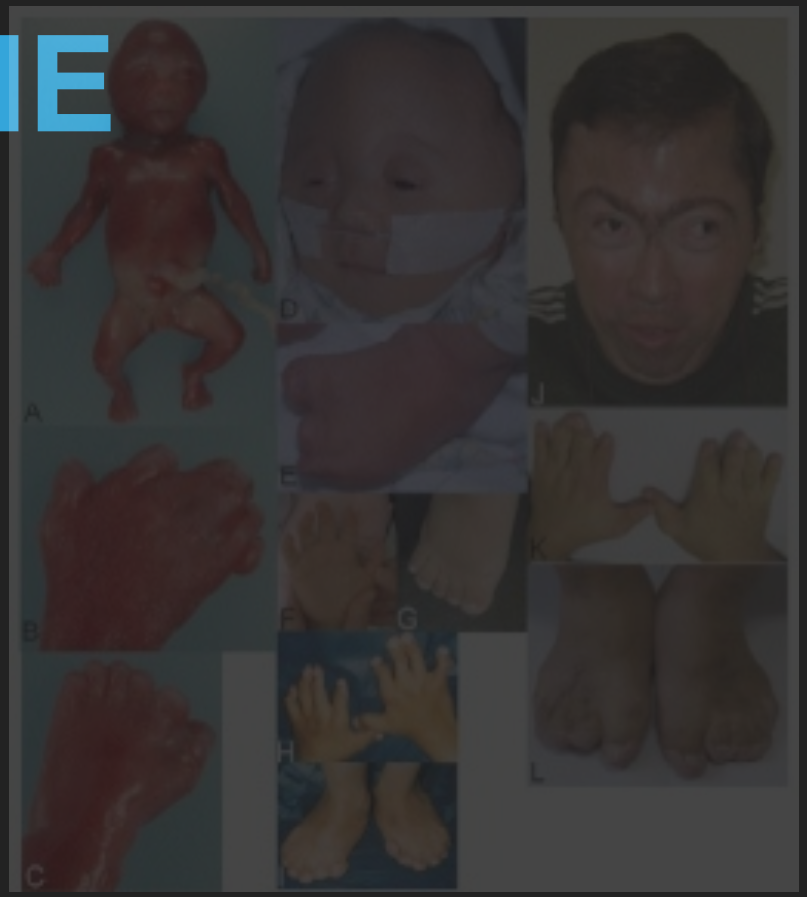
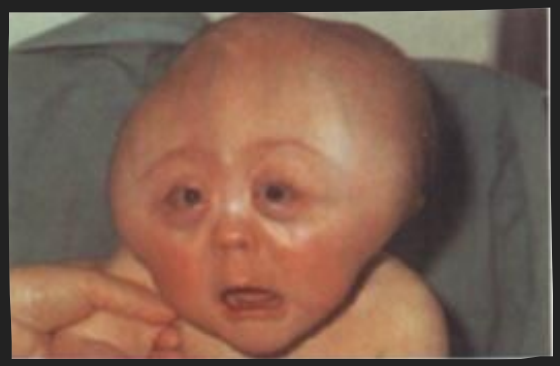
ADVANCED PERIODONTOLOGY, USC

