A Case of SHORT STATURE with GENU RECURVATUM.

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Presenting Complaints

A 9 years old girl from Jabalpur 3rd born of non consanguineous marriage was referred to our hospital for cardiac surgery after completion of which child referred to our department for

- c/o Difficulty in walking .
- Failure of eruption of teeth of lower jaw.

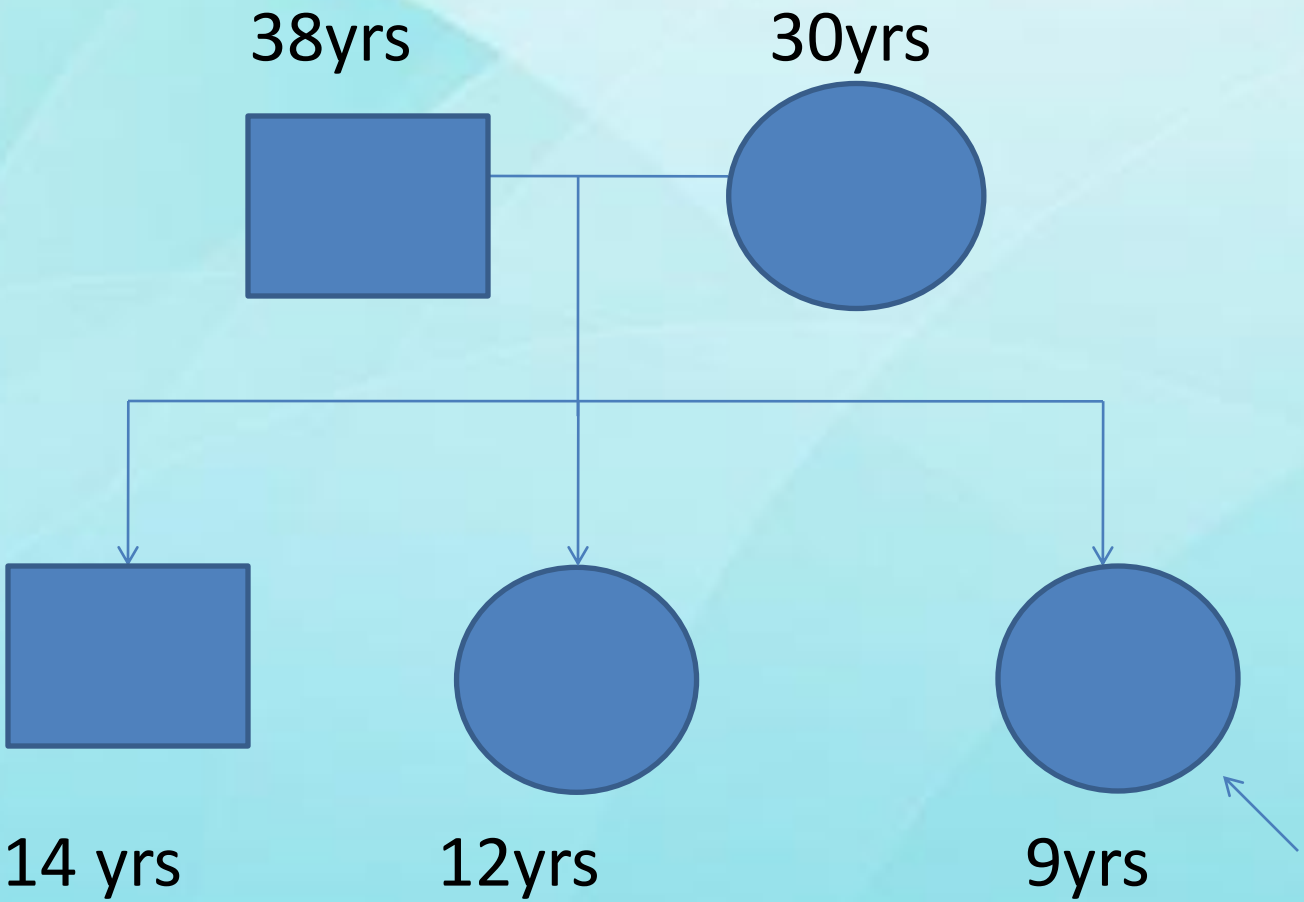
Elaboration Of Presenting Complaints

- Difficulty in walking associated with frequent falls and forward bending of legs . They took treatment in form of exercises but there was persistence of symptoms.

