

## **Lipoid Proteinosis (Urbach-Wiethe Disease)**

**Patient:** A.A., a 3-year-old girl  
**Duration:** 2 years  
**Distribution:** Face, hands and oral mucosa

**History:** Weak cry as an infant, hoarseness at a later age. Fragile skin and bullous lesions that resolve with scarring. Parents are consanguineous. Several family members affected.

**Physical Exam:** Multiple shallow linear and saucer shaped scars mainly on the face, associated with few crusted erythematous papules. Lip and tongue are infiltrated and indurated.

### **Histopathology:**

PAS positive, pink hyaline-like deposits in the dermis and around capillaries with thickened basement membranes.

**Laboratory:** None

**Treatment:** None

### **Discussion:**

- Rare autosomal recessive genodermatosis in which masses of hyaline material are deposited. Males and females are equally affected with a strong predilection for white races. The dermal deposits have been related to a perturbation of collagen metabolism and glycoprotein production. Evidence of lysosomal storage leading to deposition of complex lipids has also been documented, but the lipid component to date is not characterized.

- Clinical presentation is variable, cutaneous lesions are dominant but there may be severe visceral, CNS and ocular involvement.

The earliest manifestation is a weak cry in infancy and hoarseness that remains throughout life. Skin lesions appear during the first 2 years and occur in 2 overlapping stages.

1. Bullae and hemorrhagic crusts that resolve with ice pick scarring.
2. Skin becomes thickened yellowish and waxy. Verrucous lesions may occur on the extensor surfaces. Beaded papules on the palpebral margins (eyelids) are characteristic.

In the mucosa the mouth is most extensively affected. Lesions include infiltration of the tongue, induration and pebbling of the lip mucosa. Gingival involvement has also been reported. Adults are at risk of laryngeal obstruction.

- A unique and pathognomonic finding is bilateral intracranial and sickle shaped calcification

within the temporal lobe. Routine lab tests are within normal.

- There is no known cure except for anecdotal reports of success with DMSO, CO<sub>2</sub> laser and dermabrasion.

**References:**

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