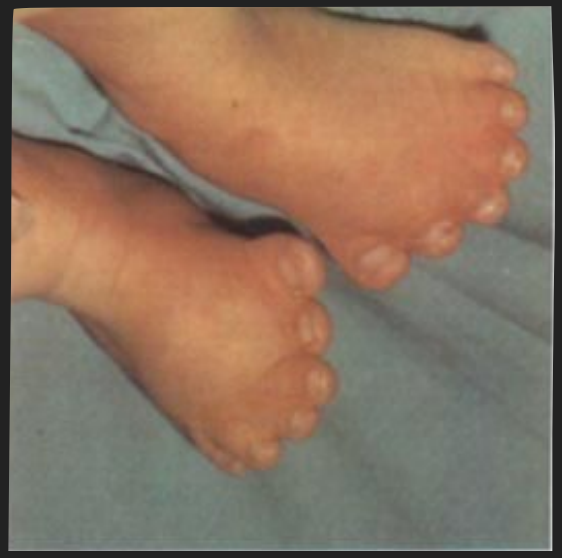
CARPENTER'S SYNDROME

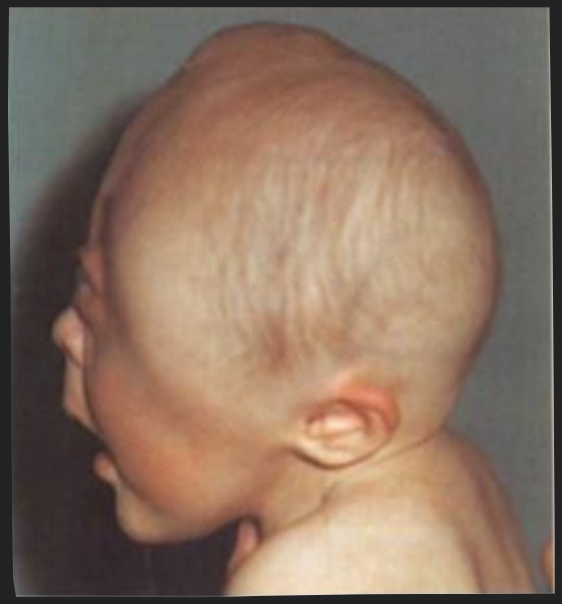
OUTLINE

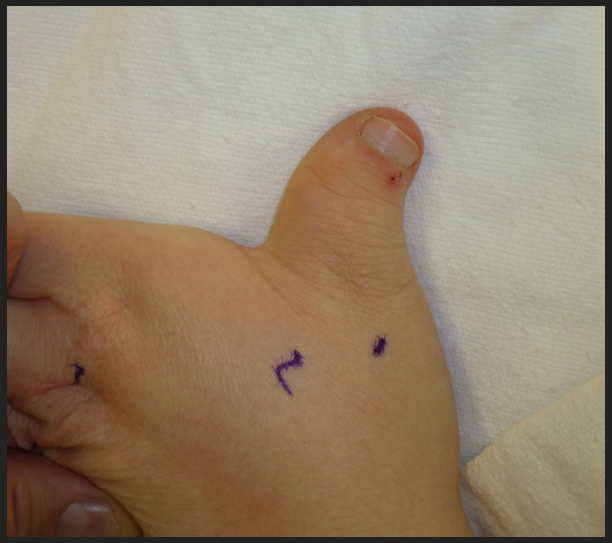
- DESCRIPTION
- FREQUENCY
- GENETIC CHANGES
- INHERITANCE PATTERN
- DIAGNOSIS & MANAGEMENT

