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CASE REPORT

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Lipoid proteinosis: A rare genodermatosis with multisystemic manifestations—A case report

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Key Clinical Message

Lipoid proteinosis (LP) is a rare autosomal recessive genodermatosis, which is characterized by the deposition of amorphous hyaline material in various tissues, including the mucosa, visceral organs, and skin. We report a case of a 11-year-old girl born to consanguineous parents presented with multisystemic manifestations of the disorder. The patient presented with progressive skin lesions evolving from blisters to papules, distinctive beaded papules along eyelid margins, hoarseness of voice, impaired speech, hair loss, and a painful jaw swelling. Clinical examination revealed waxy skin, atrophic scars, and keratotic plaques. Histopathology report revealed amorphous hyaline eosinophilic material deposition. This case report highlights the multisystemic manifestations of LP and the importance of early diagnosis and management.

KEYWORDS

case report, genetic disorders, genodermatosis, lipoid proteinosis

1 | INTRODUCTION

Lipoid proteinosis (LP) is also one of the rare autosomal recessive genodermatosis, which is characterized by the abnormal deposition of amorphous hyaline material in various tissues, including the skin, oral cavity, larynx, and internal organs.[1] This deposition leads to a wide spectrum of clinical presentations, making early diagnosis and appropriate management pivotal. The disease was first described in 1929, and fewer than 300 cases have been reported in the medical literature. Lipoid proteinosis is also known as Urbach–Wiethe disease or cutaneous-mucosal hyalinosis.[1,2]

The genetic basis of lipoid proteinosis involve loss-of-function mutations in the gene responsible for encoding extracellular matrix protein 1 (ECM1) on the chromosomal band 1q21.[2] These genetic alterations result in the aberrant accumulation of hyaline material in affected tissues. Notably, consanguineous parentage has been frequently associated with the inheritance of this rare disorder, suggesting a hereditary basis. So, the disease is inherited in not only shows an autosomal recessive pattern, which is heterozygous carriers have an apparently normal phenotype rather may have some subtle changes such as abnormal dentition.[2,3] Clinical features of lipoid

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proteinosis are myriad, and the literature describes that sometimes considerable variation found in severity between affected patients. The classic manifestation is onset in infancy with a hoarse cry due to laryngeal infiltration. Other common symptoms include multiple atrophic scars, eyelid beading, and waxy papules on the face and extremities. The oral cavity is the most extensively affected area by the disease, with thickening of the tongue, lips, and buccal mucosa.[3,4] So, we present a case of an 11-year-old girl born and something about consanguineous parents, showcasing the intricate multisystemic manifestations of lipoid proteinosis.

2 | CASE PRESENTATION

A 11-year-old girl born to consanguineous parents presented with progressive skin and mucous membrane changes since early childhood (3 years of age). At first, the skin lesions started as blisters on the back, shoulders, elbows, and scalp, which eventually healed with scarring. At present, the lesions begin as papules (1–2 mm diameter), which heal with scarring. She also presented with multiple, beaded papules embedding the eyelid margins. The mother noticed hoarseness of voice followed by impaired speech, since the child was of 3 years of age. The mother noticed excessive loss of hair as well. The child also presented with a painful swelling on the right side of the jaw. The mother gives no history of frontal headache, seizures, or any visual disturbances in the child. The child has no history of difficulty in swallowing, any frequent upper respiratory tract infections, or episodes of difficulty in breathing. There is no history of atopy. The patient has normal appetite, consumes mixed diet, and the bowel and bladder movements appear to be regular. No similar complaints were reported in the family.

On general examination, conscious, coherent, cooperative. Well-nourished and moderately built. No signs of pallor, icterus, cyanosis, koilonychias, lymphadenopathy, and edema. The patient was afebrile. Pulse rate was reported to be 90 beats min^{-1}, with regular volume and rhythm while the respiratory rate was reported at 22 breaths min^{-1}. Blood pressure was noted to be 110/70 mmHg. The cardiovascular, respiratory, and the central nervous system examination were noted to be normal.

