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Navigating the Uncommon: A Case Study of Pregnancy in Lipoid Proteinosis

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Introduction

Lipoid proteinosis (LP), also known as Urbach-Wiethe disease, is a rare autosomal recessive genodermatosis characterized by the deposition of hyaline-like material in various tissues, including the skin, mucous membranes, and internal organs. Fewer than 300 cases have been reported in the literature to date.

The condition is caused by mutations in the ECM1 gene located on chromosome 1q21, leading to a loss of function of the extracellular matrix protein 1. This protein plays a crucial role in skin integrity, angiogenesis, and binding of dermal collagen and proteoglycans. The clinical manifestations of LP are diverse, often involving the skin, oral cavity, and central nervous system. Pregnancy in patients with LP is exceedingly rare, and its implications are not well understood.

While specific complications of LP in pregnancy are not well-documented due to the rarity of the disease, following are the systems involved with associated complications in lipoid Proteinosis.

Keywords: Lipoid proteinosis, moniliform blepharosis, 2D echocardiogram

Respiratory System: Laryngeal Involvement: Thickening and infiltration of the laryngeal mucosa in Lipoid proteinosis can cause hoarseness or airway obstruction. During pregnancy, changes in hormone levels may exacerbate mucosal edema, potentially worsening respiratory symptoms and increasing the risk of airway obstruction, especially during labor or anesthesia.

Delivery Concerns: Lipoid proteinosis can cause scarring of the skin and mucous membranes. During delivery, this may lead to an increased risk of perineal tears or Impaired wound healing & may predispose patients to infection

Central Nervous system: Involvement of the temporal lobe in Lipoid proteinosis can predispose patients to seizures. Pregnancy may lower the seizure threshold posing risk to both mother and fetus.

Fetal Considerations: LP is inherited in an autosomal recessive manner. If both parents are carriers, there is a

25% chance the fetus may inherit the condition. Genetic counselling is essential for at-risk couples.

Placental Function and Vascular Complications: Although not directly reported in LP, the hyaline deposition characteristic of the disease might theoretically affect placental function, potentially leading to fetal growth restriction or preterm birth. Thickened mucosal tissues might increase the risk of vascular complications during pregnancy or delivery.

Case Report

A 29-year-old woman, born to consanguineous parents, presented at 29 weeks and 1 day of gestation to the emergency department with a 2-hour history of headache. An ultrasound performed earlier that day revealed reversal of end-diastolic flow in the umbilical artery, raising concerns for fetal well-being.

Past Medical History

- **Lipoid Proteinosis Diagnosis:** The patient was diagnosed with LP in 2019 after presenting with recurrent episodes of spontaneous blistering and subsequentatrophic scarring over the scalp and trunk since childhood. A skin biopsy confirmed the diagnosis.
- **Obstetric History:** She had a previous intrauterine fetal demise (IUFD) at 29 weeks of gestation three years prior.
- **Other Medical History:** No history of hypertension, diabetes, asthma, thyroid disorders, or tuberculosis.

Family History: No known history of similar conditions among family members.

Antenatal Course: The patient registered for antenatal care at 24 weeks and 6 days of gestation. Given her diagnosis of LP, a multidisciplinary approach was adopted:

Ophthalmology Consultation: Evaluated for multiple granular swellings over both eyelids, diagnosed as moniliform blepharosis, a characteristic finding in LP.

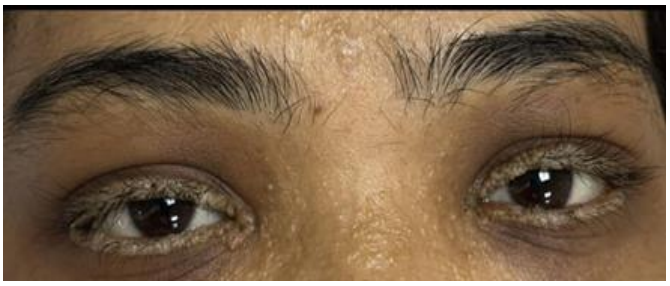

Figure 1: Eyelid lesions (moniliform blepharosis) with a background of facial scarring.

ENT Consultation: Assessed for hoarseness of voice, a common manifestation due to laryngeal involvement in LP. Laryngoscopy revealed vocal cord hypertrophy and edematous arytenoid cartilage. Speech therapy and gastroesophageal reflux disease prophylaxis were recommended.

