Gaucher disease with severe congenital Ichthyosiform erythroderma in two siblings

Patient 1: A three-year-old child presented with peeling skin for 1-month of age

H/o fever, seizures , loose stools, decreased feeding, frequent hospitalisations, multiple blood transfusions and failure to thrive
H/o delayed milestones
No h/o collodion membrane at birth and consanguinity in parents

On general examination: pallor, coarse facies, microcephaly, stunting

Skin: Ichthyosiform scaling, generalised desquamation and fissuring of skin over the body

Nails: thickened dystrophic

Oral and genital mucosae- normal

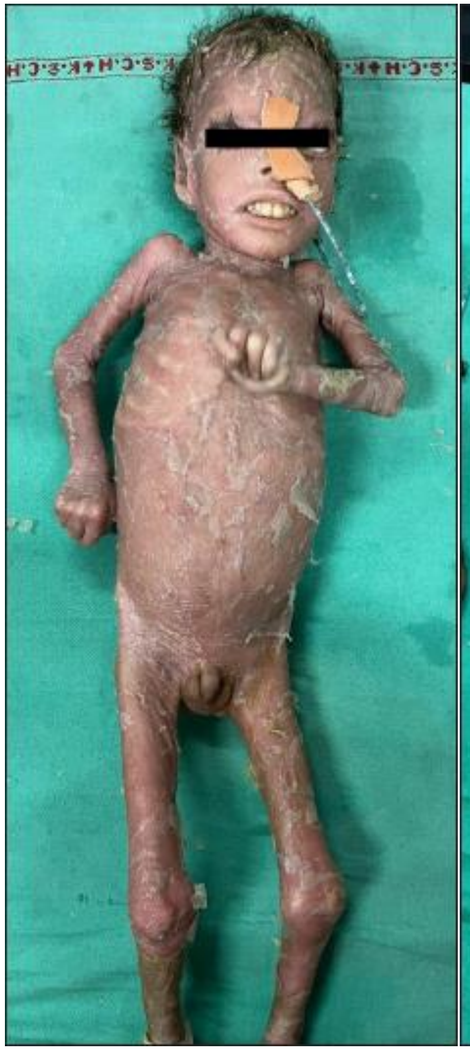
Opthalmic examination: ectropion , bilateral corneal opacities

Musculoskeletal examination: arthrogryposis, hypotonia, spasticity

MRI Brain :sub acute haemorrhage in both ventricles, pontocerebellar hypoplasia

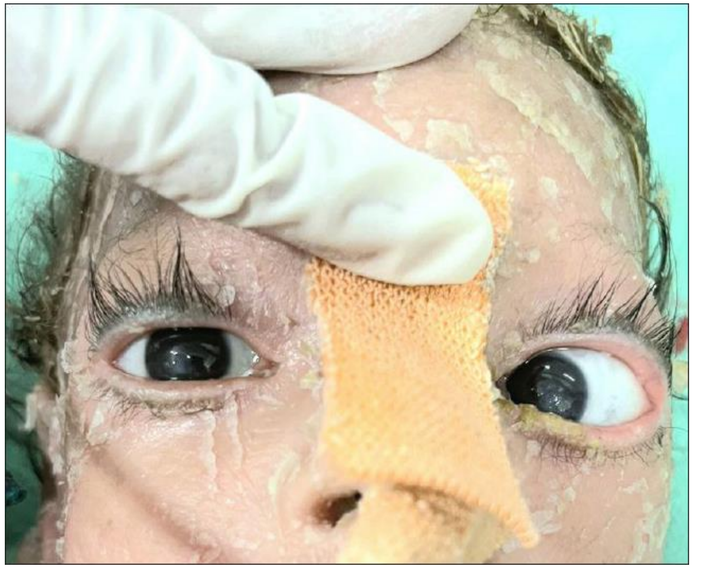
Histopathology: Epidermis shows lamellar hyperkeratosis, irregular acanthosis. Dermis shows perivascular lymphocytic infiltrate with melanin incontinence

Genomic sequencing : heterogenous missense variation in exo 10 of the glucocerebrosidase gene (substitution from leucine to proline at codon 483) suggestive of gaucher disease

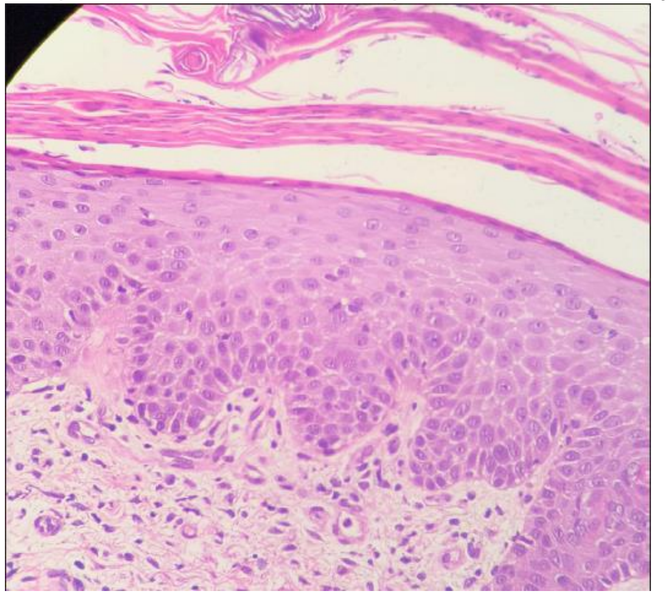


Generalised ichthyosis with desquamation and fissuring and arthrogryposis.