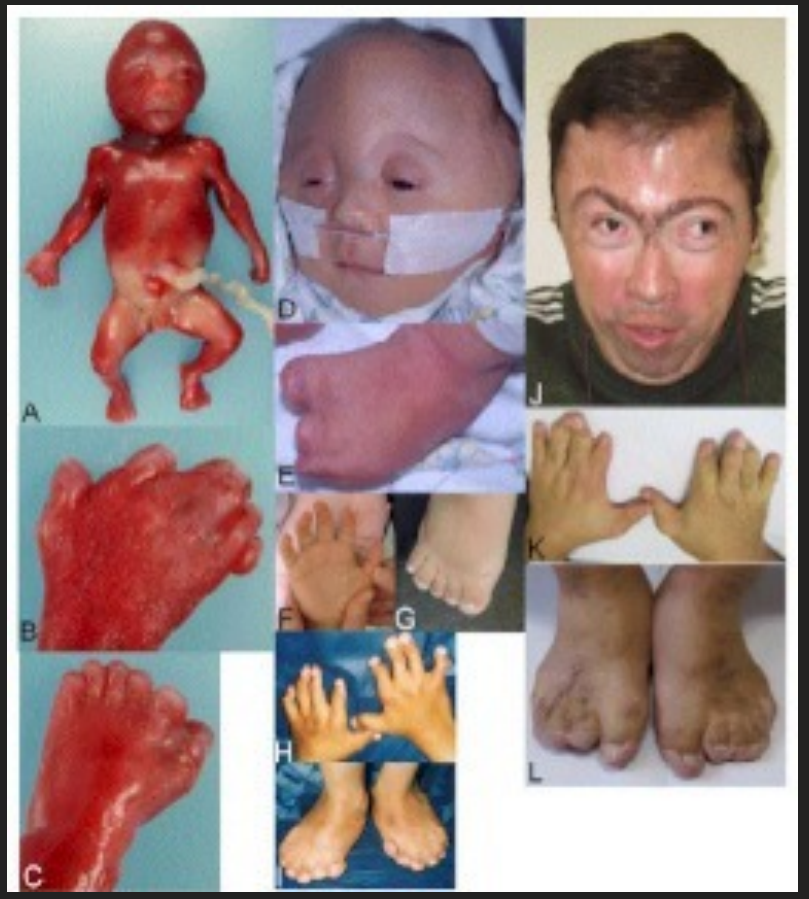








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DESCRIPTION

- Craniosynostosis
- Abnormalities of fingers and toes: polydactyly, syndactyly, brachydactyly
- Obesity
- Other developmental problems:
  - underdeveloped upper and lower jaws
  - Dental abnormalities: baby teeth

Altunhan et al., 2011

DESCRIPTION

- Craniosy
- Abnorm yndactyly, brachyd
- Obesity
- Other de
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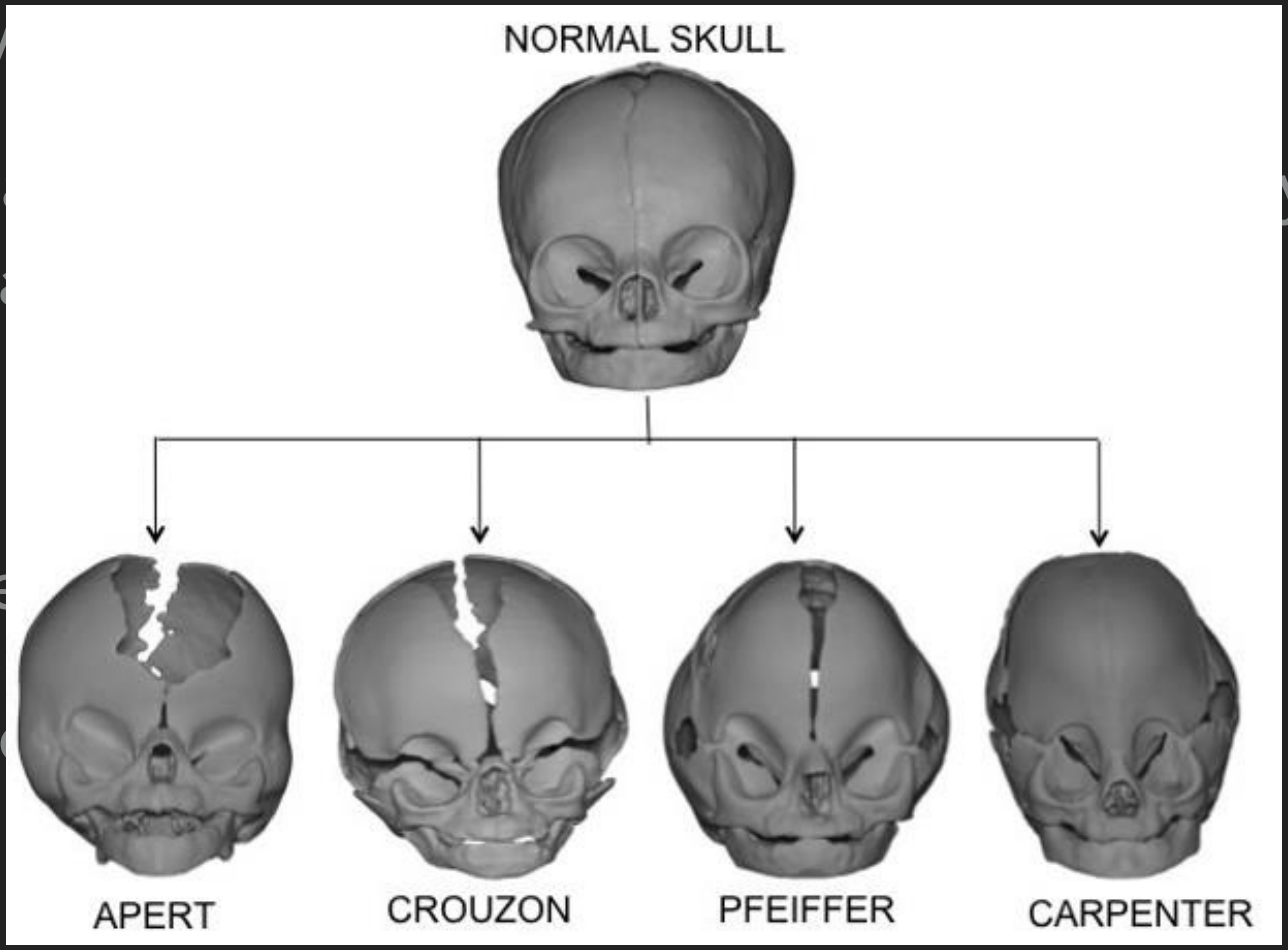