Birth History

- She was born at term, with birth weight of 1.4 kgs.
- Uncomplicated pregnancy and there was no exposure to any known teratogenic agent.
- At birth, it was noticed that her lower limbs were hyper-extended, touching her forehead.
- Echo done on d3 of life for breathlessness revealed a large sub aortic VSD, aortic over ride, severe right ventricular outlet obstruction.
- Total intra-cardiac repair of Tetralogy of Fallot was done at eight years of age.

Family history



No family h/o short stature, constitutional delay.

History

- Developmental milestones normal.
- Difficulty in speech.
- Good in studies, regular to school
- Appetite was normal
- Sleep pattern was normal
- Receiving adequate calories and proteins

On clinical examination

- Conscious, alert, active
- No pallor, icterus, clubbing, edema
- Vitals : Normal
- Anthropometry:

  Weight : 15kg (<3rd centile)

  Height : 115 cms (<3rd centile)
- Weight for Height - normal

On clinical examination

- Upper seg : lower seg ratio = 1:1.1(normal)
- Arm span : 112cms
- Midparental Ht : between 50-75 centile.

On Examination

On Examination

On examination

- Oral cavity – Sub mucosal cleft palate and Bifid uvula.
- Hearing – normal.
- Vision and Fundus – normal
- Tanner staging – Stage 1 (Preadolescent)
- Systemic examination : Normal

