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A CASE OF SCHMIDT'S SYNDROME

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ABSTRACT

The polyglandular autoimmune syndromes are rare endocrinopathies characterized by the coexistence of at least two glandular autoimmune mediated diseases. There are two major subtypes of poly glandular autoimmune syndromes. Type 1 polyglandular autoimmune syndrome consists of Addison's disease, hyperparathyroidism and mucocutaneous candidiasis. Type 2 polyglandular autoimmune syndrome consists of Addison's disease, autoimmune thyroid disease and type 1 diabetes. Type 2 polyglandular autoimmune syndrome is also known as schmidt's syndrome.

INTRODUCTION

The polyglandular autoimmune syndromes are rare endocrinopathies characterized by the coexistence of at least two glandular autoimmune mediated diseases. There are two major subtypes of poly glandular autoimmune syndromes. Type 1 polyglandular autoimmune syndrome consists of Addison's disease, hyperparathyroidism and mucocutaneous candidiasis. Type 2 polyglandular autoimmune syndrome consists of Addison's disease, autoimmune thyroid disease and type 1 diabetes. Type 2 polyglandular autoimmune syndrome is also known as schmidt's syndrome. Schmidt's syndrome is a rare autosomal dominant disease that is more common in middle-aged females with a prevalence of 1.4 to 4.5 per 100,000 population.

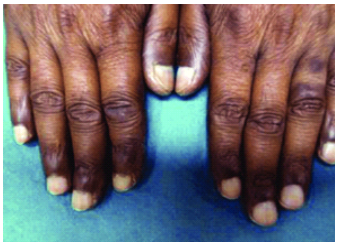

Figure 1: Hyperpigmentation over the Dorsum of the hand

CASE DESCRIPTION

A 52 year old female presented with complaints of decreased ability to do daily activities, abdominal pain, vomiting, insomnia, anorexia, significant weight loss and generalized fatigability since 2 months. There were no complaints of dysnea, fever, headache and seizures. She is a known case of hashimoto's thiroiditis on tablet thyroxine 50 mcg per day, and known case of type 1 diabetes mellitus since 24 years on insulin.

On examination pallor present, bilateral pedal edema was present, hyperpigmentation present all over the body and face. Blood pressure in supine position is 90/65 mmhg and in standing position 70/50 mmhg suggesting orthostatic hypotension, heart rate is 90 /min, respiratory rate is 19/min. Systemic examination is unremarkable. Free T3 was low and thyroid stimulating hormone was elevated. Anti thyroid antibodies were present. 8 AM plasma cortisol was 0.2 microgram/dl. FSH and ACTH was elevated. Based on the history, examination and laboratory findings patient was diagnosed to have primary adrenal in sufficiency, autoimmune hypothyroidism and type 1 diabetes mellitus.

Diagnosis of type 2 polyglandular autoimmune syndrome (schmidt's syndrome) was made. She was treated initially with intravenous normal saline and then started on hydrocortisone 100mg trice daily . She was improved symptomatically and discharged. During her follow-up after 1 month she was doing well.

DISCUSSION

APS-1 is defined by the combination of at least two of three cardinal component diseases including chronic mucocutaneous candidiasis, hypoparathyroidism, and primary adrenal insufficiency. Yeast infections of the mouth and nails are frequently reported as the first obvious symptoms, and it is more common in infants. Schmidt's syndrome, also known as APS type 2 (APS-2), is characterised by the coexistence of thyroid autoimmune disease, type 1 diabetes, and primary hypogonadism with Addison's disease. There are various disease manifestations for this condition, and familial aggregation is present. The cause of Schmidt's syndrome is not well understood, but it frequently appears after an unusual immune response. The major histocompatibility complex type 2, specifically DR3-DQ2 and DR4-DQ8, has been found to be linked to Schmidt's syndrome. The pattern of inheritance is autosomal dominance with variable expressivity and a female-to-male ratio of 3-4:1,it occurs in the third or fourth decade of life. About 50% of patients initially experience adrenal insufficiency, with about 30% and 20% of patients also simultaneously experiencing diabetes mellitus and autoimmune thyroid disease respectively . This syndrome has been linked to vitiligo, myasthenia gravis, Sjögren's syndrome, rheumatoid arthritis, and primary antiphospholipid syndrome, which are all non-endocrine autoimmune diseases. Relatives who have one or more symptoms of the syndrome should also be screened and diagnosed with the condition. Patients with autoimmune thyroid disease or diabetes mellitus who have adrenal autoantibodies but not yet developed adrenal insufficiency should also be screened and diagnosed.

CONCLUSIONS

Schimdt's syndrome is a rare disease, but it can be fatal if it is not identified and treated in a timely manner. It is advised that every patient with idiopathic endocrine insufficiency be

examined for insufficiencies of other endocrine organs due to the possibility of atypical presentations of such a disorder. For an early diagnosis of Schmidt's syndrome and suitable hormone replacement therapy, a high index of suspicion is required.

REFERENCES:

[1] Barker JM, Gottlieb PA, Eisenbarth GS. In: Williams Textbook of Endocrinology.Koronenberg H, editor. Philadelphia: Saunders: 2008. The immunoendocrinopathy syndromes. pp. 1754–55.
[2] Neufeld M, Maclaren NK, Blizzard RM. Two types of autoimmune Addison's disease associated with different polyglandular autoimmune (PGA) syndromes. Medicine. 1981;60:355.
[3] Trence DL, Morley JE, Handwerger BS. Polyglandular autoimmune syndromes. Am J Med. 1984;77:107.
[4] Dittmar M, Kahaly GJ. Polyglandular autoimmune syndromes: immunogenetics and long-term follow-up. J Clin Endocrinol Metab. 2003;88:2983.
[5] Laureti S, Vecchi L, Santeusanio F, Falorni A. Is the prevalence of Addison's disease underestimated? J Clin Endocrinol Metab. 1999;84(5):1762.
[6] Maurer A, Schwarting A, Kahaly GJ. [Polyglandular autoimmune syndromes]. Z Rheumatol. 2011;70(9):752-754, 756-759.
[7] Gupta AN, Nagri SK. Schmidt's syndrome - Case report. Australas Med J. 2012;5(6):292-295.
[8] Weiler FG, Dias-da-Silva MR, Lazaretti-Castro M. Autoimmune polyendocrine syndrome type 1: case report and review of literature. Arq Bras Endocrinol Metabol. 2012;56(1):54-66.