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 autoimmun 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, idiopathic trombositopenic purpura, pernicous anemia, chronic active hepatitis, vitiligo, alopecia, 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, fatigue, hypopigmentation, constipation and diarrhea attakes.
- He had a convulsion due to hypocalcemia and hypoparathyroidism.
- On physical examination:
  - Mucocutaneous candidiasis
  - Alopecia
  - 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.