Skeletal System Examination

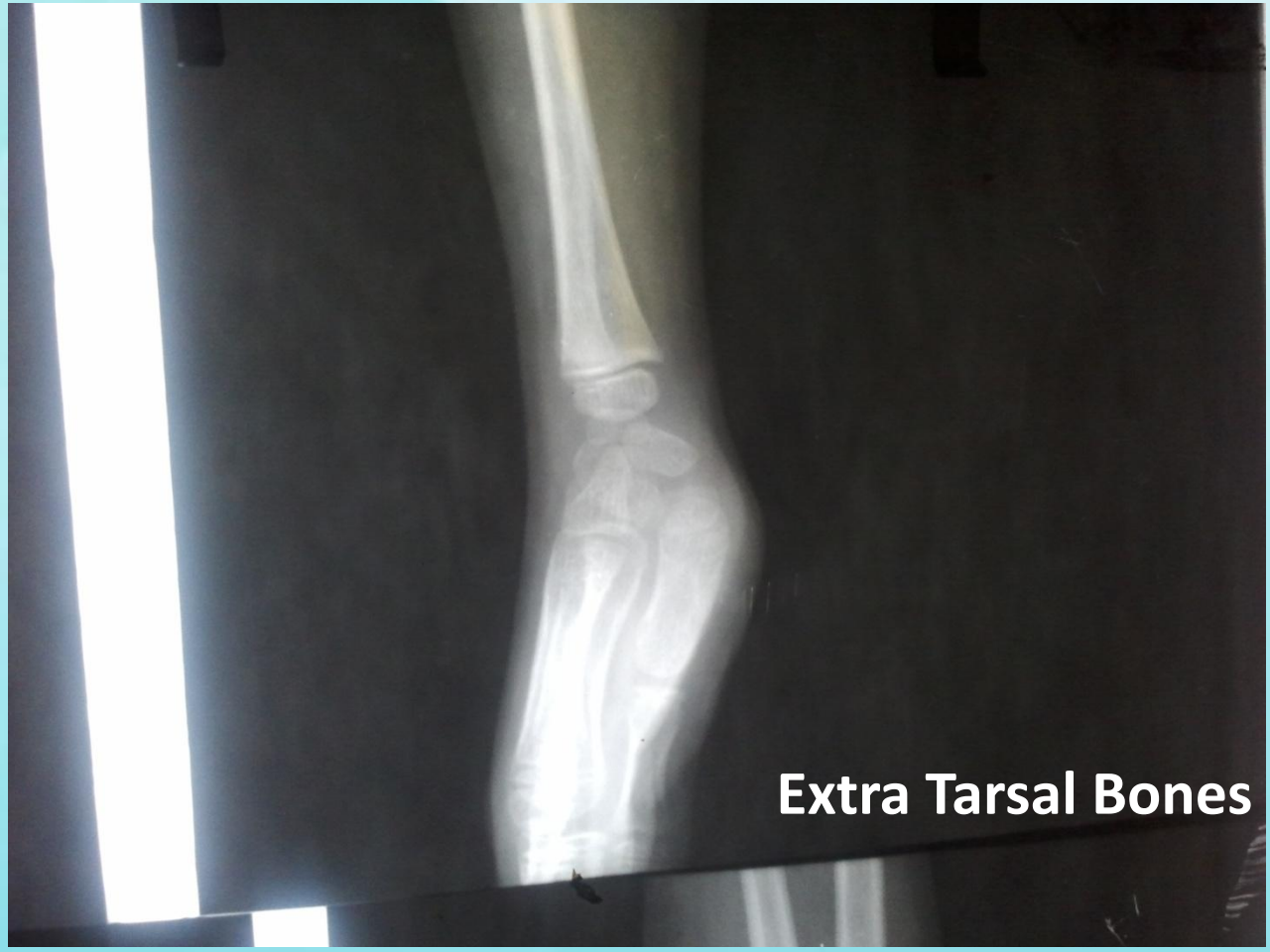
Skeletal system examination

- She had hypermobility at wrist, elbow, and ankle joints .
- Bilateral dislocation of knee joints with limited flexion at the knee joint.
- No kyphoscoliosis

Investigations

- She had spina bifida at S1.
- Ankle - accessory tarsal bones with atypical calcaneus.
- X-ray wrist – normal, no extra carpal bones.
- Chromosomal analysis revealed a normal female with 46XX (normal constitutional karyotype) (GTG band technique)
- Usg abdomen and pelvis normal.
- TFT normal.

X-Ray Findings



At present

- Cardiology status stable.
- Improvement in skeletal functions with regular physiotherapy and orthopaedic management.
- Attends school for her age with fair academic performance.
- Regular follow up.

Diagnosis

- Dysmorhic facial features- depressed nasal bridge,hypertelorism and micrognathia.
- Genu recurvatum, extra tarsal bones and short stature.
- Bifid uvula,submucosal cleft palate and missing central and lateral incissors in lower jaw.
- Heart defects.

DIAGNOSIS

LARSENS SYNDROME

DISCUSSION

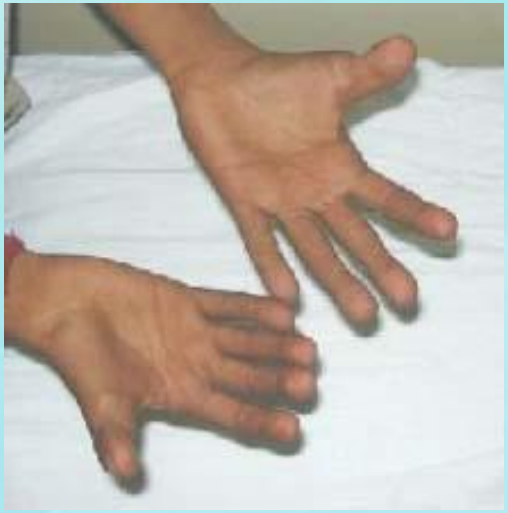
LARSEN Syndrome - Introduction

- Larsen syndrome is a rare genetic disorder.
- Incidence -1 in 100,000.
- Referred as skeletal dysplasia or hyper mobility syndrome.
- First reported by Larsen et al in 1950's on six sporadic cases.