ADDITIONAL FEATURES

Mild retardation	hydrocephalea
dysmorphic facial features	molar agenesis
abnormal male genitalia	heart defects

- Signs & symptoms vary considerably
- Life expectancy is shortened but extremely variable

Greig cephalopolysyndactyly syndrome

Overlapping features can cause these two conditions to be misdiagnosed

Genetic testing is often required for accurate diagnosis

FREQUENCY

- Very rare condition
- Aproximately **100** cases reported worldwide

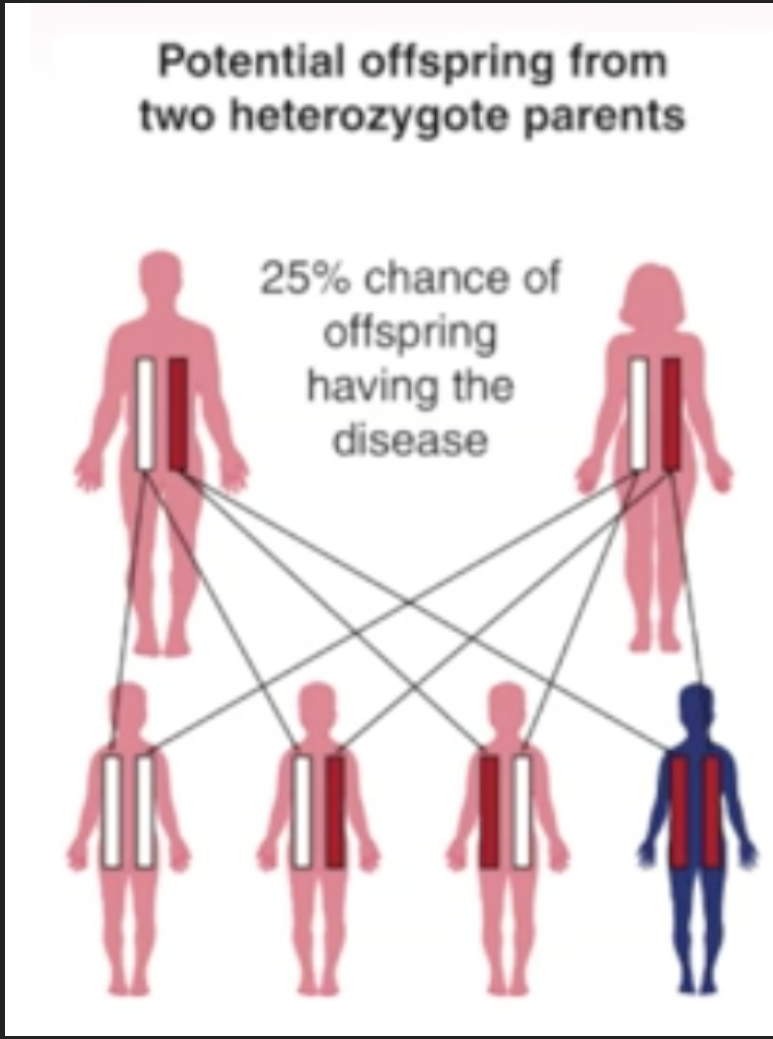
GENETIC CHANGES

RAB23 | MEGF8 ⋯⋯⋯⋯⋯⋯⋯⋯▶

- Production of proteins with little or no function
- Interference with normal body patterning

plays a role

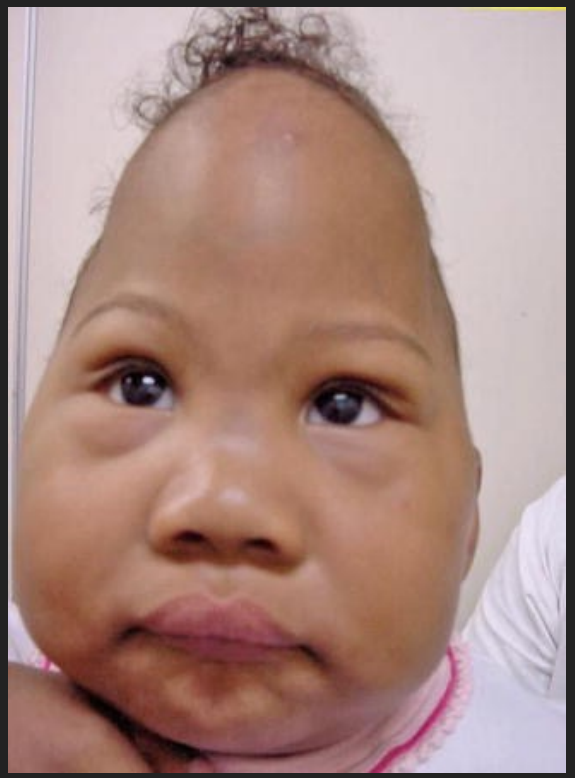
INHERITANCE



- AUTOSOMAL RECESSIVE
- Both copies of the gene in each cell have mutations.
- Parents of an individual with autosomal recessive condition each carry one cry of the mutated gene, but typically do not show signs and symptoms of the condition.

