AUTOIMMUNE POLYENDOCRINE SYNDROME TYPE I: A NEUROENDOCRINE MULTI-SYSTEMIC DISEASE WITH A VARIABLE EXPRESSIVITY

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A P E C E D

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 candidosis.**

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
- Postular acne on the face

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

Disease course: Stable
Therapy: Teriparatide, calcium, fludrocortisone and hydrocortisone.

FEMALE 14 Y OLD

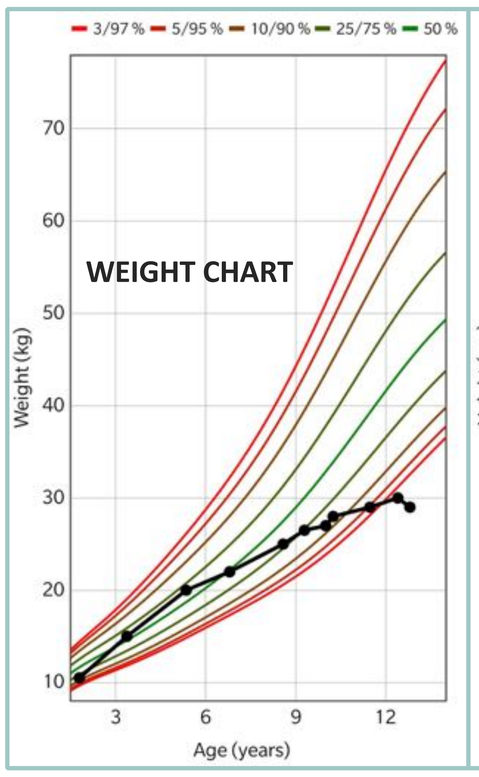
Major Manifestations:
- Hypoparathyroidism
- Vitiligo
- Alopecia
- Growth and pubertal delay
- Ectodermal dystrophy

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

Disease course: At the age of 12 abdominal pain, chronic diarrhea and hypocholic feces occured. 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.

	MALE 19 Y OLD	FEMALE 14 Y OLD
Hypoparathyroidism	+	+
Primary adrenocortical insufficiency	+	-
Chronic mucocutaneous candidosis	-	+/-
Ectodermal dystrophy	-	+
Autoimmune insulitis	+/-	+/-
Vitiligo	-	+
Alopecia	+	+
Diarrhea	-	+
Growth delay	-	+
Pubertal Delay	-	+



The CDC growth chart of female height and weight showing growth retardation and short stature

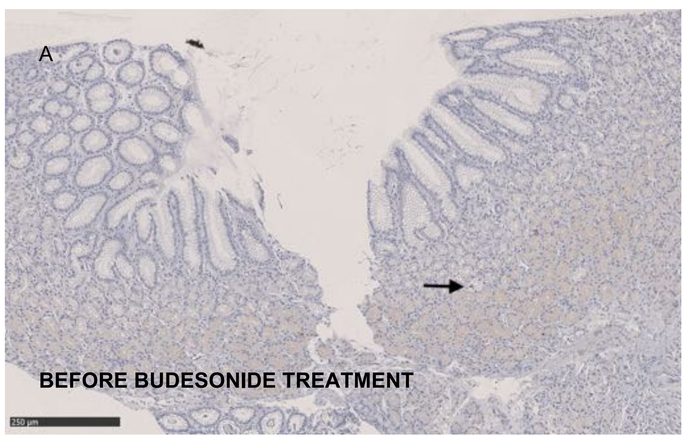
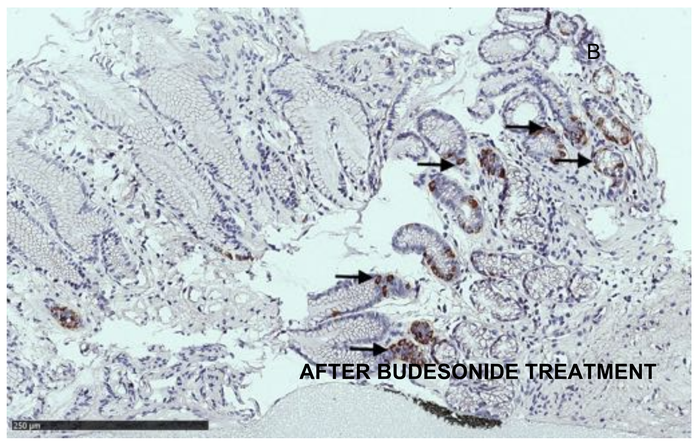


FIGURE A – B EGDs shows paucity of EEC cells in the colon and total absence in the stomach and duodenum.
After oral immunosuppressive treatment, EGDs shows new EEC cells in stomach.



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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