Epidermolytic Hyperkeratosis With Hypogonadism And Growth Retardation

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Presenting complaints

- 30 year old male patient
- Haryana
- Itchy skin lesions x birth
- Short stature

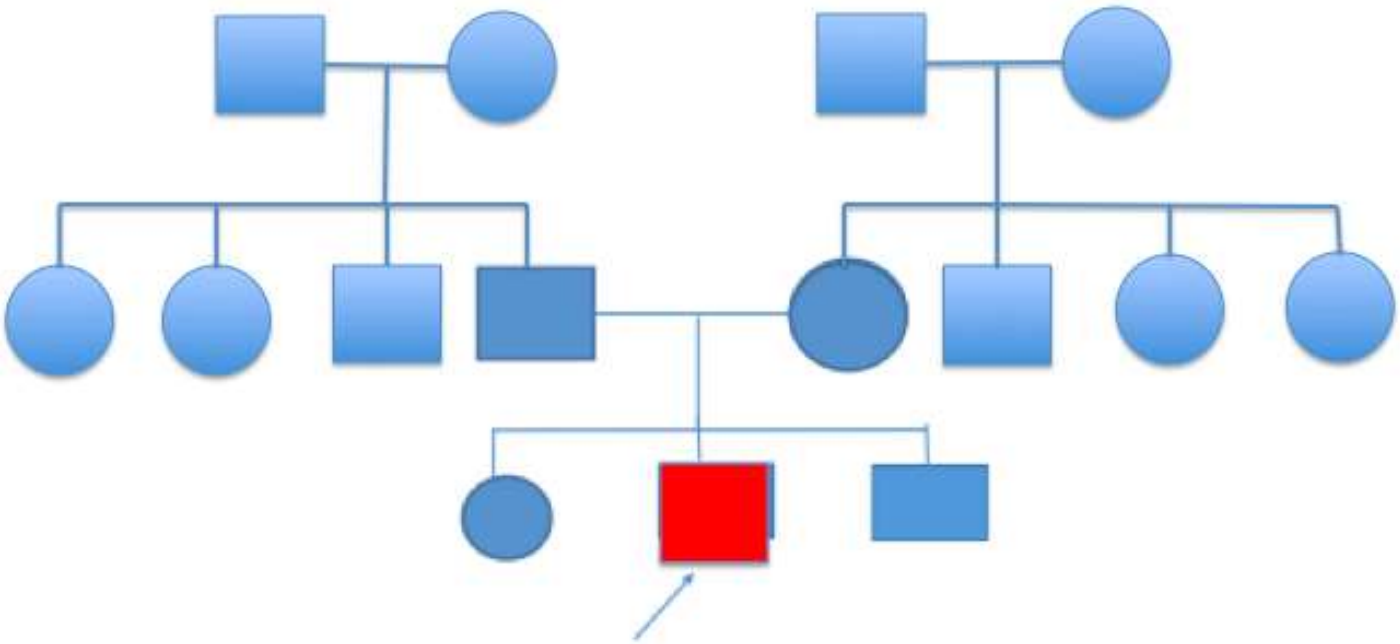
- h/o fluid filled blisters in childhood → (f/b) redness and peeling

↓

Lichenified, verrucous plaques (adult)

- No h/o collodion baby
- H/o thickened palms and soles
- H/o atopy- present
- H/o failure to gain height as compared to his peer group since 11 years of age
- a/w small sized testis and failure of change in voice during puberty
- No growth of axillary and pubic hair
- No h/o feeding difficulties, failur to thrive