Cardiology Evaluation: A 2D echocardiogram showed an ejection fraction of 60% with trivial tricuspid regurgitation, which was deemed clinically insignificant.

Examination on Admission:

- **Vital Signs:** Blood pressure was elevated at 140/100 mmHg.
- **Urinalysis:** Significant proteinuria with a dipstick reading of +3.
- **Neurological Examination:** Deep tendon reflexes were normal.
- **Obstetric Examination:** The uterus measured consistent with 24-26 weeks gestation. Fetal heart tones were not detectable with a handheld Doppler.
- **Ultrasound**: Confirmed intrauterine fetal demise.

Management: Labor was induced in light of the IUFD. Given the patient's elevated blood pressure and proteinuria, a loading dose of magnesium sulphate was administered to prevent eclampsia. Post-delivery, the patient developed oliguria progressing to anuria.

Nephrology was consulted, and she was diagnosed with pre-renal acute kidney injury secondary to thrombotic microangiopathy, a pregnancy-related complication.

Investigations

- **Lactate Dehydrogenase (LDH):** 206 U/L
- **C-Reactive Protein (CRP):** 114 mg/L
- **ANA/ Anti dsDNA/ ANCA:** Negative
- **Sr Haptoglobulin:** < 15 (normal range 35-200mg/dl)
- **Anti-complement factor H (CFH) assay:** Normal (Done to rule out HUS from TTP)
- **Lipid Profile**

➢ Total Cholesterol: 204 mg/dL
➢ Triglycerides: 94 mg/Dl
➢ High-Density Lipoprotein (HDL): 61.9 mg/dL
➢ Low-Density Lipoprotein (LDL): 176.6 mg/dL
➢ Very Low-Density Lipoprotein (VLDL): 18.8 mg/Dl

MRI Brain: Bilateral temporal lobe and hippocampal calcifications and no evidence of PRES

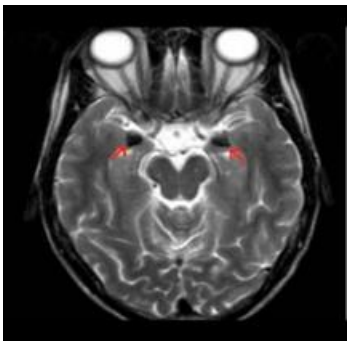

Figure 2: T1/T2 hypointense areas with blooming in SWI in temporal lobes

Outcome

She underwent plasmapheresis and hemodialysis, with gradual improvement in urine output. She was managed with strict fluid balance monitoring and regular renal function assessments.

The patient's renal function improved, and she was discharged with recommendations for regular follow-up.

Discussion

Lipoid proteinosis is a multisystem disorder with variable clinical manifestations. The deposition of hyaline material can lead to skin scarring, mucosal involvement, and central nervous system abnormalities, including temporal lobe calcifications, which may predispose patients to seizures. In this case, the patient's LP manifestations included characteristic skin lesions and laryngeal involvement leading to hoarseness. The recurrence of IUFD at 29 weeks in two consecutive pregnancies raises questions about potential links between LP and adverse pregnancy outcomes. While no direct association has been established in the literature, the possibility of systemic involvement affecting placental function cannot be excluded. Additionally, the development of thrombotic microangiopathy postpartum suggests a potential interplay between LP and pregnancy-related hypertensive disorders.

Conclusion

- The management of pregnancy in women with lipoid proteinosis requires a comprehensive and multidisciplinary approach due to the unique challenges posed by the condition.
- Early antenatal care, including timely registration, screening for pre-eclampsia, and initiation of low-dose aspirin, plays a critical role in mitigating risks.
- Frequent ultrasound examinations, including Doppler studies, are essential to monitor fetal well-being and detect any signs of placental insufficiency or fetal compromise.
- A multidisciplinary team, consisting of obstetricians, nephrologists, genetic counselors, and other specialists, ensures comprehensive care, especially in addressing potential renal complications and other systemic manifestations of the disease.

- Regular antenatal monitoring, along with an evaluation for antiphospholipid syndrome, further enhances the management of this high-risk pregnancy.
- Genetic counseling provides families with the necessary information on the hereditary nature of lipoid proteinosis, guiding future reproductive decisions. This holistic approach aims to optimize both maternal and fetal outcomes, highlighting the importance of early intervention and close monitoring throughout the pregnancy.

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