APECED Syndrome in Childhood: Rare Clinical Presentations to Keep in Mind

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APECED Syndrome; is a rare, autosomal recessive disease caused by mutations in the autoimmun regulatuar AIRE gene on the chromosome 21. Although classical triad is mucocutaneous candidiasis, hypoparathyroidism and adrenal insufficiency; endocrine / non-endocrine involvement may also be seen (1-2). Possible involvement should be evaluated without any clinical signs. We report a case of APECED syndrome with autoimmune hypophysitis secondary to growth hormone deficiency and autoimmune asplenia.

CASE PRESENTATION

History :A 12.5-year-old male patient diagnosed with hypoparathyroidism in another center and whose previously **unidentified IVS3-3C> G (c.464-3C> G) homozygote mutation** in the AIRE gene was referred to our clinic. When he presented with a history of mouth trush and fatigue when he was eight years old; it was learned that calcitriol treatment was started with the diagnosis of hypoparathyroidism.

Our patient was born with IVF at a weight of 2900 gr. The mother of our patient was LT4 due to hypothyroidism and his father was OAD due to type 2 DM. Consanguineous marriage between parents.

Physical Exam:
Weight : 35,9 kg (-1,86 sds)
Height:140 cm (-2,65 sds)
BMI : 18,32 kg/m2 (SDS: -0,51)
Bone age::10 years
Growth Velocity: **2,5 cm years*****
-Diffuse vitiligo
-Hypopigmentation in the hair,
-Photophobia, 8 ml testes.

Laboratory and Imaging:
CBC:Anemia
Peripheral Blood Smear: Acanthosis
Abdomen USG: SpleenØ
Spleen Scinthigraphy:Non-functional spleen

Autoimmune Hemolytic Anemia+Autoimmune Asplenia

Penisilin Propylaxy and protective vaccine program

Laboratory and Imaging:
Ca:9,5 mg/dL,
P:6,2 mg/dL;
ALP:197 IU/L,
PTH<2,5 pg/ml
Hypercalciurea+
Medular Nephrocalcinosis

Oral calcium treatment was discontinued.Calcitriol dose was reduced.

Laboratory and Imaging:

TFT: TSH: 2,087 µIU/mL FT4: 1,24ng/dL FT3:4,07 pg/mL
Anti TPO:+ Anti TG negative Thyroid USG: Normal
*****Autoimmune Thyroiditis**

Bazal Cortizol: 8 µg/dL ACTH: 568pg/mL
Standard Dose ACTH Test:
Bazal Cortizol :13,0 µg/dL
Peak Cortizol :13,3 µg/dL
ACTH: 792 pg/mL
*****Autoimmune Primer Adrenal Failure**
10mg/m2/day-Hydrocortisone

FSH: 1,61 mIU/mL
LH: 1,83 mIU/mL
Total Testosteron: 130,39 ng/dL
Prolaktin : 9,39 ng/mL
IGF1:114 ng/dl
IGFBP3 : 3400 ng/

Laboratory and Imaging:
Autoimmune Disease Screening:
(Islet) antibody 2 (IA2):)+
ANA : Negative Anti ds DNA (mikroelisa): Negative
Anti-GAD :+
Gastrin 328*** Anti parietal antibody: Negative
Endoscopy: Normal Celiak Serology : Negative
Ophthalmologic examination: Mild chorioretinal atrophy in the retina, photosensitivity compatible with albinism.

Pitutiary MRG: Sella tursika sizes are normal. The anterior gland height of the pituitary is 5 mm. An area measured 3 mm in the right lateral section of the gland is more hypointense than the gland in the pre and post contrast series. Adenoma?
Infindibulum thickness 1.6 mm.

*****Autoimmune Hyphopysitis**

*****Autoimmune Hyphopysitis**
Peak L-dopa test:7,83ng/dl ** Peak Clonidine test :1,27 ng/dl ** Growth Hormone Therapy was started. Growth Velocity: 2.1 cm / 2 months

Conclusion: APECED syndrome has a wide clinical spectrum. Keeping rare clinical presentations in mind will reduce the morbidity and mortality of the disease

Refereences:
1- Weiler, Fernanda Guimarães, Magnus R. Dias-da-Silva, and Marise Lazaretti-Castro. "Autoimmune polyendocrine syndrome type 1: case report and review of literature." *Arquivos Brasileiros de Endocrinologia & Metabologia* 56.1 (2012): 54-66.
2-Husebye, E. S., Anderson, M. S., & Kämpe, O. (2018). Autoimmune polyendocrine syndromes. *New England Journal of Medicine*, *378*(12), 1132-1141.

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