Genetics

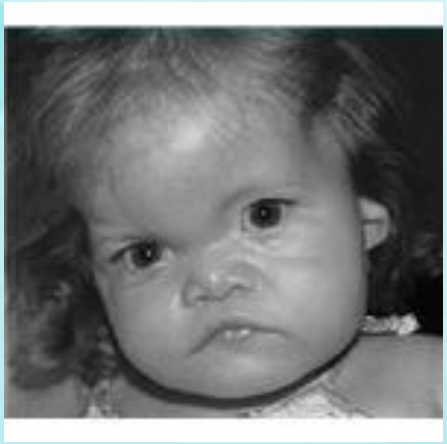
- Autosomal dominant or recessive. Most cases are dominant.
- Disorder of connective tisssue migration during fetal life.
- Missense mutations or small in frame deletions in FLNB gene (filamin B, beta gene) located on chromosome 3, cytogenetic location “3p14.3”.
- Gene FLNB encodes the cytoskeletal protein filamin B and mutations or deletions in it result in the severe bone and cartilage abnormalities.

Clinical Features



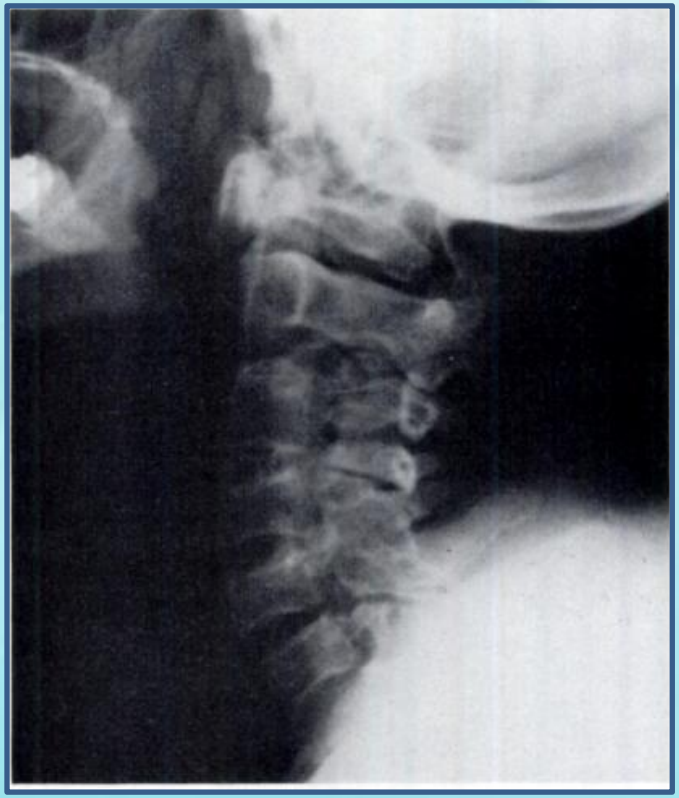
Joint hypermobility and multiple joint dislocations, especially of knees and feet.