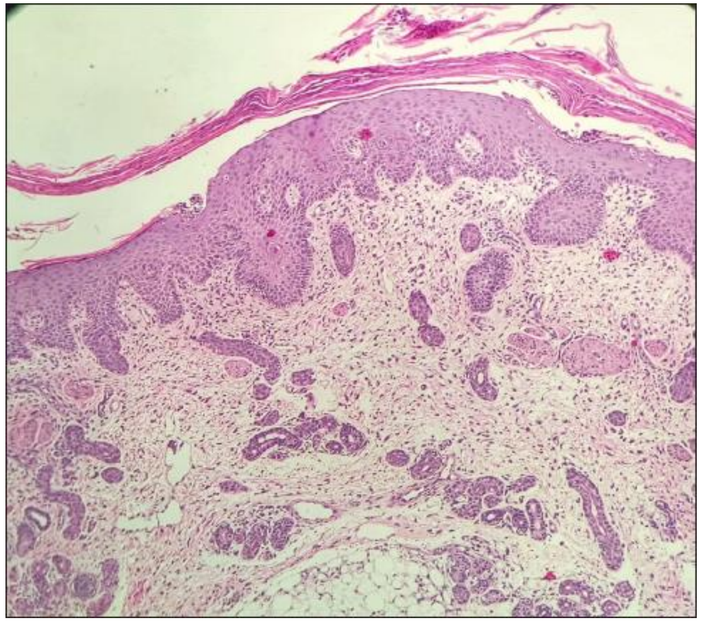
Ichthyosiform, yellow-brown coarse scales with desquamation of skin over the back.



Ectropion and visible corneal opacities in bilateral eyes.



Histopathological examination (Haematoxylin & eosin, 400×) shows lamellar hyperkeratosis with parakeratosis and exocytosis of neutrophils and lymphocytes.



Histopathological examination (Haematoxylin & eosin, 100×) shows hyperkeratosis and irregular acanthosis of the epidermis. Dermis shows mild perivascular lymphocytic infiltrate with melanin incontinence.

Patient-2: 3 months old female (younger sibling of above patient) presented with similar cutaneous and systemic complaints as patient-1 since 1.5 months of age

H/o fever, cough, feeding difficulties, delayed milestones

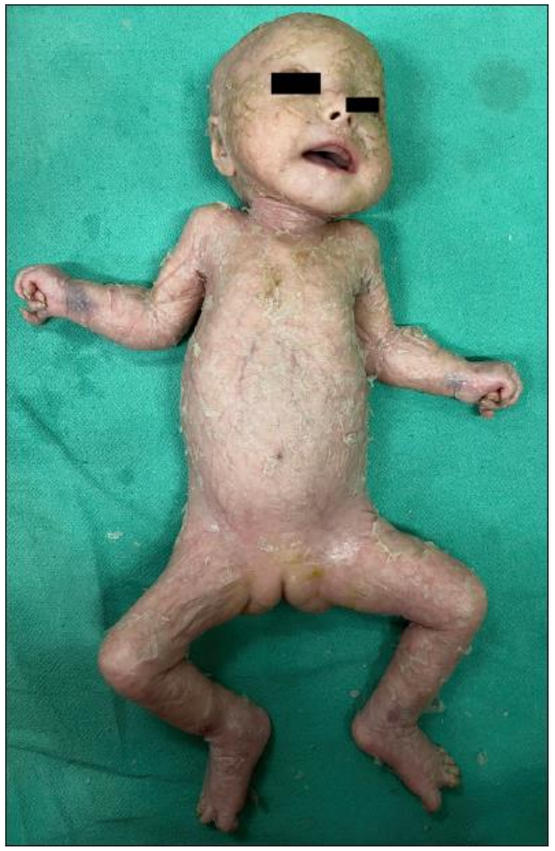
On general examination: pallor, microcephaly, severe stunting

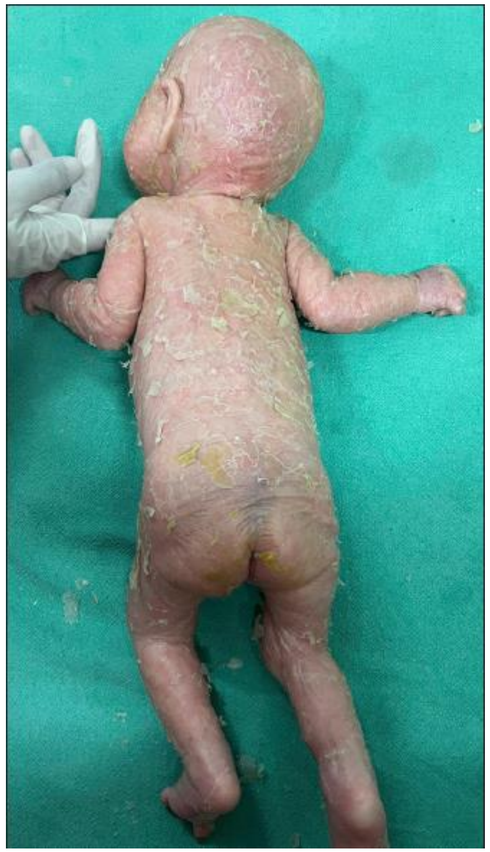
Cutaneous examination: similar to elder child

Abdominal palpation : hepatomegaly, splenomegaly

Musculoskeletal examination: reduced power in upper and lower limbs with hypotonia

Biopsy- features consistent with CIE





- Patient-2 expired after a few days due to aspiration pneumonia
- Patient-1 was started on treatment with systemic antibiotics, topical emollients and anti
- Parents were advised genetic counselling for the disease