

Unusual Case of Autoimmune Polyglandular Syndrome (Case Report)

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Background/Aims: Incidence of Autoimmune disease dramatically increases in children and adolescents in the past decades. But in other hand case of APS is rare. Signs and symptoms appear with different combination during the lifespan in different patients. Here we report case of APS syndrome with unusual presentation.

Case presentation: Patient 16,4 years old boy with diabetes mellitus since the age of 2,6 years. Born term, healthy, with no previous medical problems. Mother died at age 40, with gastric cancer, after that he had psychological problems. At the age of 12,6 years were diagnosed hypothyroidism-autoimmune thyroiditis, after that he received L-thyroxine. At the age of 14 appears problems with the skin, that at first was diagnosed as Psoriasis. Last hospitalization - at the age of 16,4 years with clinical features of severe anemia. Based on clinical and paraclinical data analyses and catamnestic observation (The child had the same anemia at the age of 13 years, which was cut with appropriate treatment) this case was diagnosed with megaloblastic anemia caused by a B-12 deficiency. The situation was stabilized after therapy of vitamin B12, without glucocorticoids. After that, the boy was consulted with the dermatologist and diagnoses of psoriasis were changed with cutaneous candidiasis after diagnostic test. A physical examination shows retardation of growth (since 11 years) weight -32kg(-4,91SDS), height-147cm(-3,3SDS), Tanner stage 2 pubic hair development, testes<4ml, penile length-5cm. His bone age was 13 years by the standards of Greulich and Pyle at a chronological age of 16 years. Electrolytes: K, Na, Ca, P-were normal. . In ultrasound liver is always enlarged since 11 years by 2-4cm, ALT-65U/L, AST-36U/L, Cortisol-531,48ng/ml. Mean HbA1c-7,3%, TSH-2,4mIU/l, Anti-TPO-216,5 IU/ml, Anti-Tg-76,4 IU/ml. Insulin daily requirement- 1,1U/kg, L-thyroxine-100mcg/day.

Age	Diseases
2 years 6 months	Diabetes mellitus
Since 11 years	Hepatomegaly
Since 11 years	Retardation of growth
12 years 6 months	Hypothyroidism-autoimmune thyroiditis
13 years	Megaloblastic anemia
14 years	Cutaneous candidiasis
	Hypogonadism

Hematologic investigations:

- Complete blood count: severe anaemia: hemoglobin concentration Hb-5,2g/dl, Ht-16%, anaemia is macrocytic(MCV>105fL), reticulocyt count is low, total white cell and platelet counts reduced: tr-52, leik-2,4(Pancytopenia).
- Blood smear: megalocytes, macrocytes, neutrophils with show hypersegmented nuclei.
- Serum unconjugated bilirubin and lactate dehydrogenase are raised.
- Iron metabolism, serum folic acid-normal, Serum B12 is low<51ng/l.
- Bone marrow: hypercellular with large erythroblasts, giant and abnormally shaped metamyelocytes.

Conclusions:

Conclusion: Patient 16 years has DM, AIT, Pernicious anemia, cutaneous candidiasis, hypogonadism, that we give a possibility to diagnose APS. We think, that it is type 1 syndrome, although genetic testing for the AIRE gene was not made because of financial problems. Early identification of this syndrome gives us a chance to for early detection and proper management of associated conditions and its complication with maximal efficacy.

References:

- References:
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