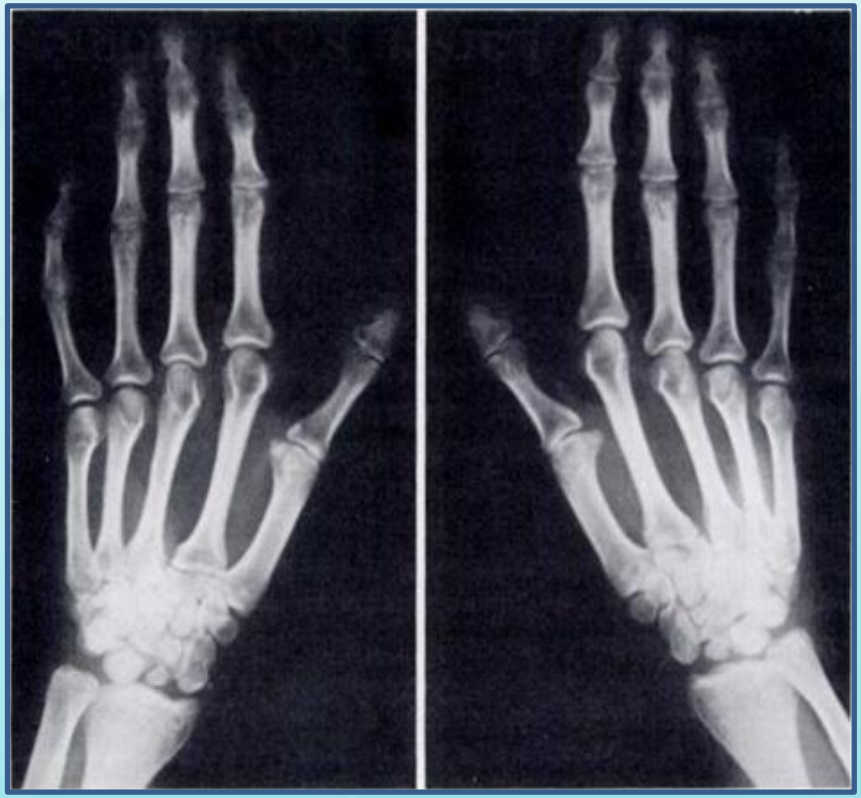
Clinical Features



Facial features include midfacial hypoplasia with a depressed nasal bridge , hypertelorism and prominent forehead.

Clinical features

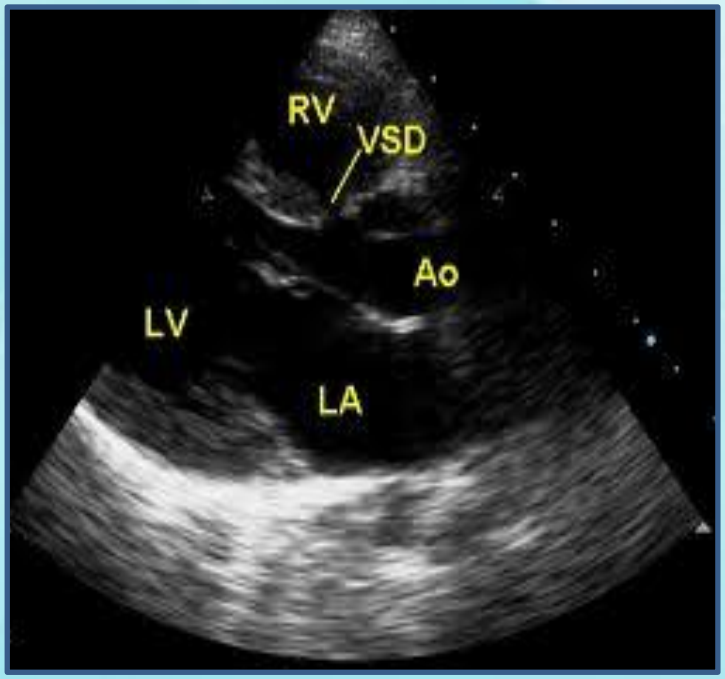




Failure of segmentation of posterior elements and cervical instability.

Numerous carpal or tarsal bones.

Clinical Features



Structural heart defects like VSD.
Cleft palate, cataracts and hearing abnormalities

Clinical features.

