

Urbach-Wiethe syndrome

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DESCRIPTION

A 14-year-old girl was referred for skin tightening in the fingers. She did not have Raynaud's phenomenon, gastroesophageal reflux disease or other systemic symptoms. She had had a hoarse voice since birth and was developmentally normal. She was born of a second degree consanguineous marriage. Examination revealed pearly papules around the eyelids, face (figure 1) and fingers (figure 2), and hyperkeratotic plaque on the elbows (figure 3). Oral cavity examination revealed ankyloglossia (figure 4). Systemic examination was normal. Haemogram, and liver and renal function tests were within normal limits.

The patient had a younger sibling (12 years old) with similar facial features (figure 5), elbow rash and oral cavity findings. CT of head of both siblings revealed bilateral basal ganglia calcification (figure 6). Both sisters were diagnosed to have lipid proteinosis, or Urbach-Wiethe syndrome. Since they were asymptomatic, they were not given any medications and remained on follow-up.



Figure 3 Elder sibling—hyperkeratotic plaque on dorsum of elbows.

Papular lesions on the face, eyes and fingers merit consideration of congenital erythropoietic protoporphyria, lepromatous leprosy and amyloidosis. Absence of painful photosensitivity ruled out erythropoietic protoporphyria in this case; and absence of hypoaesthetic skin lesions, paresthesias and joint pains ruled out leprosy. Onset in early childhood with family history, and absence of renal and gastrointestinal tract involvement, ruled out amyloidosis.^{1 2} Hoarseness of voice with onset in early childhood merits a differential diagnosis of hypothyroidism, which can also be associated with coarse skin. However, hypothyroidism with onset from infancy is likely to be associated with mental retardation, and our patients had normal mental development. The symptom complex encountered in our patients regarding the papular lesions on the face and hands, and around the eyes, the hyperkeratotic plaque on the elbows, the ankyloglossia and hoarse voice, along with bilateral basal ganglia calcification, was characteristic of Urbach-Wiethe syndrome.³



Figure 1 Elder sibling—beaded pearly papules around eyelids and on face.



Figure 2 Elder sibling—papules on dorsum of hands and fingers.



Figure 4 Elder sibling—restricted tongue protrusion.



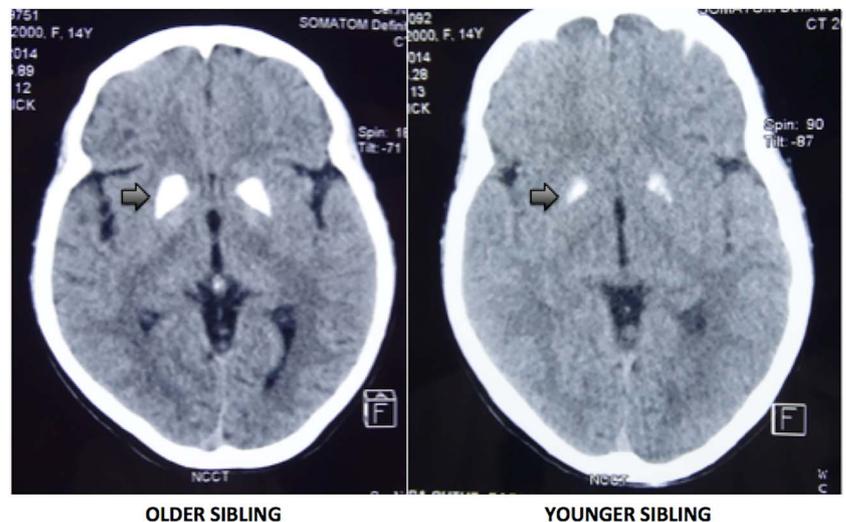
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Figure 5 Younger sibling—beaded pearly papules around eyelids and on face.

Urbach-Wiethe syndrome is a rare autosomal recessive disorder due to mutation of extracellular matrix protein 1 (ECM1), leading to deposition of collagenous material in the skin and soft tissues. The characteristic facial appearance includes waxy papules, also called moniliform blepharosis, on the face, including the eyelids.⁴ Patients may develop hyperkeratotic plaque over sites of frequent trauma, such as the elbows. Ankyloglossia, or inability to protrude the tongue, is due to collagen deposition causing a thick frenulum.^{1, 3} Complications encountered in these patients include dental hypoplasia, gum hypertrophy, dryness of mouth, recurrent parotid and submandibular gland infections, and predisposition to respiratory distress during an episode of upper respiratory infection, due to thickened tongue and larynx.^{5, 6} Neurological complications include basal ganglia calcification, usually bilaterally symmetric, which can predispose to seizures, subtle cognitive abnormalities⁷ and, rarely, spontaneous intracerebral haemorrhage.⁸ Usually, patients have a normal lifespan. Anecdotal reports suggest improvement in skin features with acitretin; its teratogenic potential merits cautious use in young females.

Figure 6 Bilateral basal ganglia calcification (black arrow) in both siblings seen on non-contrast CT of the head.



Learning points

- ▶ Urbach-Wiethe syndrome is a rare autosomal recessive disorder predisposing to increased collagen deposition in the skin and soft tissues.
- ▶ Characteristic features include moniliform blepharosis, ankyloglossia and bilateral symmetric basal ganglia calcification.
- ▶ It is important to recognise this rare non-life-threatening syndrome to avoid unnecessary investigations.

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