

Delayed evolution: The chronological puzzle of APS- 1 diagnosis

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Abstract

Background: Autoimmune polyendocrinopathy syndrome type 1 is a rare autosomal recessive disorder characterized by autoimmune multiorgan attack. The disease is caused by mutations in the autoimmune regulator gene, resulting in defective AIRE protein, which is essential for self-tolerance. We describe a case of APS 1 with various endocrine and nonendocrine manifestations.

Case summary: A 30-year-old man presented with difficulty and pain during deglutition for 1 year. He also had a history of perioral paresthesia and tingling of the limbs. On examination, we found oropharyngeal candidiasis, carpopedal spasm, pigmentation of the lips and hard palate, yellow dystrophic nails, hypoplastic dental enamel and alopecia. On investigation, his serum levels of calcium and PTH were found to be extremely low, and S. phosphate was in the upper normal reference range. He was diagnosed as a case of primary adrenocortical insufficiency at the age of 26 and was on Hydrocortisone replacement. Despite adequate calcium supplementation, he developed repeated tetany and subsequently adrenocortical crisis requiring hospital admission.

Conclusion: APS type 1 is a complex syndrome. Onset typically begins in childhood, but sometimes it may present at a later age, as in this patient. Careful monitoring of patients with components of APS type 1 is necessary to reduce the risk of development of malignancy, mortality and morbidity. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S62]

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