A CASE OF RICKETS - RARE ASSOCIATION.

I.S.P, Stanley medical college.
Dr. Chandan Kumar k m (DCH).
Dr. Ekambarnath .MD.
Dr. Amudha Rajeshwari .MD.
Dr. Sujatha Sridharan. MD.
Dr. Karunakaran .MD.
Case report

A 2yrs female child with C/o

- Fever, cough, cold - 1 week.
- Difficulty in breathing - 4 days.

Relevant history:

- Non consanguineous marriage,
- Diagnosed to have skin disorder at birth,
  Vesico bullous lesions with erythematous base, membrane covering all over the body.
  became scaly, hyper pigmented by 3-4 months.
H/o On and off blistering present.
• No positive family history.
• H/o recurrent cold, cough, wheezing attacks since 6 months of age.
• H/o Refusal to stand with excessive crying due to leg pain since last 6 months.
• H/o contact with TB + *uncle completed ATT 6mo prior.
• Not on any long term medications except liquid paraffin external application.
Developmental history

Developmental delay +.

Dietic history.

- Exclusively breast fed for 4-5mons, Complimentary feeding started from 5month.
- Mixed diet.
- Caloric deficit of 380cal.
- Protein deficit of 4 g.
On examination

- Afebrile.
- Signs of Respiratory distress +.
- Generalized scaling, erythema, hyperpigmentation +.
- Large, thick, ridge like scaling around wrist, elbow, ankle, back +.
- Few vesicles, bullae seen over back +.
- Frontal bossing, rachitic rosary, Harrison groove, protruding abdomen, wrist widening +.
- Generalized hypotonia +.
**Anthropometry**

- **Wt**: 7.1kgs (expected- 12kgs) **Grade 3 PEM (IAP)**.
- **Length**: 72cms (expected- 89cms) **Grade 3 stunting (Water low's)**.
Systemic examination

- CVS- WNL.
- RS- Bil. wheeze+, creptations +
- P/A- Soft, liver 2cms below RCM.
- CNS- NAD.
**Investigations**

- CBC-WNL.
- PS study-WNL
- RFT-WNL.
- LFT-WNL.
- Sr. calcium-8.7 mg/dl (N-9to11mg/dl).
- Sr. phosphorus-3.3 mg/dl (N-3.8to6.5mg/dl).
- Alk pho-601 u/l (N-145to200u/l).
- Urine and stool examination-N.
- Mx-negative.
- RGJ-negative.
CXR- Bil. hyper inflation with infiltrates.

Widening of anterior end of ribs.
• Multiple #s of both bones of Rt. Forearm with minimal callus.
• Widening, cupping and fraying of metaphysis.
• X-ray Spine-N, X-ray LL-no #’s.
- **Hearing and vision assessment normal.**

- **Orthopedic opinion**- Undisplaced Multiple # of both bones of forearm with minimal callus, Nil surgical intervention.

- **Dermatology opinion**- Epidermolytic hyperkeratosis ichthyosis.
Skin biopsy report:

- Squamous epithelium with acanthosis with marked hyperkeratosis with hypergranulosis with parakeratosis, sub epithelium appears normal.
- Impression – Epidermolytic hyperkeratosis Ichthyosis.

**FINAL DIAGNOSIS**

**EPIDERMOLYTIC HYPERKERATOSIS Ichthyosis with Vit D Deficiency RICKETS with Pathological #’s with WALRI with PEM Grade 3.**
**TREATMENT**

- **SUPPORTIVE**: nasal O₂, IVF, Antipyretics, bronchodilators.
- **SPECIFIC**: IV antibiotics and steroids.
- **Vit D**: 6L IU IM [stoss regimen] followed by oral Vit D₄₀₀ IU/d.
- Ca, P. supplementation.
- Nutritional supplementation.
- Liquid paraffin local application.
Rpt. investigations after Rx

- Sr. Ca-9.3mg/dl, Sr. P-4.4mg/dl, SAP-201u/l.
- Radiological sign of healing.
The word ichthyosis = ‘fish skin disease’
Epidermolytic hyperkeratosis AD inheritance, up to 50% sporadic (spontaneous mutation).
Prevalence -1 in 1L-2L.
Onset at birth with generalized blistering, peeling with redness. With time hyperkeratosis, hyperpigmentation develop. Prominent over flexural, cubital, popliteal fossa, & axilla.
Variable involvement of palm and sole.
Nail/hair/mucosa not involved.
Underlying defect of keratin synthesis or degradation defect involving keratin 1, 2e and 10 with gene clustering on chr12p and 17q.

- The association of rickets with EHK is due to decreased synthesis of Vit D
  - hyperkeratosis of Horney layer.
  - increased amount of melanin lead to decreased penetration of UV rays.
Rickets commonly reported in Lamellar Ichthyosis & X linked Ichthyosis,

1st case of rickets with Epidermolytic hyperkeratosis Ichthyosis reported in 2004.
Fractures in Rickets

- Pathological #'s & Epiphyseal displacement can occur.
- Green stick #'s are most common.
- Commonly seen in mobile infants & toddlers.
- Patho physiology-Abnormality in arrangement of biochemically inferior collagen fibers. Osteoclastic resorption of uncalcified osteoid does not occur.
- Rx - Conservative management with plaster cast/splint along with Vit D, Ca & P supplementation.
Literature review

The 1\textsuperscript{st} case of rickets wit EHK reported in 2004 from dept of dermatology, university of Bari, Italy.

- Mother with features of EHK
- Child diagnosed with EHK at birth.
- Recurrent episodes of wheeze since 1yr.
- By 3yr complained of bone pain with rachitic features.
- Radiological and lab. features of Vit D deficiency rickets.
Investigations after sun exposure.

No significant increase in Vit D level.
Rickets in this child was due to defective synthesis of Vit D secondary to skin condition. Responded to Vit D treatment.
• Dept of dermatology, M.K.C.G. medical college, Orrisa.

• A 6yrs old child with EHK with radiological and laboratory features of rickets. Responded to Vit D treatment.

• Dept of dermatology, AIIMS, Delhi.

• 5 children with ichthyosis, 4 with lamellar ichthyosis and 1 with EHK, all had radiological and laboratory features of rickets, responded to Vit D treatment.
Take home message.....

- In severe ichthyosis with pigmented skin, *evaluation done for Rickets*, especially in developing countries where there is a background of Vit D deficiency. They need *life long Vit D supplementation*, as dietary intake alone not sufficient to prevent Rickets.

- Rickets to be thought of in DD of #’s, in Children with Non-accidental trauma & atraumatic limb pain as Rx of Rickets is vital for healing of #’s.
References...

- Rockwood and Wilkin’s -Fractures in children.
- Tachdjian’s pediatric orthopedics.
Thank you........