Lipoid Proteinosis: A Rare Genetic Disorder

**ABSTRACT**

Lipoid Proteinosis or Urbach-Wiethe Disease is a rare genetic disorder that causes hoarseness, skin, mucosa and eye lesions and neurological symptoms due to a buildup of hyaline material. The diagnosis is made through biopsy of skin or mucosa. The disease has a benign course and there isn't still a specific and effective treatment.

**INTRODUCTION**

Lipoid Proteinosis or Urbach-Wiethe Disease was first described in 1929. It's a rare recessive genetic disorder, which causes a buildup of hyaline material in the skin and mucosa. There are approximately 300 cases reported in the international scientific literature. Their main symptoms are hoarseness and lesions on the skin and eyelids. Some cases may present with neurological findings. The usual age of diagnosis is still under the age of 6 months old. The objective of this paper is to report a case of Lipoid Proteinosis or Urbach-Wiethe Disease with the emphasis on their otolaryngological manifestations.

**CASE REPORT**

Report of a 38 years old patient that presented since she was 6 months old skin lesions similar to pustules in buttocks, face, arms and legs and a weak cry. The skin lesions got less important through the years and they were related to emotional stress. The patient has a hoarseness that keeps her from talking on the telephone. There were no neurological or ophthalmological symptoms. The physical exam showed an infiltrated skin; the absence of teeth; pale, stiff and irregular oral mucosa, especially in the tongue and lingual frenulum. The nasal vestibules were narrowed. (Pictures 1-4)

The videolaryngoscopy showed an infiltration of the arytenoids and epiglottis. The vocal cords had normal mobility and coaptation. (Picture 5)

A skin biopsy was requested and the deposition of hyaline material PAS positive was found in the papilar derme, confirming the diagnosis of Lipoid Proteinosis. The patient received treatment with topic and systemic corticosteroids and also phonotherapy and obtained some improvement. The patient doesn't want to be submitted to a surgical procedure. She is in follow-up in Gafrée Guinle University Hospital.

**DISCUSSION**

Lipoid Proteinosis shows their first symptoms still in childhood, with skin lesions and a hoarse cry. It has a positive family history in 20% of the cases, it's more common in Europeans citizens and there's not a preference for sex. The cause of the disease is still unknown; however, recent studies suggest that an imbalance between the production of type 1 and type 4 collagen fibers could be involved. This disease has a chronic and benign course, and the symptoms get less exuberant as the patient grows older and with pregnancy.

The most common symptoms are hoarseness, skin lesions and moniliform blefarosis. In some cases the central nervous system (CNS) may be involved, causing neurological symptoms such as epilepsy. Other than the hoarseness, the infiltration of the nose, oral mucosa, tongue, lingual frenulum, tonsils and uvula can cause some speech restriction and the narrowing of the nasal vestibules. Other findings are the absence of teeth, gingival hyperplasia and recurrent parotitis.

**CONCLUSIONS**

Lipoid Proteinosis is a rare disease that has a chronic and benign course, still without a specific treatment. Due to the variety of symptoms in the oral cavity, larynx and nose it is important that the ear, nose and throat professional is familiar with this affection.

**REFERENCES**

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