DIAGNOSIS

- Clinical manifestations: malformation of the skull.
- Genetics tests



TREATMENT

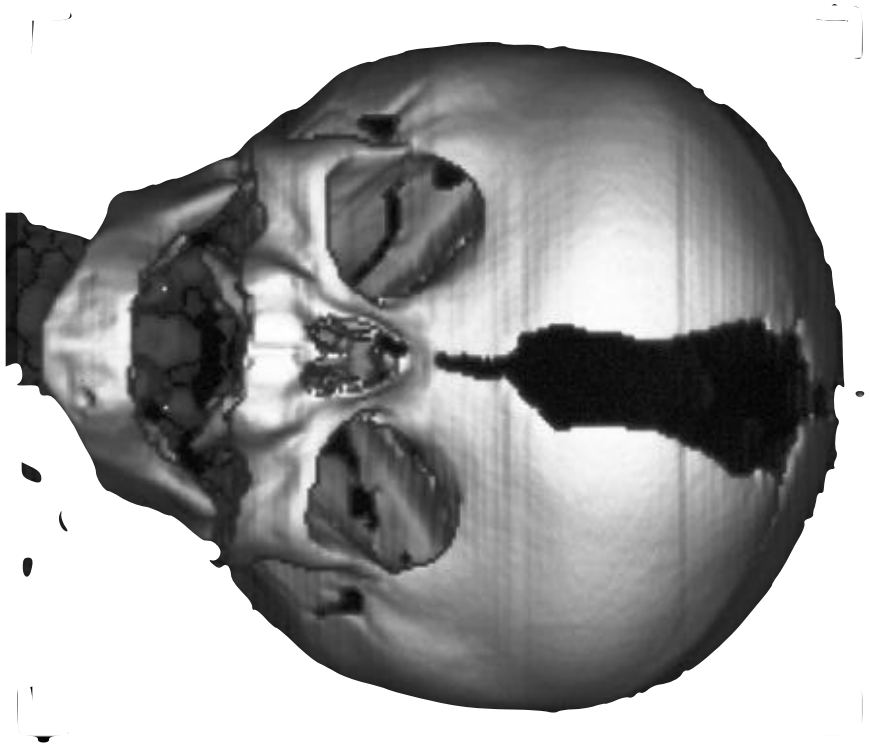
- Options to correct malformations of skull: within 1st year of infancy.
- Surgery to correct malformation of the heart
- Speech and occupational therapy
- Lifelong diet plan

Thank you

Crouzon Syndrome

by Dr. Jasveen Wadia
USC Advanced Periodontology Class of 2019

Background



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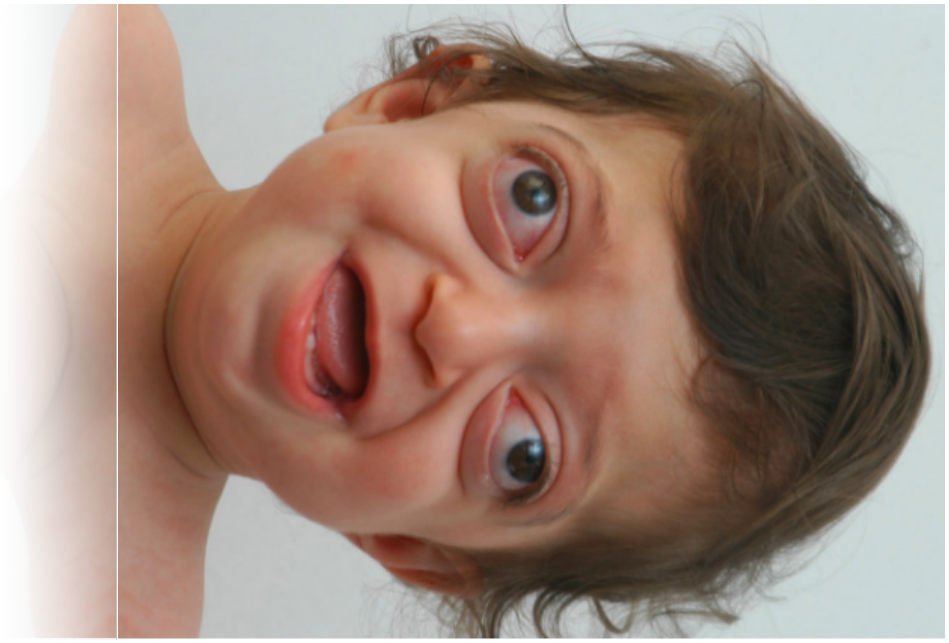