The facial skin was waxy, which in appearance with a multiple, superficial scars, ill-defined, atrophic scars of varying size, ill-defined plaques, corrugated, rough, keratotic, were present over bilateral elbows, shoulders, and back. Multiple skin colored linearly, which is arranged closely aggregated 1–2 mm sized papules predominantly over both upper and lower eyelid margins. Tender painful swelling on the right mandible in favor of parotitis. The scalp revealed scarring alopecia. Nails were normal. Genitals were normal. Intraoral examination revealed hypertrophied tongue with whitish plaques, erosions and fissures and restricted tongue movements. Histopathological evidence from scalp and shoulders reveals amorphous acellular hyaline eosinophilic material in the dermis. Skin appendages show similar type of deposition surrounding them. Slit-lamp examination revealed multiple yellow waxy beaded papules along the eyelid margins and distichiasis. CT brain findings include bilateral medial temporal sickle-shaped calcifications. The treatment includes Tab ISOTROIN 10 mg OD, Tab LIMCEE 500 mg OD, APIFIL Moisturizing Cream TID, SPER Sunscreen, and three sessions of microdermabrasion at an interval of 3 weeks. The patient was under follow-up for 4 weeks, and no recurrence was noted (Figures 1 and 2).

3 | DISCUSSION

Lipoid proteinosis, a rare autosomal recessive genodermatosis, represents a clinical challenge due to its infrequency

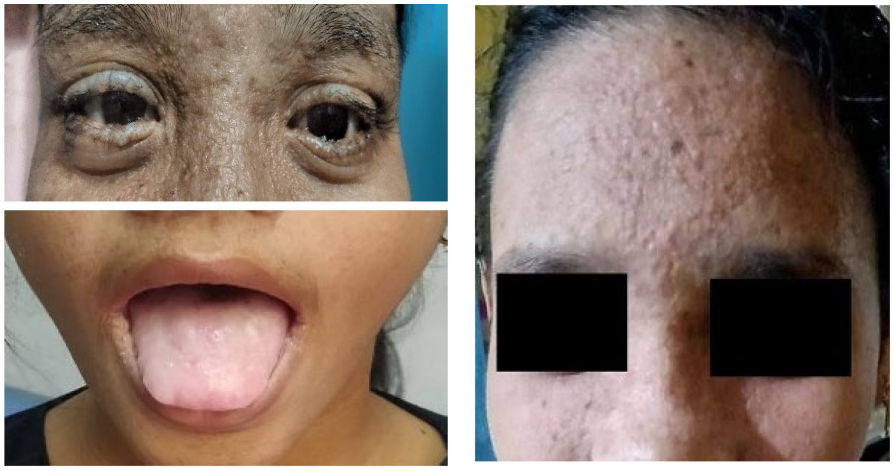

FIGURE 1 Multiple, skin-colored, some linearly arranged, closely aggregated 1–2 mm papules on eyelid margins. Hypertrophied tongue with whitish plaques, fissures, and erosions. Waxy facial skin appearance with multiple atrophic ill-defined, superficial scars of varying size.

