AUTOIMMUNE POLYENDOCRINOPATHY-CANDIDIASIS-ECTODERMAL DYSTROPHY: A CASE REPORT

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Introduction

▪Autoimmune polyendocrinopathy, candidiasis and ectodermal dystrophy (APECED)

- •A rare hereditary disorder
- •Autoimmun manifestations affecting both endocrine and non-endocrine tissues
- •It is caused by mutations in the autoimmune regulatory (AIRE) gene
- •Defined by the presence of two of the three major components:
  - ➢Chronic mucocutaneous candidiasis
  - ➢Autoimmune hypoparathyroidism
  - ➢Addison's disease

▪Clinical manifestations may be developed during early years of life and may continuous throughout decades.

▪Moreover, the syndrome also includes many other autoimmune diseases such as type 1 diabetes mellitus, idiopatic trombositopenic purpura, pernicious anemia, chronic active hepatitis, vitiligo, alopesia, Hashimoto thyroiditis and sistemic lupus eritematosus.

▪Here, we present a case with APECED from a consanguineous family, who had mucocutaneous candidiasis, hypoparathyroidism, Addison's disease, Hashimoto thyroiditis, pernisious anemia and trombositopenia.

Case Report

▪Our case was a six years old boy who was presented to our outpatient endocrinology clinic with nause and vomiting, fatique, hypopigmentation, constipation and diarrhea attackes.

▪He had a convulsion due to hypocalcemia and hypoparathyroidism.

▪On physical examination:

- ➢Mucocutaneous candidiasis
- ➢Alopesia
- ➢Teeth-nail deformations
- ➢Normal vital signs.

▪On laboratory examination:

- ➢Anemia
- ➢Trombositopenia
- ➢Hypoglycemia
- ➢Hyponatremia

▪The results of ACTH stimulating test confirmed primary adrenal insuffiency.

▪Genomic DNA from the periferal blood lymphocytes was extracted with QIA amp DNA Blood Mini Kit (Qiagen GMBH, Hilden, Germany) using standard procedures. AIRE gene mutation analyses has demostrated a homozygous missense mutation p.Arg15 His (c.44G>A) in exon 1. Mutation analyses of the both parents have revealed heterozygous mutation p.Arg15 His (c.44G>A) in exon 1.

Conclusion

▪Although first clinical manifestation of APECED usually begin in childhood, appearence order of other components might be delayed to make diagnosis more challenging.

▪In case, patients might be undiagnosed or misdiagnosed. Clinicians should be aware of this antity in terms of endocrine and non endocrine problems, because of the broad clinical spectrum.

▪In many cases, the diagnosis should be considered presenting at least one of the major clinical manifestations because of its high morbidity and mortality.

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