- Facial features include midfacial hypoplasia with a depressed nasal bridge , hypertelorism and prominent forehead.
- Joint hypermobility and multiple joint dislocations, especially of knees and feet.
- Bifid uvula,bifid tongue , cleft lip and cleft palate.
- Failure of segmentation of posterior elements and cervical instability,Numerous carpal or tarsal bones.
- Structural heart defects like VSD.
- Cataracts and hearing abnormalities.

Diagnosis

- Diagnosis- a step-wise process done by clinical examination , radiographic and facial anomalies initially.
- Confirmation - It is confirmed by carrying out FLNB molecular genetic testing, like DNA analysis using linkage analysis & sequencing or others.
- A karyotype test will reveal any abnormalities in the inherited chromosome 3.

Treatment

- Treatment depends on the symptoms shown by the individual patient.
- Joint defects, dislocations - physiotherapy, specialized casts and braces and in serious cases corrective surgery.
- Spinal issues- posterior arthrodesis.
- Myelopathic indications- anterior decompression and circumferential arthrodesis.
- Cleft palate - surgical interventions or speech therapy.
- Respiratory issues (caused by flaccid cartilage) - physiotherapy or tracheotomy, and depending on the severity, the use of a ventilator.

PROGNOSIS

- Lifespan is till adulthood, with normal intelligence.

- Once a child has been diagnosed with Larsen syndrome for effective management, lateral cervical spine tests, audiometric testing and the extent of hip dislocation are essential to allow the proper choice of surgery procedures.

REVIEW OF LITERATURE

- **Cervical Spine Involvement in Larsen's Syndrome: A Case Illustration Pediatrics 2003;111;199**

  Oakes and Paul A. Grabb.

  13 year old male presented with cervical instability and neurological symptoms and effectively managed with orthopedic procedures.

REVIEW OF LITERATURE

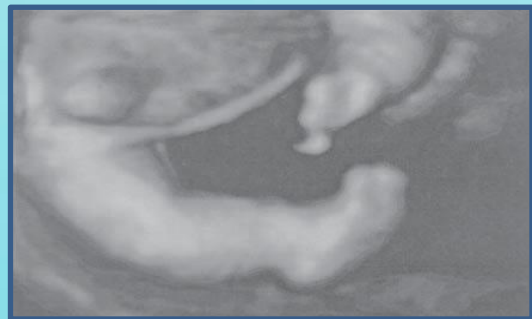
- **INDIAN PEDIATRICS VOLUME 45__SEPTEMBER 17, 2008**
- An eleven year old male child born of non consanguineous couple on examination had
- Short stature, flat facies, prominent forehead, depressed nasal bridge,hypertelorism , bilateral rhizomelic shortening of upper limbs, spatulate and dislocated thumbs *, bilateral elbow, ankle, and* hip dislocation **.**
- *Parents* showed no features of Larsen syndrome.

REVIEW OF LITERATURE

- **American Academy of Pediatric Dentistry-17:l, 1995**
- 29 year old Afro American female
- Intraoral examination revealed a permanent dentition with two mandibular central incisors missing.
- Full mouth radiographic examination revealed 40 to 95% alveolar bone loss associated with all the teeth, especially maxillary and mandibular incisors and periodontitis.

REVIEW OF LITERATURE

- **Indian Journal of Pediatrics, Volume 77—July, 2010.**
- A 30-year-old consanguineously married women with a pregnancy of 20 weeks of gestation, with history of previous child diagnosed as Larsen syndrome on the basis of characteristic clinical and radiological features.
- With this history Larsen syndrome was diagnosed antenatally in present pregnancy on the basis of multiple joint dislocations, resulting in abnormal position of legs and club feet, hypertelorism and depressed nasal bridge on ultrasonography .
- Antenatal genetic counseling was done and the couple decided to terminate the pregnancy. The abortus was sent for autopsy, and confirmed the diagnosis.



Take home message

- Larsen syndrome though rare, early clinical diagnosis needed for effective multidisciplinary management.

- Need to prevent occurrences in family by proper genetic counseling.

THANK YOU