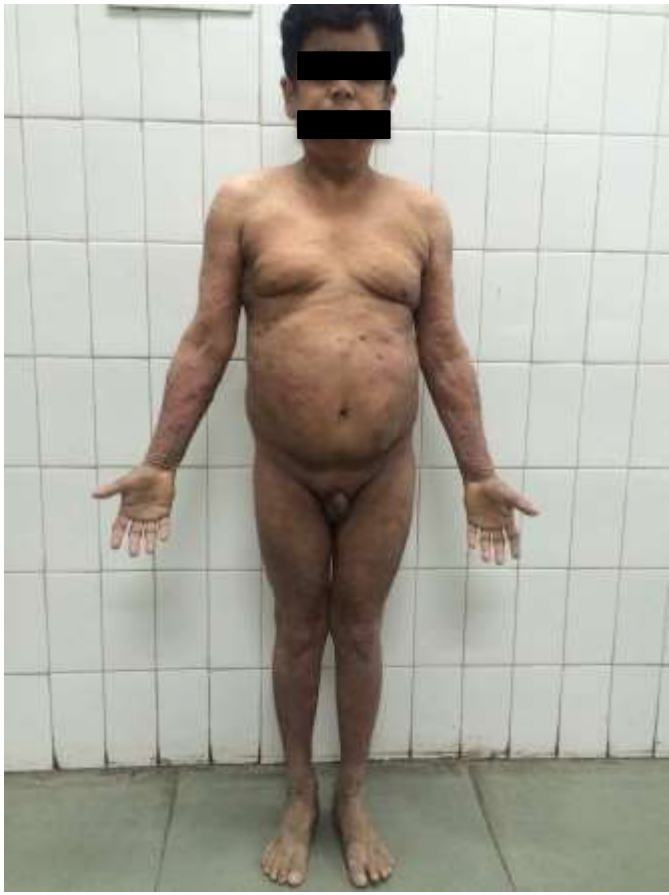
- **Past history-** not significant
- **Developmental milestones-** were normal.
- **Birth history-** non-consanguineous marriage; healthy parents; normal vaginal delivery; normal siblings.
- **Family history-**



- **Psychosocial history**- not significant
- **Treatment history-** Retinoids

  - Oral steroids- local practitioner intermittently

EXAMINATION



Anthropometry-

- Height: 130 cm (<5th percentile)
- US:LS= 1:1 (Proportionate)
- Weight: 45 kg

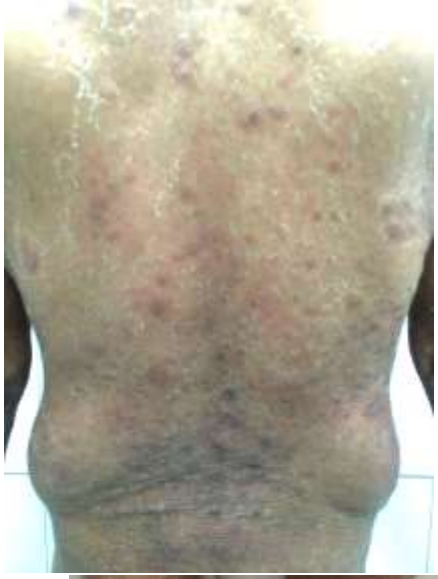
Vitals – stable

Secondary sexual characters-

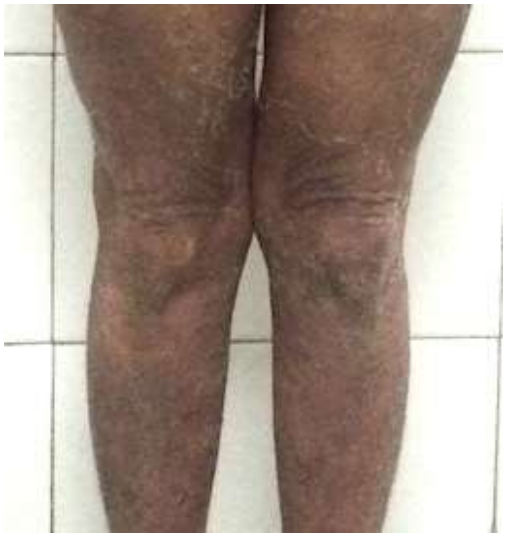
- Axillary, pubic hair- absent
- High pitched voice
- Testicular volume ~ 4ml (smaller)
- Stretched penile length was 3.5cm (smaller).

