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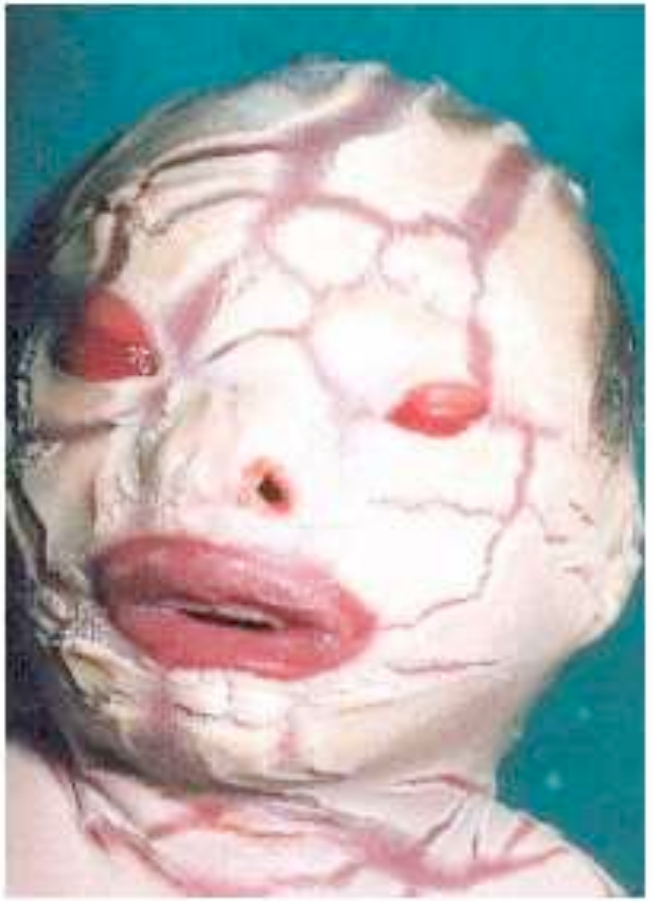
Genomics & Medicine
January 21, 2009

Ichthyosis, Harlequin Type

Introduction

- *Very* rare disease – 1 in 300,000 are affected
  - Autosomal recessive disease
  - Usually because of consanguinity
- Earliest report of harlequin ichthyosis by Reverend Oliver Hart (Charleston, South Carolina, 1750-colonial times)



Classical Diagnostic Methods

- Born prematurely
- Encased in armor-like shell
- Between plates are deep fissures where skin is cracked
- Underdeveloped ears
- Flattened Nose
- Wide lips
- Prenatal diagnosis of harlequin fetus by skin biopsies in the 22nd week of gestation

Classical Treatments

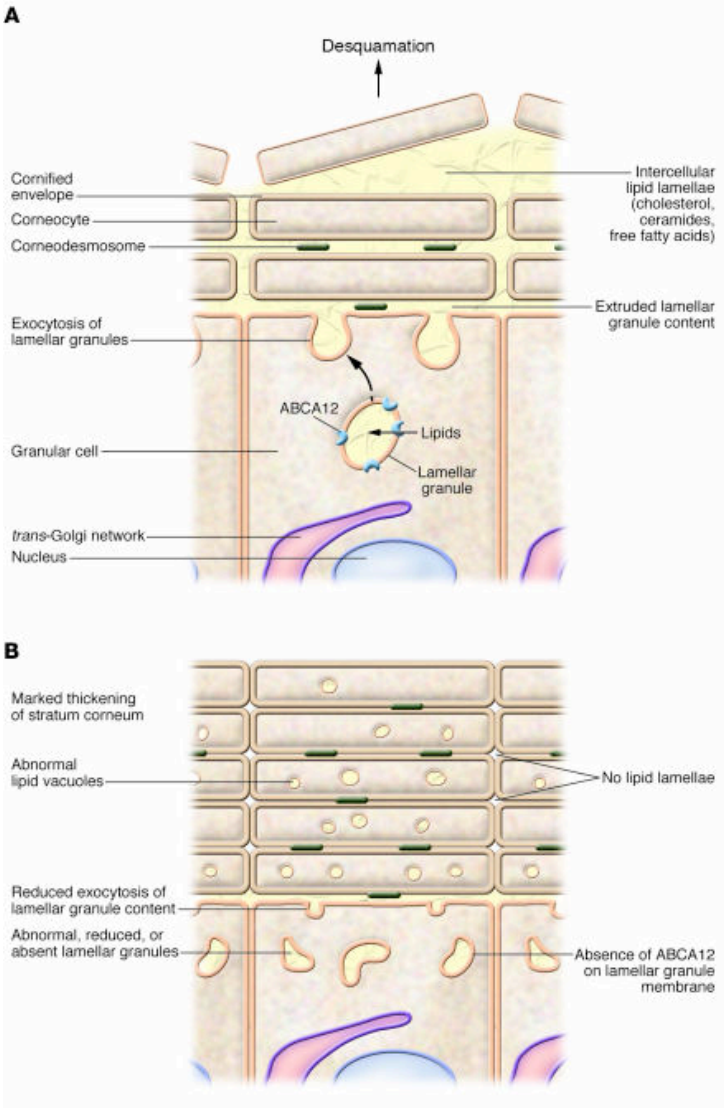
- Very high intake of protein
- Moist environment
- Petroleum-based creams/ointments
- Alpha-hydroxy acid or urea preparations to promote peeling and thinning of skin
- Prevention of infection and dehydration
- Release of collodion membrane on digits and around the chest area

Novel Diagnostic Methods

- HI patients have mutations in ABCA12, (region 2q34, chromosome II)
  - Heterozygous and homozygous for mutation => HI
- Analysis of extracted DNA

Novel Treatments

- Genetic correction of ABCA12 deficiency by gene transfer in patients' keratinocytes restored lamellar granule formation and lipid secretion
- Still searching for specific therapeutic approaches
  - Systemic administration of functional peptides with ABCA12-like properties
  - ABCA12 gene delivery approaches prior to or after birth



References

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