A CASE OF POLAND SYNDROME

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- 14 yrs 5 mo old adolescent female child
- 1st born of 3rd degree consanguinous marriage presented with

  - H/o poor breast development Rt Side noticed since 3 yrs
  - Not attained Menarche
  - Not gaining height

- No History S/O any Chronic systemic illness
- No H/o Polyuria / Polydipsia
- No History S/o Hypothyroidism
- No H /o Headache / vomiting

- **Past History :**

  - Underwent surgery for rt hand syndactyly at 4 yrs of age at Govt.hospital

- **Birth History:**

  - Antenatal period : Uneventful
  - FT Forceps / B.Wt : 3.5 kg
  - Natal,Postnatal & Neonatal Period: uneventful
  - Short rt upper limb & syndactyly noticed at birth between II-III-IV fingers rt hand

- **Immunised for Age**

- **Family History:**
  - **3rd degree consanguinous marriage**
  - **Mother attained menarche at 14 yrs**
  - **Two Siblings – Female, younger**

    2nd – 13 yrs old – not attained menarche

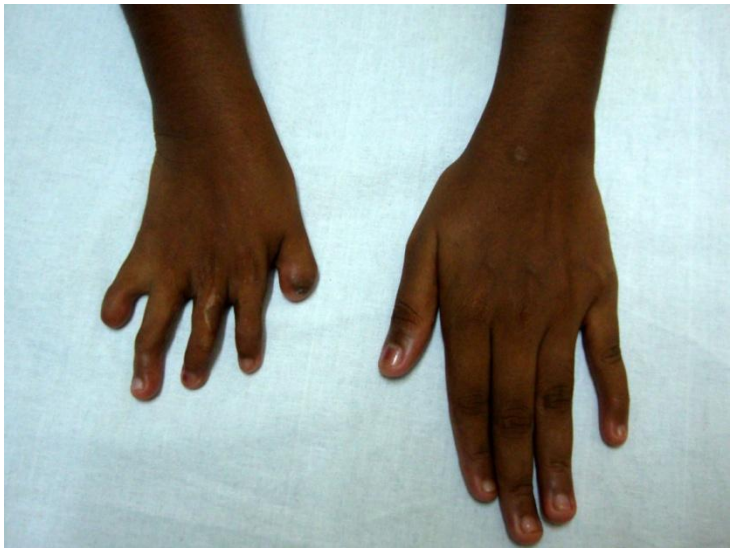
    3rd – 8 yrs old
- **Diet history:**

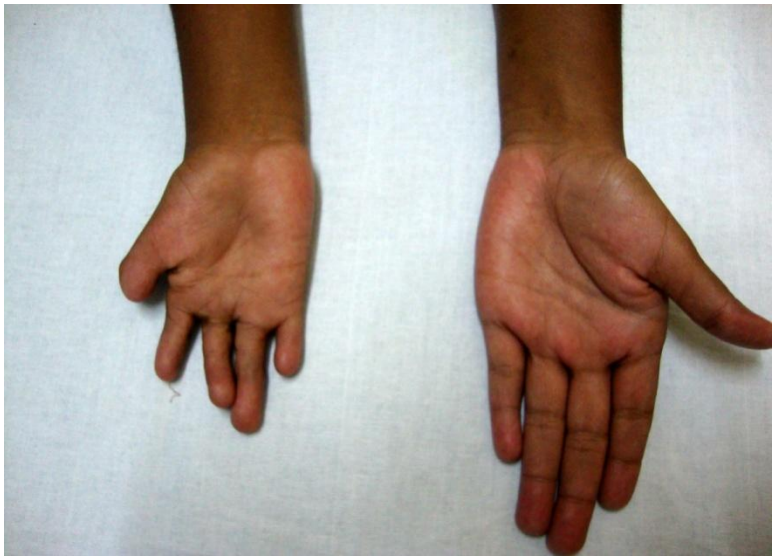
  -Adequate intake
- **Development History:**
  - **Appropriate for age**
  - **-Now Studying in Class X**
  - **Average in Studies**