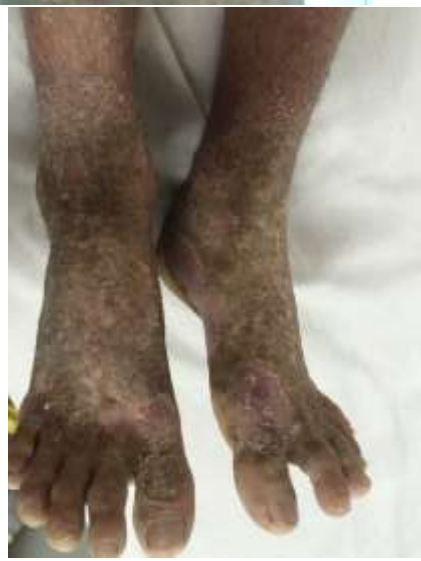
Cushingoid features- present

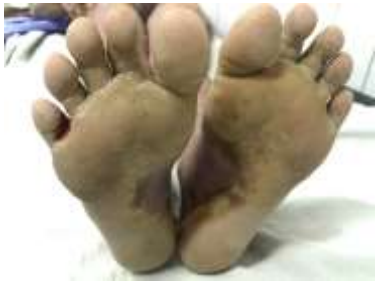
Cutaneous Examination

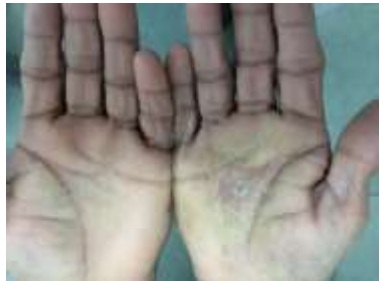


- Generalized involvement
- Erythematous, lichenified to hyperkeratotic plaques
- Scales- adherent scaling at few places, predominantly around extensors.
- Diffuse erythema & exfoliation over the trunk and lower limbs.
- Palmoplantar karatoderma- (+)
- Mucosa, teeth, nail and hair- (-)









