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A CASE OF LIPOID PROTEINOSIS – URBACH WIETHE DISEASE

Medicine

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ABSTRACT

A 52-year-old normotensive and non-diabetic female presented with complaint of change in voice, beading papules, vesicles and bullae on bilateral eyelids, whitish discolouration of skin and mucus membrane of upper lip and difficulty in chewing and decreased tongue movements for 2 months. Skin biopsy revealed LIPOID PROTEINOSIS. Life span is normal unless altered by laryngeal obstruction. Few may require permanent tracheostomy.

KEYWORDS

Lipoid proteinosis, multi-system involvement.

BACKGROUND

Lipoid proteinosis also known as URBACH-WIETHE disease is a rare, chronic, autosomally inherited disorder with multisystem involvement due to extracellular deposition of an amorphous hyaline material. Named after Erich Urbach and Camilo Wiethe. This disease presents with hoarseness of voice followed by pox-like and acneiform scars along with infiltration and thickening of the skin and certain mucus membranes. Autosomal recessive inheritance, with no sex predilection

CASE REPORT

A 52-year-old normotensive and non-diabetic female presented with complaint of

- Change in voice, beading papules, vesicles and bullae on bilateral eyelids
- Whitish discolouration of skin and mucus membrane of upper lip
- Difficulty in chewing and decreased tongue movements for 2 months

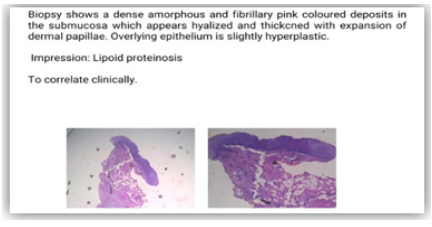
No history of similar complaints in past.
The patient underwent total abdominal hysterectomy with bilateral salpingo-oophorectomy 25 years ago.

Routine investigations such as LFT, RFT, Serum electrolytes, haemogram and Chest X-ray were normal.

Ultrasound study of abdomen was normal, except for absence of uterus and adnexae.

Skin biopsy revealed LIPOID PROTEINOSIS [Fig. 1]

FIGURE 1



DISCUSSION

Lipoid proteinosis also known as URBACH-WIETHE disease is a rare, chronic, autosomally inherited disorder with multisystem involvement due to extracellular deposition of an amorphous hyaline material. Named after Erich Urbach and Camilo Wiethe, this disease presents with hoarseness of voice followed by pox-like and acneiform scars along with infiltration and thickening of the skin and certain mucus membranes. Autosomal Recessive in inheritance, with no sex predilection. Life span is normal unless altered by laryngeal obstruction. Few may require permanent tracheostomy. Mutation of Extracellular Matrix Protein 1 [ECM 1] gene located on chromosome 1 affects a protein in the extracellular space that is seen in all tissues and organs, leads to deposition of hyaline like material in the skin and viscera in abnormal amounts which causes clinical manifestations. (1) CT scan of brain may reveal bilateral intracranial bean shaped calcifications in temporal lobe, amygdala and hippocampus (2)

Differential diagnoses include amyloidosis, nodular localized cutaneous amyloidosis, leprosy, lichen myxedematosus, xanthomas. (3)

CONCLUSION

Lipoid proteinosis also known as URBACH-WIETHE disease is a rare, chronic, autosomally inherited disorder with multisystem involvement due to extracellular deposition of an amorphous hyaline material. Life span is normal unless altered by laryngeal obstruction. Few may require permanent tracheostomy.

REFERENCES

1. Hamada T, McLean WHI, Ramsay M, Ashton GHS, Nanda A, et al. 2002. Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). Human Molecular Genetics 11:833–40
2. Appenzeller, S; Chaloult, E; Velho, P; De Souza, E. M.; Araújo, V. Z.; Cendes, F; Li, L. M. (2006). "Amygdalae Calcifications Associated with Disease Duration in Lipoid Proteinosis". Journal of Neuroimaging. 16 (2): 154–156.
3. DiGiandomenico S, Masi R, Cassandrini D, El-Hachem M, DeVito R, Bruno C, Santorelli FM. 2006. Lipoid proteinosis: case report and review of the literature. Acta Otorhinolaryngol Ital 26:162–7