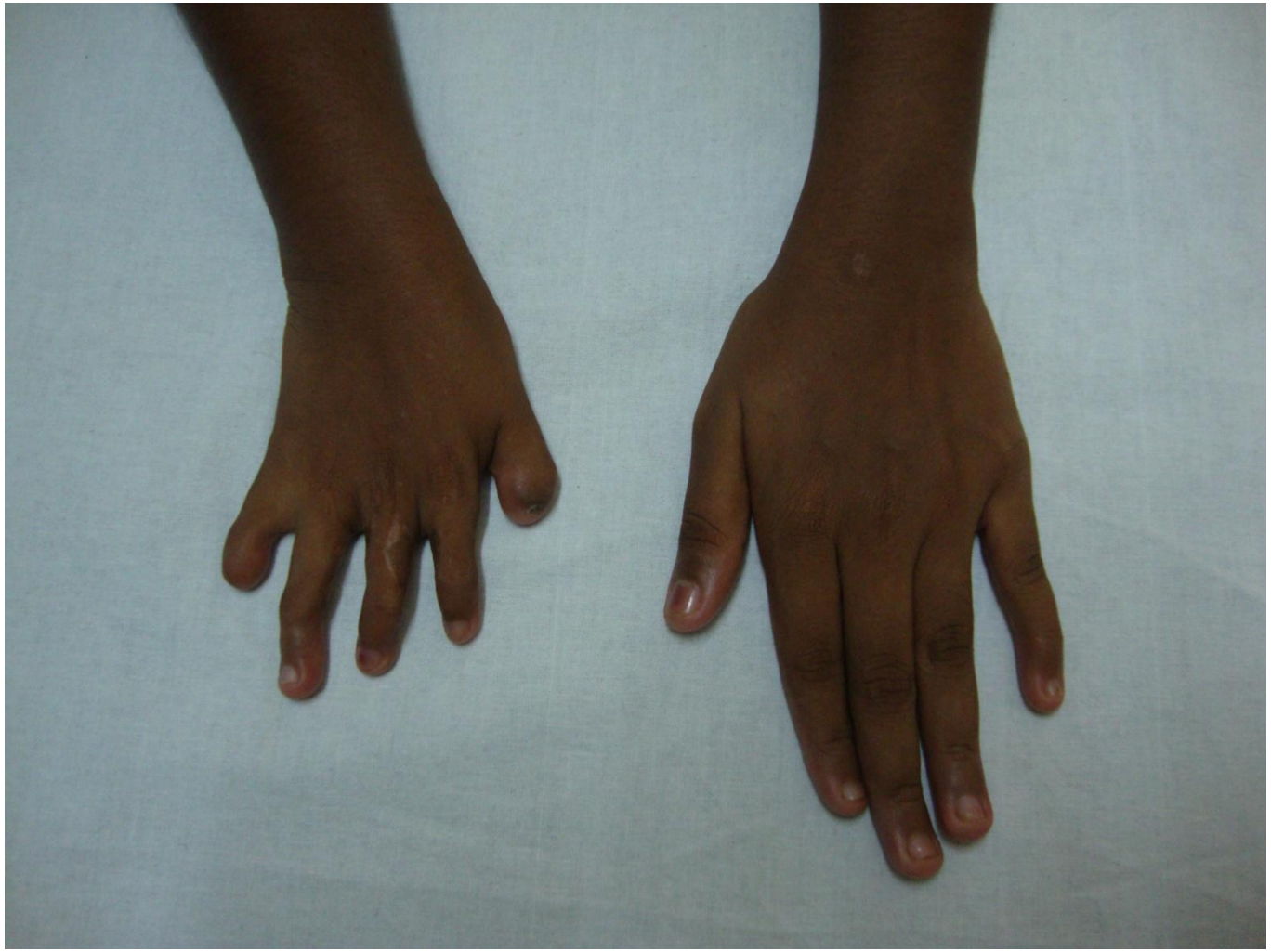
- **GENERAL EXAMINATION:**
  - Alert , Co-operative
  -Head,Hair,Eyes,Ear : Normal
  -Oral Cavity ,Palate : Normal
  -Neck , Skin : Normal