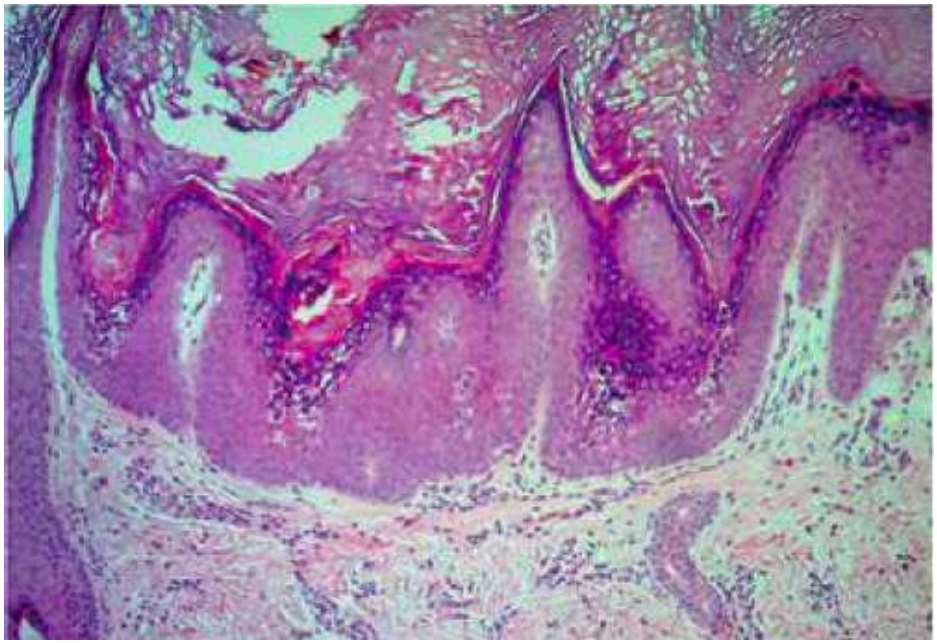
DIAGNOSIS

EPIDERMOLYTIC HYPERKERATOSIS WITH SECONDARY GROWTH FAILURE

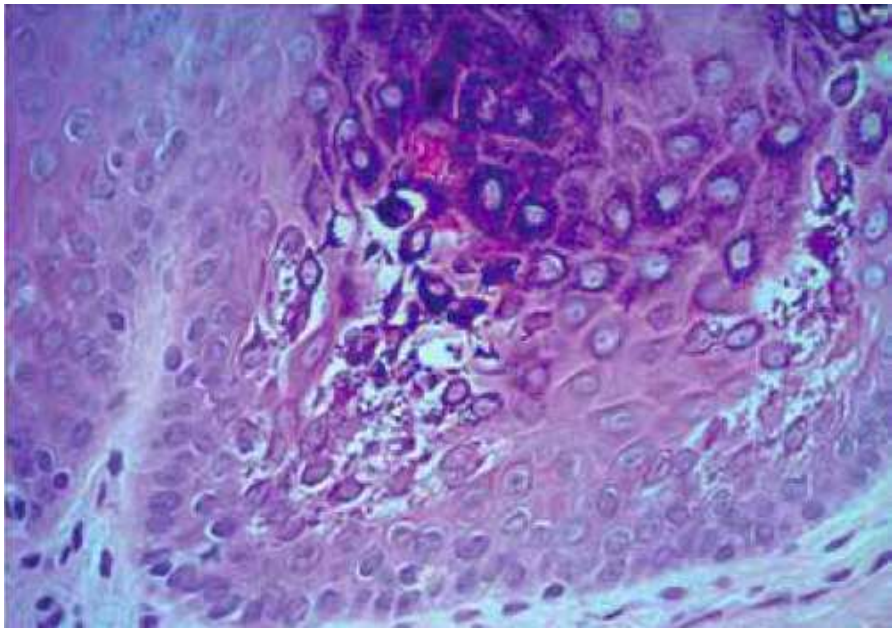
Cause- ???Endocrine disorder

Iatrogenic short stature

Histopathology



H&E stain,100X (magnification).
Epidermis- hyperkeratosis, papillomatosis and hypergranulosis.
Vacuolated keratinocytes (+)
Dermis- moderate lymphomononuclear cell infiltrate



H&E stain,400X (magnification).
Keratohyalin granules in vacuolated keratinocytes

