

Cachexia: An Unsolved Puzzle

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Abstract: APS type 1 (APS - 1) is a rare, autosomal recessive inherited disorder encompassing chronic mucocutaneous candidiasis, hypoparathyroidism, and autoimmune adrenal insufficiency, with the presence of two of these being essential for diagnosis. APS - 1 primarily affects young patients, both males and females equally. The pathogenesis of the syndrome involves the development of T cells with a high affinity to auto antigens; a defect in the body's immune system fails to eliminate these T cells, and thus they bind to and attack the host's cells. Apart from the manifestations listed above, APS - 1 patients can also develop hypogonadism, vitiligo, chronic diarrhea and gastritis, autoimmune hepatitis, autoimmune hypothyroidism, type 1 diabetes mellitus, and pituitary failure.

Keywords: Autoimmune polyglandular syndrome, mucocutaneous candidiasis, T cells, cortisol, adrenal insufficiency

1. Introduction

Autoimmune polyglandular syndrome (APS) type 1, also called autoimmune polyendocrinopathy - candidiasis - ectodermal dystrophy/dysplasia (APECED) or Whitaker syndrome is defined as the presence of two or more of the following: Addison's disease, chronic muco - cutaneous candidiasis, and hypoparathyroidism [3]. It occurs in childhood with 1: 1 male to female ratio, an autosomal recessive pattern, and has an incidence of about 3/1, 000, 000 people. Although treatment is identical whether it occurs in isolation or as part of an APS, clinicians must be cautious to syndromic associations of autoimmune diseases to prevent delayed diagnosis.

2. Case

A 28-year-old man with no known comorbidities was admitted with progressive weight loss, fatiguability and progressive darkening of the skin for 1 year duration. On examination, BMI - 19.4 kg/m² and BP - 102/64 mm Hg. Examination revealed hyperpigmentation over the body and mucocutaneous candidiasis. Abdominal examination showed Liver is palpable 2cm below right costal margin.

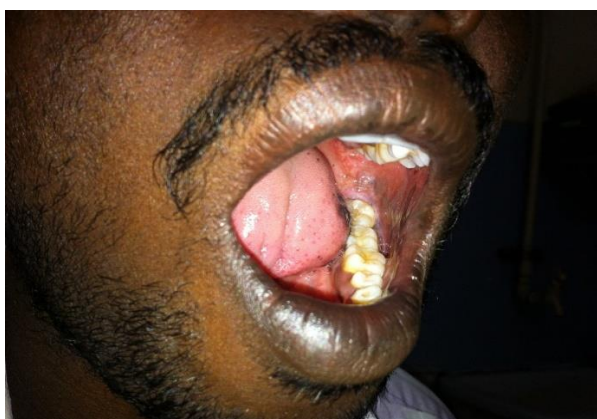


Figure 1: Mucocutaneous candidiasis



Figure 2: Hyperpigmentation

3. Observation

Routine blood investigations were within normal limits except anemia, hyponatremia and hyperkalemia. On further investigations showed low serum cortisol levels (3mcg/dL) and raised ACTH levels (1240pg/m) with bilateral adrenal masses in CT abdomen and evidence of granulomatous inflammation in histopathology. Patient was treated with parenteral hydrocortisone and intravenous saline and showed clinical improvement on follow up.

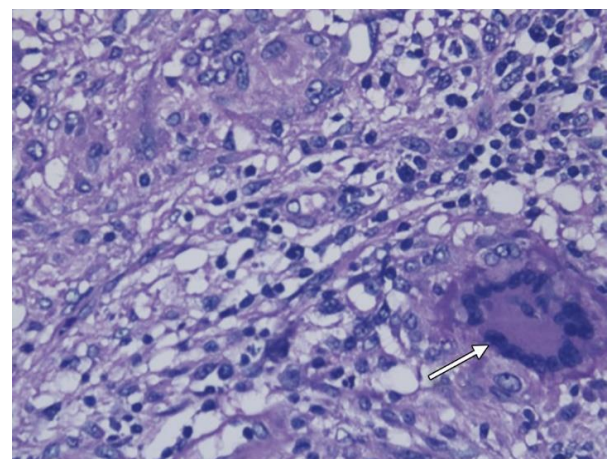


Figure 3: Granulomatous inflammation



Figure 4: Bilateral adrenal masses

4. Clinical Significance

Physicians should have a high index of suspicion for adrenal insufficiency while evaluating young patient with cachexia in the setting of hyperpigmentation. Since tuberculosis can lead to adrenal insufficiency, infections may be a major factor in the evolution of Autoimmune polyglandular syndrome. Patient education (sick day management) is the key to avoid hypocortisolemic crisis that increases the risk of mortality.

References

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