LIPOID PROTEINOSIS: URBACH- WIETHE DISEASE

Shubhangi Raju Kori- IInd year MBBS student, Dr. John Stephan, HOD Dermatology

INTRODUCTION

- Lipoid proteinosis is an autosomal recessive disorder that occurs due to loss of fuction mutation in ECM 1 gene encoding for extracellular matrix protein located on chromosome 1q21
- Lipoid proteinosis is characterized by the deposition of hyaline material in the skin, mucous membranes, and internal organs.

CASE REPORT

History

Chief Complaints:

- Beaded papules on the eyelid margin since birth.
- Hoarseness of voice since infancy.
- Multiple scarred lesions over the back, present since 4 years of age.

History of Presenting Illness: An eight-year-old boy presented with a history of beaded papules on the eyelid margin present since birth, persistent hoarseness of voice starting in infancy, and multiple scarred lesions over the back that have been present for the last 4 years. According to the patient's father, the boy had a weak cry as a baby and began developing white papules around the age of 4 months. These papules progressively worsened, and by 4 years of age, they had resulted in scarring on the back.

Past Medical History:

- No history of photosensitivity, seizures, or delayed developmental milestones.

Family History:

- The child was born to consanguineous parents.
- No family history of similar symptoms or skin lesions.

Birth and Developmental History:

- Developmental milestones were normal.

Systemic Examination:

- Vitals and general systemic examination were normal.
- The oral mucosa appeared leathery with thickening of the frenulum.
- There was a slightly pigmented, well-circumscribed, infiltrated plaque with mild scaling over both elbows.

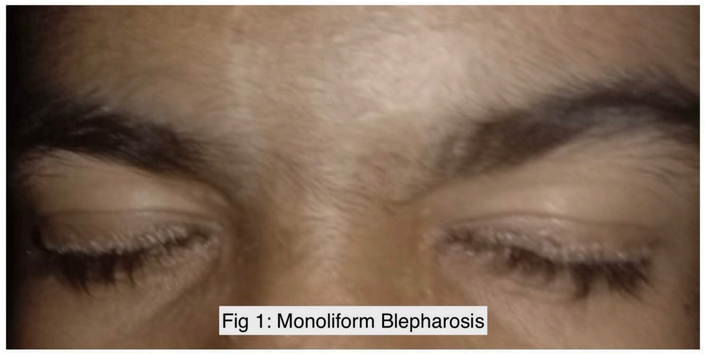

Fig 1: Monoliform Blepharosis

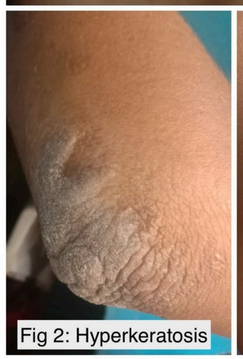

Fig 2: Hyperkeratosis

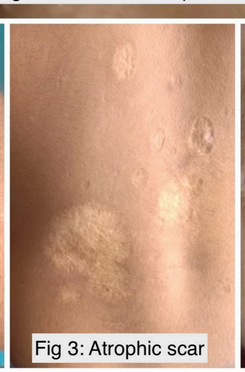

Fig 3: Atrophic scar

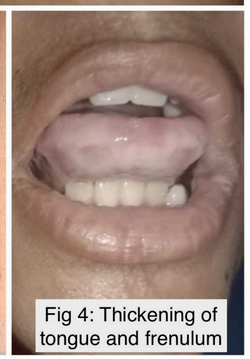

Fig 4: Thickening of tongue and frenulum

INVESTIGATION

- Histopathological examination of skin revealed focal epidermal atrophy, orthokeratosis, and vacuolar changes in the keratinocytes.

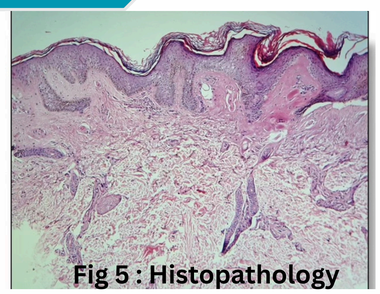

Fig 5 : Histopathology

- Amorphous, acellular hyaline material was observed surrounding the blood vessels in the papillary dermis. Alcian blue and Periodic Acid-Schiff (PAS) staining revealed hyaline deposits in the dermis.
- Congo red staining was negative for amyloidosis.

DISCUSSION

- Lipoid proteinosis is a rare disease with around 400 cases recorded worldwide. Only 51 cases have been reported from India.
- This condition is marked by the deposition of hyaline material in the skin, mucous membranes, and multiple organs, potentially causing widespread systemic effects.

These Include :

1. Skin Changes (Monoliform Blepharosis)
2. Development of Verrucous and Keratotic lesions on the extensor surface, particularly elbows
3. Hyaline deposition in the mucosa, thickening of the tongue and hoarseness of the voice.
4. Deposition of hyaline material in the scalp leads to the destruction of hair follicles causing baldness
5. In a few patients, CNS involvement is seen which results in seizures, memory loss, loss of fear and decreased pain sensitivity.

MANAGEMENT

There is no cure for this condition, treatment focuses on managing symptoms to enhance the patient's quality of life. This includes:

- Excision of laryngeal deposits
- Retinoids, such as acitretin or isotretinoin, have been used to improve skin texture and reduce the appearance of verrucous and keratotic lesions
- Seizures if present are to be managed by with anti-epileptic drugs
- Oral dimethylsulfoxide (DMSO) and D-penicillamine have been used as therapeutic options in the treatment of lipoid proteinosis

CONCLUSION

The particular case is reported due to rarity.

REFERENCES

1. LeWitt TM, Paller AS, Bell A, Zhou XA. Lipoid Proteinosis [Internet]. PubMed. Treasure Island (FL): StatPearls Publishing; 2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK568769/

2. LeWitt TM. Lipoid proteinosis [Internet]. U.S. National Library of Medicine; 2023 [cited 2024 Sept 13]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK568769/

3. Chatterjee A, Viswanathan LG, Madhu Nagappa, Sinha S. Lipoid proteinosis (urbach-wiethe disease): A rare genodermatosis with characteristic dermatological and neuroimaging findings. Annals of Indian Academy of Neurology. 2021 Jan 1;24(5):761–1.

4. Vahidnezhad H, Youssefian L, Uitto J. Lipoid Proteinosis [Internet]. Adam MP, Feldman J, Mirzaa GM, et al., editors. PubMed. Seattle (WA): University of Washington, Seattle; 1993. Available from: https://www.ncbi.nlm.nih.gov/books/NBK338540/