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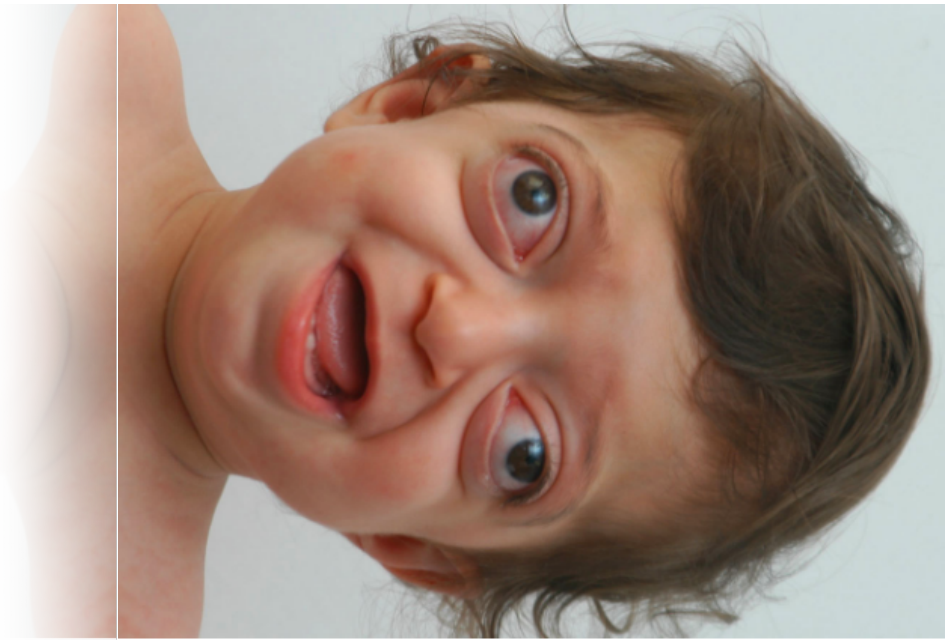

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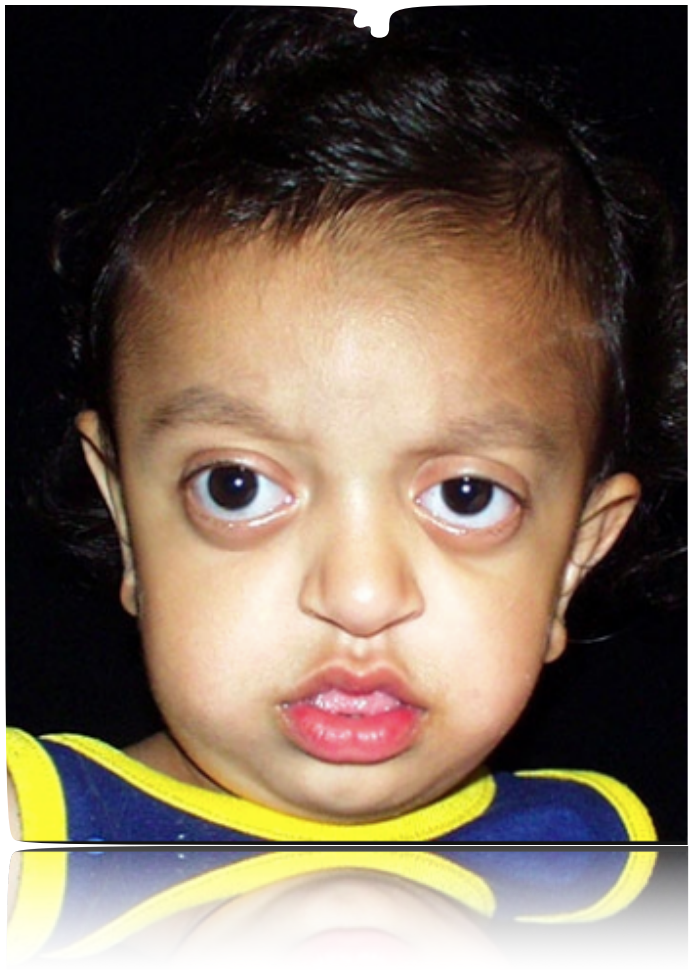

Genetics

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Genetics

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Diagnosis

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Dental Impact