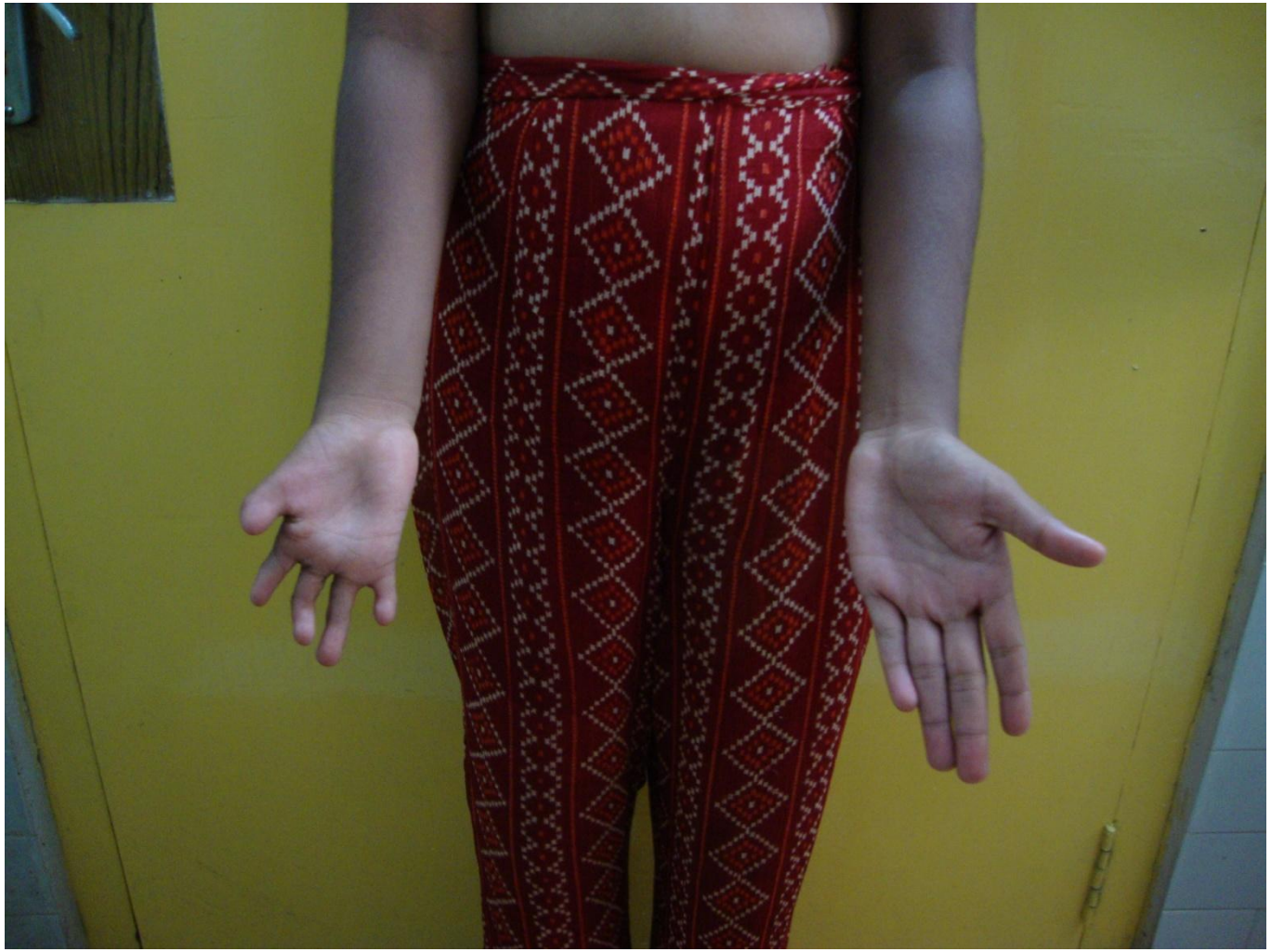
Rt Hand: Brachydactyly

Nails: Atrophic Nails in Rt hand









- **SMR STAGING:**

  Breast : RT – Stage I / LT - Stage III

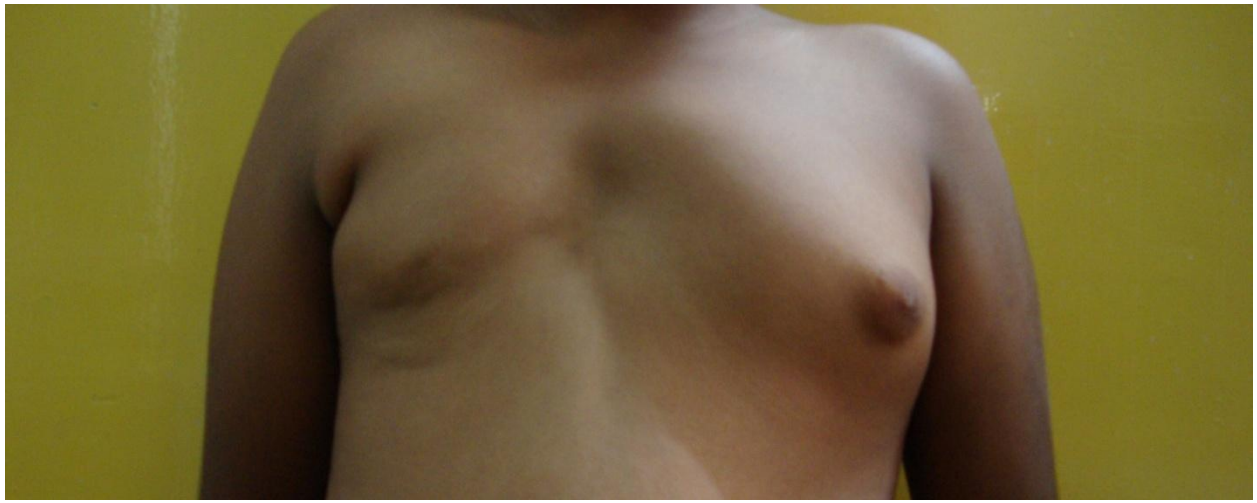
  Pubic Hair : Stage II

  Axillary Hair : Stage I

- **RT CHEST:**

  Amastia, Hypoplastic Areola & Nipple

  Absent Sternal Head of Pectoralis



- **Anthropometry :**

  HT: 144.5 cm [< 3rd centile]

