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Case Report

Autoimmune Polyglandular Syndrome Type-2 A Case Report on a Rare Disease

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ABSTRACT

Autoimmune polyglandular syndromes (APS) are rare disorders involving multiple endocrine and non-endocrine organs. These are often difficult to diagnose, as the clinical presentation of these is insidious. We present a case where a 29-year-old man presented to the Emergency Department in a state of altered sensorium with a history of focal seizures. His clinical presentation further included hypotension, malaise, and diplopia. A detailed workup revealed multiple endocrine gland involvement, and a diagnosis of APS was made.

Keywords: Autoimmune polyglandular syndrome, Addison's disease, Myasthenia gravis.

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INTRODUCTION

Autoimmune polyglandular syndromes (APS) destroy multiple endocrine and non-endocrine organs.[1] There are three types of APS; of these, APS type-2 is the most common, which may appear in incomplete and complete forms. The incidence of the complete form is only 1–2/100,000 per year.[2] However, an incomplete form that only partly meets the diagnostic criteria is relatively more common.[3] APS type-2 is characterized by the presence of Addison's disease along with autoimmune thyroid disease and diabetes mellitus in the majority of the cases.[4] Primary hypogonadism, myasthenia gravis, and celiac disease are also commonly observed in this syndrome. APS_2 is associated with HLA alleles and has numerous inheritance mechanisms, which may be diagnosed by genetic studies.[5] We present a case of autoimmune polyglandular syndrome type-2.

CASE REPORT

A 29-year-old gentleman with a known case of Hashimoto's thyroiditis (with positive anti-microsomal antibodies) and vitiligo for three years was brought to the Emergency Department with a one-day history of altered state of consciousness. He has also had recurrent focal seizures involving facial twitching and myoclonic jerks for the past three weeks. His attendants also reported that he had off-and-on diplopia along with generalized fatigue for the past two years. However, the cause of these symptoms was not established. His clinical examination showed a GCS of 13/15, Blood pressure of 90/60 mmHg and low volume pulse. He had dystonic posturing of all limbs with myoclonic jerks and hyperpigmented palmar creases. The patient was admitted, and his initial laboratory workup revealed normal baseline tests except for mildly raised potassium. His MRI Brain showed no signal abnormality. His CSF studies showed normal cell count and glucose, mildly increased proteins and raised IgG index. PCR for HSV virus RNA and CSF culture were both negative. An electroencephalogram was done that showed slowing with Delta waves. Keeping in view his presentation and investigation results, a presumptive diagnosis of Autoimmune Encephalitis was made. Autoimmune encephalitis panels, including NMDA, were tested, which came out to be positive. The patient was started on IV Methylprednisolone pulse therapy for five days. Over the next few days, the patient started improving. He regained consciousness, his blood pressure improved, and his dystonia and focal seizures settled. However, on discontinuation of steroid treatment, he experienced a postural drop of more than 20 mmHg of systolic Blood pressure. Because of low blood pressure, hyperpigmented creases and electrolyte abnormalities, a suspicion of Addison's disease was raised. His HbA1c was 3.9%, and his hormone profile revealed low serum cortisol and raised ACTH levels. Next up, the short synacthen test was done and turned out to be positive. He was started on IV hydrocortisone, which improved his blood pressure and general well-being.

Since the patient had evidence of multiple autoimmune disorders, including vitiligo, Hashimoto's thyroiditis, Addison's disease and autoimmune encephalitis, a diagnosis of APS_2 was suspected and a Complete antibody and fertility profile were done,

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which revealed positive ANA and normal LH, FSH and Testosterone. Anti-acetylcholine receptor antibodies were positive, indicating Myasthenia gravis and explaining his longstanding diplopia and fatigue.

The patient was started on pyridostigmine and azathioprine for myasthenia gravis. Maintenance doses of oral prednisolone for Addison's disease and thyroxine for Hashimoto's thyroiditis were also given. His diplopia settled over the next few weeks. The patient was advised to follow up with a laboratory workup.

DISCUSSION

Autoimmune polyglandular syndrome type-2 (APS_2) is a rare disorder where Addison's disease occurs concurrently with other endocrine and non-endocrine autoimmune disorders. These include Thyroid Disease, Diabetes Mellitus type-I, Myasthenia Gravis, Celiac Disease and Primary Hypogonadism forming diagnostic criteria.[6,7] Our patient had three of these illnesses (Addison Disease, Myasthenia Gravis and Hypothyroidism) along with vitiligo and autoimmune encephalitis.

Diagnosis of APS is often difficult due to the non-specific nature of symptoms and signs that do not appear simultaneously. Stress states like surgery, infection, pregnancy or trauma may unmask the underlying syndrome and may prove to be fatal for affected patients.[8] However, since antibodies denoting different components of this syndrome may appear well before clinical symptoms develop, these disorders can be identified early on the basis of suspicion, and patients can be managed accordingly. Accurate and timely diagnosis with subsequent appropriate hormone replacement therapy is often life-saving and reduces the physical, psychological and financial burden of the patients.[9]

Conflict of Interest: None.

Authors' Contribution

Following authors have made substantial contributions to the manuscript as under:

WHK & UZF: Conception, study design, drafting the manuscript, approval of the final version to be published.

KA & UN: Data acquisition, critical review, approval of the final version to be published.

manuscript, approval of the final version to be published.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

REFERENCES

1. Perniola R, Fierabracci A, Falorni A. Autoimmune Addison's disease as part of the autoimmune polyglandular syndrome type 1: historical overview and current evidence. Front Immunol 2021; 12: 298. https://doi.org/10.3389/fimmu.2021.606860.
2. Banerjee M, Mondal SK, Maisnam I, Mukherjee AK. Jaundice and anaemia as presenting features of an incomplete autoimmune polyglandular syndrome type II. BMJ Case Rep 2019; 12(4): e228490. https://doi.org/10.1136/bcr-2018-228490.
3. PV AA, Kumar J, Unnikrishnan D, Raj R. Case of autoimmune polyglandular syndrome type-2: how we uncovered the diagnosis. BMJ Case Rep 2019; 12(2): e227187. https://doi.org/10.1136%2Fbcr-2018-227187.
4. Bain A, Stewart M, Mwamure P, Nirmalaraj K. Addison's disease in a patient with hypothyroidism: autoimmune polyglandular syndrome type 2. Case Rep 2015; 2015: bcr2015210506. https://doi.org/10.1136/bcr-2015-210506.
5. Dittmar M, Kahaly GJ. Polyglandular autoimmune syndromes: immunogenetics and long-term follow-up. J Clin. Endocrinol Metab 2003; 88(7): 2983-2992. https://doi.org/10.1210/jc.2002-021845.
6. Betterle C, Lazzarotto F, Presotto F. Autoimmune polyglandular syndrome Type-2: the tip of an iceberg? Clin Exp Immunol 2004 ;137(2):225-33. https://doi.org/10.1111/j.1365-2249.2004.02561.x.
7. Husebye ES, Anderson MS, Kämpe O. Autoimmune polyendocrine syndromes. NEJM. 2018; 378(12): 1132-1141. https://doi.org/10.1056/nejmra1713301.
8. Prylutskyi O, Prylutska O, Degonskyi A. A case of autoimmune polyglandular syndrome. ype 2 associated with atypical form of Scleromyxedema. Ethiop J Health Sci 2016; 26(5): 503.
9. Kahaly GJ, Frommer L. Polyglandular autoimmune syndromes. J Endocrinol Invest 2018; 41(1): 91-98. https://doi.org/10.1007/s40618-017-0740-9.