CASE REPORT

Autoimmune Polyglandular Syndrome Type 2: An Unusual Presentation

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Abstract- Autoimmune polyglandular syndrome (APS) type 2 is characterized by the presence of Addison's disease, in association with autoimmune thyroid disease and/or type 1 diabetes mellitus. APS type 2 occurs most often in middle aged females and is rare in children. Here an 11 year old boy is reported with Addison's disease who developed symptom's of diabetes mellitus, goiter, malabsorption, macrocytic anemia and keratitis. APS type 2 occurs most often in middle aged females and is quite rare in children but one should think to autoimmune poly glandular syndrome type II in patient at any age especially in patients with Addison's disease.

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Key words: Polyendocrinopathies, autoimmune; Addison disease; diabetes mellitus

Introduction

Autoimmune polyendocrine syndrome type II (APS:II) is more common than APSI (1). It is a rare syndrome which may occur at any age and in both sexes but it is most common in middle aged females (2) and very rare in childhood (3). APS type 2, also known as schmidt's syndrome, is condition occurring with a prevalence of 1.4-2.0 per 100000 inhabitants (4). APS 2 is usually defined by the occurrence in the same patient of two or more of the followings: primary adrenal insufficiency (Addison's disease), Grave's disease, autoimmune thyroiditis, type 1 diabetes mellitus (1,5,6). Primary hypogonadism, myasthenia gravis, celiac disease, vitiligo, alopecia and serositis. Pernicious anemia also occurs with increased frequency in patients with this syndrome. Rare autoimmune disorders can also occur for example, a woman has been reported of autoimmune hyperparathyroidism in association with autoantibodies to calcium receptor who also had multiple autoimmune diseases including autoimmune hypophysitis (1,7).

Addison disease may occur as a component of 2 autoimmune poly endocrinopathy syndrome. Type 1 autoimmune poly endocrinopathy (APS-1) consists of chronic mucocutanous candiadiasis, hypoparathyroidism and Addison's disease. Type II autoimmune polyendocrinopathy (APS-2) consists of Addison disease associated with autoimmune thyroid disease (Schmidt syndrome) and type 1 diabetes (carpenter syndrome) (8).

We report an 11years old boy with Addison disease sequently development diabet mellitus, hypothyroidism.

Case Presentation

An 11 years old boy a known case of Addison's disease who was on hydrocortion and fludrocortison replacement therapy for 4 year admitted with polyuria and polydepsia without any follow up during 2 years (Figure 1). In physical examination, his height was 126cm and weight 21 kg both below the third percentile darkly pigmentd, ptosis and corneal opacity in the right eye with a firm goiter (grade II) were detected.

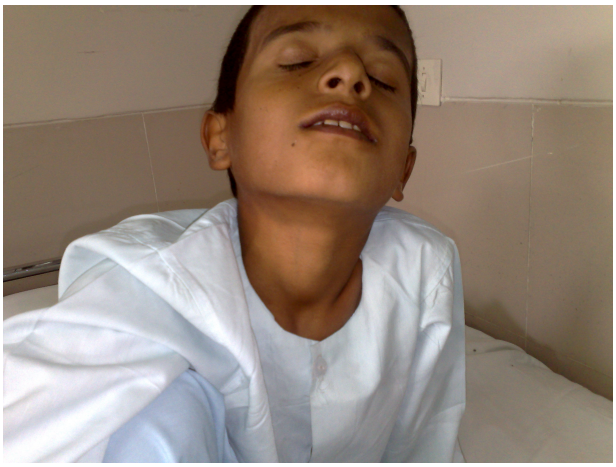

Figure 1. Polyglandular type 2 patient with Goiter and hyperpigmentation

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Table 1. Laboratory finding of patient

	Patient	Normal range
Hb	9.9	11-13
MCV	110	70-100
C3	0.988%	0.89-1.87
ANA	4.9 U/ml	≤ 10
Ds-DNA	10.3 iu/ml	Up to 50
ACLA	7.3 Gplu/ml	<8
P-ANCA	1.2	3.1
CANCA	0.1	<0.4
T4	8.4 µg/dl	8-15
TSH	5.73	0.3-5.1
Tissue total . transglutamines (IgA)	3.2 u/ml	Up to 12
Total IgA Elisa	2.21 g/l	0.71-3.6
Fasting blood glucose	174	75-115
Calcium	9.7	8.5-10.4
Phosphorous	3.9	2.5-4.5
Tpo. Ab Microsomal	564.1 Iu/mL	>40
Anti TGAb	2513 Iu/mL	Up to 25

He was pre pubertal; pubic and axillary hair tanner stage 1. (Even though he took irregular medication such as hydrocortisone and fludrocortison.) Laboratory data is reviewed in Table 1. Laboratory investigation showed macrocytic anemia, subclinical hypothyroidism T4:8.4, TSH:5.7 with positive antithyroid antibodies, anti TPO antibody: 564.1, anti TgAb: 25.3 high blood glucose: 173 mg/dl, negative tissue transglutamines antibody:3.2 and normal liver function test.

Discussion

APS type 2 occurs most often in middle aged females. Its presentation during childhood is uncommon Schmidt syndrome is also a rare condition occurring with a prevalence of 1.4-2.0 per 100,000 in the population presents in middle-aged women. Mostly (female- male ratio ranges from (2-3.7 to 1) and is very rare in childhood (2,3). In patients with APS type 2, Addison's disease is present in 100% of the cases, autoimmune thyroid disease in 69-82% and type 1 diabetes mellitus in 30-52% of the syndrome (5-6). Other autoimmune diseases that are not the major component may be present in APS type 2: hypergonadotropic hypogonadism (4-9% of patients), chronic hepatitis (4% of patients), alopecia (1-4% of patients) chronic atrophic gastritis with or without pernicious anemia (4.5-11% of patients) and hypophysitis (2).

In this communication we report an 11 old boy with Addison's disease autoimmune thyroiditis and DM type 1, macrocytic anemia and keratitis. This condition is very rare in childhood as far as we know there is only report if another child with this syndrome from Greece (2007) (7). In comparison between our presented case with previous reported case, diabetes mellitus was found in our case but it was absent in previous case, but growth hormone deficiency secondary to autoimmune hypophysitis was determined in previous case but it was not possible to the determine the level of growth hormone in this case because he had not an follow up for 2 years.

The age range of two presented case are also different (11 years old v.s 12 years). In conclusion, one should think of autoimmune polyglandular in all age, especially with presence Addison's disease and we need to careful observation in these patients.

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