INVESTIGATIONS

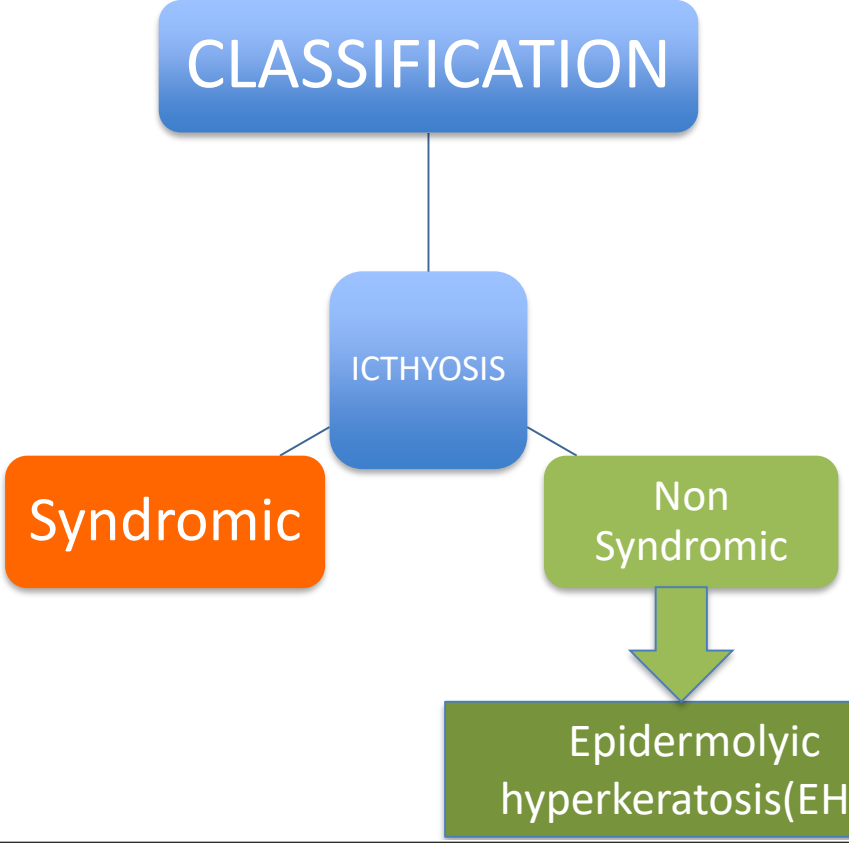
- Intelligence quotient- WNL
- LH- N
- FSH-N
- Testosterone- 155.46 (241-827 ng/dl) **L**
- Prolactin-N
- Growth hormone- 0.95ng/ml (N)
- Morning cortisol levels- <0.50 µg/ml. **L**
- T3, T4, TSH- WNL
- cHmg, RFT, LFT, FBS, RBS- WNL
- Sperm analysis- Azoospermia

DIAGNOSIS

- EPIDERMOLYTIC HYPERKERATOSIS with Hypogonadism (SECONDARY) and Iatrogenic Cushing's Syndrome.

DISCUSSION

- Epidermolyic hyperkeratosis(EHK)- Epidermolytic Ichthyosis or Bullous Congenital Ichthyosiform Erythroderma- clinically heterogeneous **disorder of cornification.**
- Rare Genodermatosis
- Prevalence- 1:100,000 to 1:300,000.
- M=F
- Inheritance- autosomal dominant (most common)
- Rarely- recessive or semi-dominant
- SPORADIC CASES- spontaneous mutations 50% cases.
- Mutation - KRT1 and KRT10→ suprabasal keratins, keratin 1 and keratin 10, respectively.



Although rare, Growth failure reported- severe form of EHK.

	NPS - 1 N 11	NPS - 2 N7	NPS - 3 N5	PS - 1 N25	PS - 2 N3	PS - 3 N1
Plamar-plantar hyperkeratosis	-	-	-	+	+	+
Palmar-plantar surface	Normal	Normal	Superlinear, minimal flaking	Flat	Flat	Cerebriform
Digital contractures	-	-	-	-	+	-
Flaking	Hystrix	Brown	Thin, white	Soft	White, rough	Brownish
Distribution	General	General	General	Local	General	General
Erythroderma	-	-	+	-	+	-
Blisters	+	+	+	Local	+	Neonatal

J Invest Dermatol. 2013;133(11):2626–8.

- Clinical spectrum- Broad

- Rarer variants of EHK- annular and nevoid form
- Hypogonadism is not common for EHK

- A single case report of EI with hypogonadism by Das et al has been found.

Indian J Dermatol Venereol Leprol 2007;73:280.

CONCLUSION

- Non-syndromic forms of icthyosis may also rarely present with underlying systemic abnormalities and is worth investigating as per the clinical suspicion of the treating physician.
- Early intervention in the form of hormone supplementation (pediatric endocrinologist consultation) in such patients may lead to better quality of life.

References

1. Nousbeck J, Padalon-Brauch G, Fuchs-Telem D, et al. Semidominant inheritance in epidermolytic ichthyosis. J Invest Dermatol. 2013;133(11):2626–8.
2. Das S, Roy AK, Kar C, Maiti A. Epidermolytic hyperkeratosis with a rare digital contracture. Indian J Dermatol Venereol Leprol 2007;73:280.

THANK YOU