Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy is a rare monogenic autosomal recessive disease known for the triad of the major components:
- Hypoparathyroidism
- Primary adrenocortical insufficiency
- Chronic mucocutaneous candidiasis.
Minor components such as gastrointestinal manifestations, ectodermal manifestations and others endocrinological manifestations may be present.

We report two siblings affected by APECED with the same genotype (AIRE 260 T>C; 967-979 del) but with extremely different phenotypes. Hypoparathyroidism and alopecia are the only two components they have in common.

### MALE 19 Y OLD

**Major manifestations:**
- Hypoparathyroidism
- Primary adrenocortical insufficiency
- Alopecia
- Postural acne on the face

**Other:** Preclinical autoimmune insulitis (GADA Ab positive without glycaemia alterations)

**Disease course:** Stable

**Therapy:** Teriparatide, calcium, fluordrocortisone and hydrocortisone.

### FEMALE 14 Y OLD

**Major Manifestations:**
- Hypoparathyroidism
- Vitiligo
- Alopecia
- Growth and pubertal delay
- Ectodermal dystrophy

**Other:** Preclinical autoimmune insulitis (GADA Ab positive without glycaemia alterations)

**Disease course:** At the age of 12 abdominal pain, chronic diarrhea and hypocholic feces occurred. Esophagogastroduodenoscopy and a colonoscopy (EGDS) were performed showing normal appearing mucosa and moderate inflammation.

**The lack of EECs was the only abnormality described.**

**Therapy:** Teriparatide, calcium. A trial with oral budesonide has been proposed for autoimmune enteropathy.

### CONCLUSIONS

APECED is a monogenic disease, but its expressivity can be extremely variable even in two identical genotypes in the same family. Chronic diarrhea may be due to several causes such as exocrine pancreatic insufficiency (EPI), autoimmune enteropathy (AE), lactose intolerance and celiac disease. Autoimmune enteropathy should be taken into consideration when abdominal pain and diarrhea occur and loss of EEC cells may be the only abnormality described. Oral immunosuppressive therapy with budesonide in the patient described allowed to improved symptoms.

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**REFERENCES**

2. N. Roifman et al. Clinical manifestations and management of patients with autoimmune polyendocrine syndrome type 1. Journal of Internal Medicine, 205(5), 514-29.