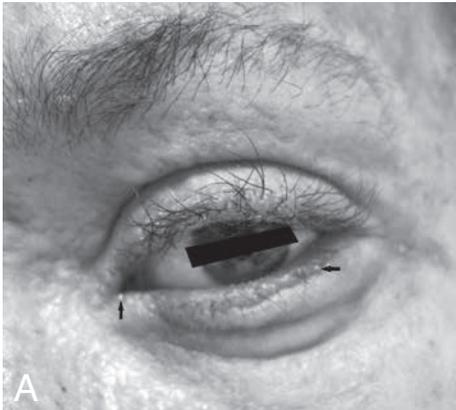


IMAGES IN CLINICAL RADIOLOGY



Bilateral selective amygdala calcifications: lipoid proteinosis

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A 14-year-old boy presented with progressive skin and mucous membrane changes including multiple papules on the eyelid margins. His parents also noted hoarseness since early childhood. His rashes emerged first when he was 1-month-old and healed by leaving scars. Voice distortion appeared at the age of 1. At the age of 5, he developed bilateral multiple papules on the eyelids. His sister also developed similar cutaneous and eyelid lesions as well as voice hoarseness.

On physical examination, bilateral beaded papules were present on the margins of eyelids (arrows in Fig. A). Also noted were widespread facial acneiform scars, thickening of the tongue associated with restricted tongue movement.

Comment

Lipoid proteinosis was firstly defined as "lipoidosis cutis et mucosae" by Urbach and Wiethe in 1929. The disorder has recently been shown to result from loss-of-function mutations in the extracellular matrix protein 1 gene on chromosome 1q21. It is rare autosomal recessive disorder that presents in early childhood with hoarseness, skin infiltration and thickening with beaded papules on eyelid margins, and facial acneiform or pox-like scars. Histological examination reveals widespread deposition of hyaline like material and disruption of basement membrane around vessels and at the dermal-epidermal junctions. The first symptom of the disease is hoarseness, which is attributed to laryngeal infiltration. It occurs in the majority of patients soon after birth. Congenital dystonia, congenital hypothyroidism and lipoid proteinosis should be included in differential diagnosis when hoarseness is seen in early childhood. The other clinical manifestations of LP may vary considerably between affected individuals. Multiple systemic involvement may occur at some individuals. Moniliform blepharosis is a pathognomonic finding and is seen in 50% of patients.

LP involves the central nervous system (CNS) infrequently. The amygdalae, hippocampus, parahippocampal gyrus and the striatum are the most commonly affected sites. CNS involvement mostly occurs as a result of accumulation of hippocampal perivascular calcium deposits. Patients with involvement of the amygdalae may experience symptoms related to limbic system dysfunction such as alteration in fear recognition, modulation of attention, perception, learning and memory. Dystonia is seen in some cases with striatal calcifications.

The essential imaging findings in LP are atypical intracranial calcifications which are mostly seen in medial temporal lobes, amygdala, hippocampus and parahippocampal gyrus. Bilateral horn or comma-shaped amygdala calcifications are almost pathognomonic (arrows in Fig. B, C) occurring in approximately 50% of cases.

Despite the progressive nature of the disease, the prognosis of LP patients is rather good. Laser microlaryngoscopy and dissection of the vocal cords may be performed to improve voice quality. Although intralesional heparin and oral dimethylsulphoxide and steroid therapies have been reported in the literature, there is currently no effective treatment. Only symptomatic therapeutic approaches are available.



Reference

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