  WT: 46.5 kg [50th - 75th centile]

  BMI: 23.06 [75th -85th centile]

  US / LS Ratio : 1 : 1

  MPH : 153.5 cm [< 3rd centile]

- VITALS : STABLE
- SYSTEMIC EXAMINATION : NAD

INVESTIGATIONS

- Sr.FSH - 5.37 MIU / ml [N]
- Sr. LH - 0.75 MIU / ml [N]
- Sr.Prolactin – 4.49 ng / ml [N]
- Growth Hormone study: Normal
- MRI Brain : Normal

- **THYROID PROFILE:**
  - **T3 : 2.1 nmol / L**
  - **T4 : 12.4 pmol / L**
  - **TSH : 1 micro units / ml**
- **USG PELVIS : PRE-PUBERTAL UTERUS**
- **CXR : Normal**
- **Bone Age : corresponds to chronological age**
- **Karyotyping : 46 XX**

FINAL DIAGNOSIS

- Based on clinical features of absent pectoralis sternal head,hypoplasia of right breast,upper limb asymmetry,syndactyly ,brachydactyly diagnosis of poland syndrome with familial short stature was made

MANAGEMENT

- Reassurance
- Rt Breast : Adv… Reconstructive Surgery
- Rt Hand : Physiotherapy

POLAND SYNDROME

- According to the National Human Genome Research Institute, Poland syndrome affects MALES THREE TIMES as often as females
- Affects the RIGHT SIDE of the body TWICE as often as the left.
- The incidence is estimated to range from one in 7,000 to one in 100,000 live births.

- Unilateral absence or hypoplasia of the pectoralis muscle, most frequently involving the sternocostal portion of the pectoralis major muscle

- Variable degree of ipsilateral hand and digit anomalies, including synbrachydactyly.

- Poland syndrome is most commonly a sporadic condition but familial cases have been reported.

ETIOLOGY

- Unknown

- The primary defect could be in the development of the PROXIMAL SUBCLAVIAN ARTERY with early deficit of blood flow to the distal limb and the pectoral region, resulting in partial loss of tissue in those regions

CLINICAL FEATURES

FREQUENT SIGNS

- **Absent pectoral muscles**
- **Brachydactyly ,Oligodactyly , Syndactyly**
- **Dextrocardia**
- **Diaphragmatic hernia /defect**
- **Humerus ,Radius , Ulna - absent/abnormal**
- **Liver /biliary tract anomalies**

- Rhizomelic micromelia (relatively shorter proximal segment of the limbs compared to the middle and the distal segments)
- Upper limb asymmetry
- Simian crease on affected side
- Hypoplastic /absent nipples
- Scapula anomaly

OCCASIONAL SIGNS

- Agenesis /hypoplasia of kidneys
- Abnormal morphology of hypothalamic-hypophyseal axis
- Abnormal function of hypothalamic-hypophyseal axis
- Microcephaly
- Preaxial polydactyly
- Ureteric anomalies (reflux/duplex system)
- Vertebral segmentation anomaly

NOTABLE PERSONALITIES

- British TV presenter Jeremy Beadle (1948-2008) Manifested in the form of his withered right hand.
- Olympic boxer Jérôme Thomas - left arm and hand are significantly shorter and smaller than his right. Thomas also lacks a left pectoral muscle.
- PGA golfer Bryce Molder- Absent left pectoral muscle and a small left hand. Several surgeries in his childhood repaired syndactyly on the left hand.

ASSOCIATED SYNDROMES

- Moebius syndrome and Poland anomaly. They suggested a common fetal mesodermal defect.
- C/F :dextrocardia, agenesis of the pectoralis muscle, aplasia of the abdominal muscles, mammary hypoplasia, agenesis of the nipple, and various hand and finger anomalies.

- Poland anomaly and the Adams-Oliver syndrome were coexistent.
- Result from the interruption of early embryonic blood supply in the subclavian arteries
- Autosomal dominant pattern of inheritance.
- Poloand anomaly and Goldenhar syndrome

- Klippel-Feil syndrome and Poland anomaly
- Typical triad of KFS, including very short neck, low occipital hairline, and reduced bilateral neck movements.
- Radiographic examination showed fusion of C1 and C2 vertebrae.
- Also had absence of the right pectoralis muscle, hypoplastic right breast, hypoplastic costochondral junctions, and hypoplastic sternum consistent with Poland anomaly.
- No cardiac, vascular, or renal anomalies.

LITERATURE REVIEW

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THANK YOU