CHONDRO ECTODERMAL DYSPLASIA

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Case history

- 8 years old
- born of 2nd degree consanguineous marriage male child
- admitted with complaints of fever- 3 days, high grade, associated with chills and rigor, Intermittent fever with no history of vomiting, loose stools, abdominal pain, or any swellings.
- Child was not admitted in the past for any other illness.
- Antenatal, birth and neonatal history was not significant.
- No other significant history
On Examination

General examination:
- Comfortable,
- no pallor/icterus/cyanosis/clubbing/pedal edema

Vitals
- PR – 102/min regular, good volume, felt in all peripheral pulses, no radio-radial/radio-femoral delay, no pulse deficit.
- RR- 24/min
- Bp- 100/70 mm hg
Examination

Anthropometry:
- Ht: 102 cms against 125 cms (81.6%)
- Wt 18.5 kg against 24 kg (77.08%)
- Hc-49 cms
- US : LS ratio- 1.25:1
- Arm span 97 cms.
- Defect in upper lip
- Dental anomalies - absence of both upper and lower incisors
- Short limbs
- Post axial polydactyl
- Short stubby fingers.
- Nails were hypoplastic
- P/a - spleen 3 cms below left costal margin. Firm.
- CVS - fixed split S2
- RS - NVBS, BAE
- CNS - NFND
Dental anomalies
Lip anomalies
Dysplastic nails and polydactyl
Short stubby fingers
Investigation

- CBC- normal
- QBC for MP – positive
- urine R/E- Normal
- X-Ray- chest- high clavicle,
  Lower limbs- shortening of tibia and fibula compared to femur, spur in tibia, widened tibial shaft.
  Wrist- fusion of capitates and hamate
  Skull- dentigerious cyst
- ECHO- ASD with cleft in anterior mitral leaflet
- USG abdomen- splenomegaly
Radiology - lower limbs
X-ray skull
Exostosis
Discussion

- Ellis and van-Creveld described in 1940

**Prevalence:** occurs 1 in 60000-200000 live births.
- Most common in Amish community in Lancaster County, Pennsylvania.
- It's fairly very rare in general population

**Genetics:** autosomal recessive trait
- Mutation have been identified in one of the two genes, EVC and EVC2 which may be very close to each other on chromosome 4p
- Function of this gene products is unknown.
Discussion

Growth: short stature of prenatal onset.

Skeletal:

- disproportionate, irregular short extremities, polydactyl of the fingers occasionally toes,
- small broad middle phalanges and hypoplastic distal phalanges,
- malformed carpals, fusion of capitate and hamate, extra carpal bones
- narrow thorax with short poorly developed ribs
- hypoplasia of upper lateral tibia with knock knees
- pelvic dysplasia with low iliac wings and spur like projection at med and lat aspects of acetabula.
- exostosis projecting distally from medial aspect of proximal tibial metaphysis
Discussion

- **Nails**: hypoplastic

- **Teeth**: neonatal teeth, partial anodontia, small teeth

- **Mouth**: defects in alveolar ridge with accessory frenula

- **Cardiac**: approx 60% have cardiac defects, m/c ASD often with single atrium
Prognosis

- About 30% of patients die of cardiac or respiratory problems during infancy
- Life span is otherwise normal.
- Adult height ranges from 109-152 cms
References

- Smith book on congenital anomalies
- Caffey ‘s text book of radiology

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Thank you