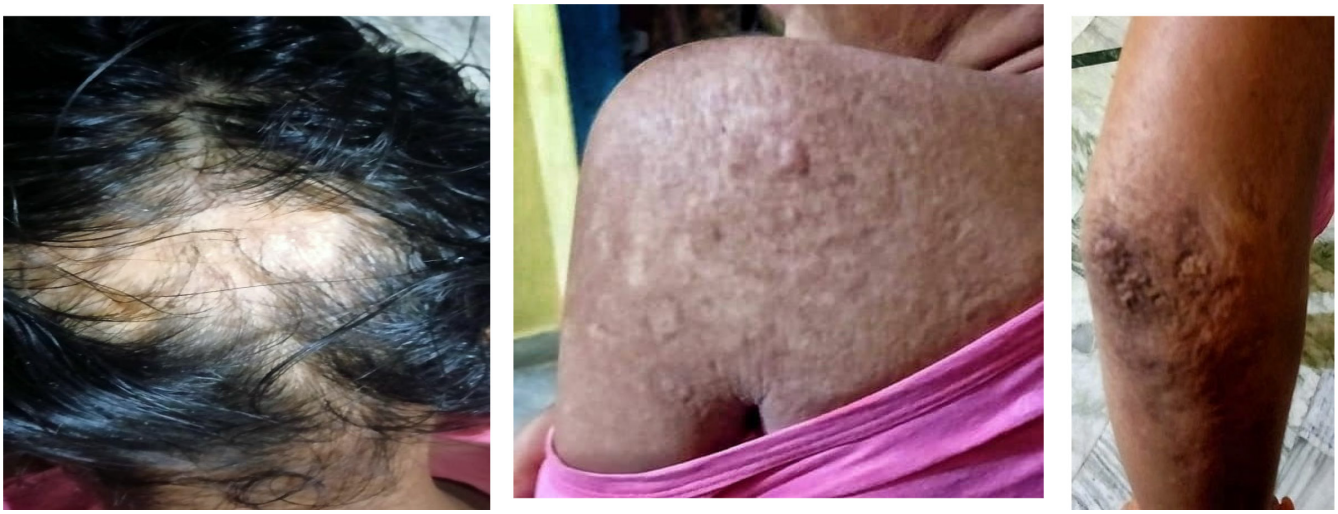

FIGURE 2 Scarring alopecia and corrugated, rough, ill-defined, keratotic plaques on bilateral elbows and shoulders.

and multifaceted presentations. Lipoid proteinosis primarily manifests as the deposition of amorphous hyaline material in various tissues, including the skin, oral cavity, larynx, and internal organs.[4,5] Clinically, patients often exhibit cutaneous and mucosal alterations, such as blistering, scarring, and beaded papules along eyelid margins, which were strikingly evident in our case. Furthermore, the disease is frequently associated with voice and speech abnormalities, as seen in our patient who displayed hoarseness of voice and impaired speech.[6]

Genetically, LP is attributed to the loss of function of the mutations in the gene encoding ECM1 on the chromosomal band 1q21. These mutations lead to the aberrant deposition of hyaline material in various tissues. Consanguinity in patients' parents is identified in approximately 20% of lipoid proteinosis cases.[7] It is notable that the consanguineous parentage in our case is consistent with previous studies that have suggested a hereditary basis for this disorder. Genetic counseling and prenatal genetic testing, as mentioned in our case report, become pivotal in such familial scenarios. These interventions can offer informed choices to families at risk, thereby potentially preventing the birth of affected individuals.[8,9]

Management of lipoid proteinosis remains a complex endeavor. Various treatment modalities have been explored, although no curative therapy exists. The primary goals of treatment are to alleviate symptoms, improve quality of life, and minimize complications.[1,3] Various therapeutic options available for LP, which includes topical and systemic corticosteroids, oral dimethyl sulfoxide, etretinate, acitretin, D-penicillamine, and intralesional heparin.[1,4,8] Furthermore, chemical peeling and surgical interventions such as dermabrasion and CO2 laser procedures can be considered for managing scarring and skin changes. Nonetheless, the effectiveness of these procedures should be carefully weighed against the potential for adverse effects and recurrence.[9,10]

4 | CONCLUSION

Lipoid proteinosis is an exceedingly rare genodermatosis with distinct clinical and genetic features. Our case report underscores the importance of accurate diagnosis, multidisciplinary collaboration, and tailored therapeutic strategies. Genetic counseling and prenatal genetic testing emerge as vital tools for families at risk. While therapeutic options exist, they are primarily symptomatic, emphasizing the need for ongoing research to discover more targeted treatments and potentially a cure for this enigmatic disorder. By disseminating knowledge and fostering collaboration among clinicians, researchers, and genetic counselors, we can contribute to improved outcomes and enhanced quality of life for individuals affected by lipoid proteinosis.

AUTHOR CONTRIBUTIONS

Farah Naaz Hashmi: Investigation; validation; writing – original draft; writing – review and editing. **Sumera Huma:** Data curation; validation; visualization; writing – original draft. **Harshini Singireddy:** Data curation; software. **Nikhat Zareen:** Funding acquisition; investigation; software. **Tarun Kumar Suvvari:** Conceptualization; data curation; formal analysis; investigation; methodology; writing – original draft. **Mustafa Hussain Ansari:** Investigation; visualization. **Nudrat Sultana:** Investigation; validation. **Md. Al Hasibuzzaman:** Project administration; supervision; writing – original draft.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

None declared.

ETHICS STATEMENT

In our university, ethics approval was not required for case reports and case series.

CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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