ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 04 | April - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

HYPERPIGMENTATION SYNDROME

Endocrinology

Mohammed Shanil P DM Cardiology Resident, Government Medical College, Kannur

Farsana Jasmin K MD Radiotherapy resident, Government Medical College, Kozhikode

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

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.

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/m^2 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.

Figure1:mucocutaneous candidiasis

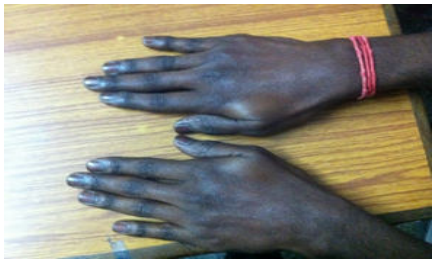

Figure 2:Hyperpigmentation

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.

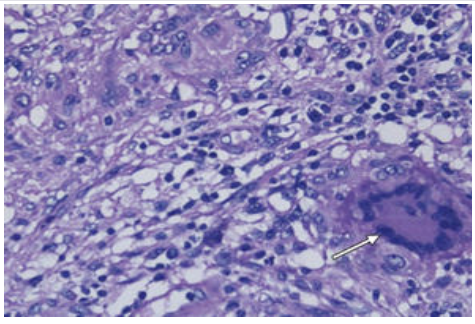

Figure 3-Granulomatous inflammation

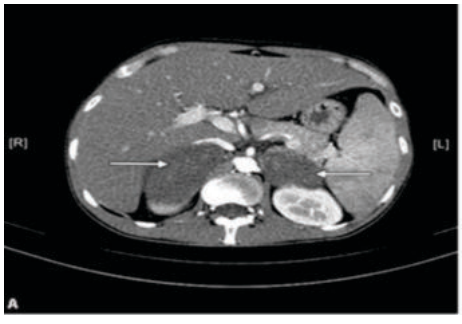

Figure 4-Bilateral adrenal masses

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

1 Ferré EMN, Schmitt MM, Lionakis MS. Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy. Front Pediatr 2021; 9: 723532, DOI: 10.3389/ fped. 2021.723532.
2 Zaidi G., et al. Autoimmune polyendocrine syndrome type 1 in an Indian cohort: a longitudinal study. Endocr Connect. 2017;6(5):289–296.
3 Livo Hetemäki,et al.Patients with autoimmune polyendocrine syndrome type 1 have an increased susceptibility to severe herpesvirus infections,Clinical Immunology,Volume 231,2021,108851,ISSN 1521-6616.
4 Sperling MA, Angelousi A, Yau M. Endotext [Internet] South Dartmouth (MA): MDText.com, Inc.; 2021. Autoimmune polyglandular syndromes.
5 Astley CM, Smith JR, Wassner AJ. Nelson Textbook of Paediatrics. Vol. 2. Philadelphia PA: Elsevier ; [Jul; 2022]. 2018. Autoimmune Polyglandular Syndrome Type 1; pp. 2937–2942.
6 Shaikh SB, Haji IM, Doddamani P, Rahman M. A Study of Autoimmune Polyglandular Syndrome (APS) in Patients with Type1 Diabetes Mellitus (T1DM) Followed Up at a Teritiary Care Hospital. J Clin Diagn Res. 2014 Feb;8(2):70-2. doi: 10.7860/JCDR/ 2014/7013.4011.