Lipoid proteinosis of the larynx in siblings: exploring new modalities of treatment

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Abstract Lipoid proteinosis is a rare autosomal recessive disease inheritance with deposition of hyaline-like material at cutaneous and non-cutaneous sites. This is a report of two siblings presenting with typical cutaneous and laryngeal manifestations of lipoid proteinosis since birth who were treated successfully with placental extract.

Keywords Lipoid proteinosis · Siblings · Laryngeal manifestations · Placental extracts

Introduction

Lipoid proteinosis, also called Urbach-Weithe disease [1] or hyalinosis cutis et mucosae, is a rare disease with much rarer occurrence in siblings. This genodermatosis is an autosomal recessive disease with involvement of the oral cavity, oropharynx and larynx along with characteristic cutaneous lesions occurring due to deposition of amorphous hyaline material. The usual presentation is with hoarseness of voice and shrill cry since childhood [2, 3].

Case report

A 12-year-old girl and her younger brother, a 5-year-old boy, both born out of a second-degree consanguineous marriage presented to our outpatient department with complaints of hoarseness of voice and shrill cry since childhood. They also complained of skin lesions which followed the hoarseness. The other three siblings were normal. Physical examination revealed typical skin beading around the eyelids and papular lesions over the flexor surface of elbows and knuckles. The tongue was woody, thickened and irregular. The patients had difficulty in protruding the tongue fully. The voice was husky and easily fatigable.

Flexible laryngoscopy showed diffusely thickened and nodular epiglottis. An elevated irregular lesion with intact mucosa was seen at the interarytenoid region with phonatory gap between the true vocal cords. Aryepiglottic folds, false cords and true cords appeared thickened (Fig. 1). Upper end esophagoscopy and bronchoscopy did not show any abnormality. Biopsy and histological examination of the lesions over the epiglottis, skin and tongue were consistent with findings of lipoid proteinosis, which showed perivascular deposition of periodic acid schiff (PAS)-positive and diastase-resistant hyaline-like material which was negative for Congo Red staining (Fig. 2). The karyotyping of both siblings was done and showed no abnormality. Radiography

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of the skull did not reveal any abnormal calcification. Lipid profile and other investigations were normal.

In view of the diffuse involvement and absence of respiratory obstruction, the patients opted for medical line of management with etretinate, which the patients could not tolerate because of gastric irritation. Subsequently, after obtaining consent, both the patients were given intramuscular injections of human placental extract on alternate days with topical application of placental extract gel for cutaneous lesions, as placental extract is reportedly useful in inflam-

Fig. 1 Laryngeal photograph showing irregular growth with intact mucosa at the interarytenoid region

Fig. 2 Histopathology of the tongue showing hyperkeratosis, deposition of PAS-positive hyaline material in the perivascular area with onion skin appearance of hyaline material deposition

matory processes involving collagen synthesis [4, 5]. After two months of treatment the elevated cutaneous and oral lesions flattened and showed significant cosmetic improvement. There is a definite improvement of the quality of voice which the patients could appreciate. The patients are on follow up for further improvement.

Discussion

The lipoid proteinosis is a rare disease more common in the European and South African population. It has an autosomal recessive pattern of inheritance with hoarseness of voice as a primary symptom when the larynx is involved. If left untreated, it may also present with stridor and respiratory embarrassment. Hence, knowledge of this disease with a proper follow-up and treatment is essential to avoid respiratory complications. The cutaneous lesions are classical in their appearance with beading of the margins of the eyelids (moniliform blepharosis). The laryngeal involvement is diffuse with primary involvement of supraglottis. Histological examination shows a PAS-positive thickening of the mucosa and deposition of hyaline-like material which is negative for Congo Red staining in contrast to amyloidosis. Neurological symptoms and behavioral abnormalities were reported in patients with abnormal calcification in the suprasellar area. The pathogenesis is unknown. It is mainly thought to be a metabolic defect in collagen synthesis. There is an increase in type IV collagen with a decrease in type I collagen. The defect is mapped to chromosome 1q21 [6].

The prognosis for life is good and treatment for laryngeal lesions has been mainly microlaryngeal surgery with carbon dioxide laser. Oral dimethyl sulphoxide, pencillamine and etretinate [7] have also been tried.

Conclusion

With our experience in treating the two siblings with lipoid proteinosis, we would like to bring forward the observation that placental extract injections have a definite role in the favorable outcome in the treatment of lipoid proteinosis. In our literature review we did not find any reported usage of placental extract for lipoid proteinosis. The treatment outcome with placental extract is better, safer and cheaper than other conventional modes of treatment. We are presenting this paper as lipoid proteinosis is a rare disorder of the larynx and also because of the success achieved with placental extract treatment.

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