Unusual clinical presentation of autoimmune polyendocrinopathy type 1

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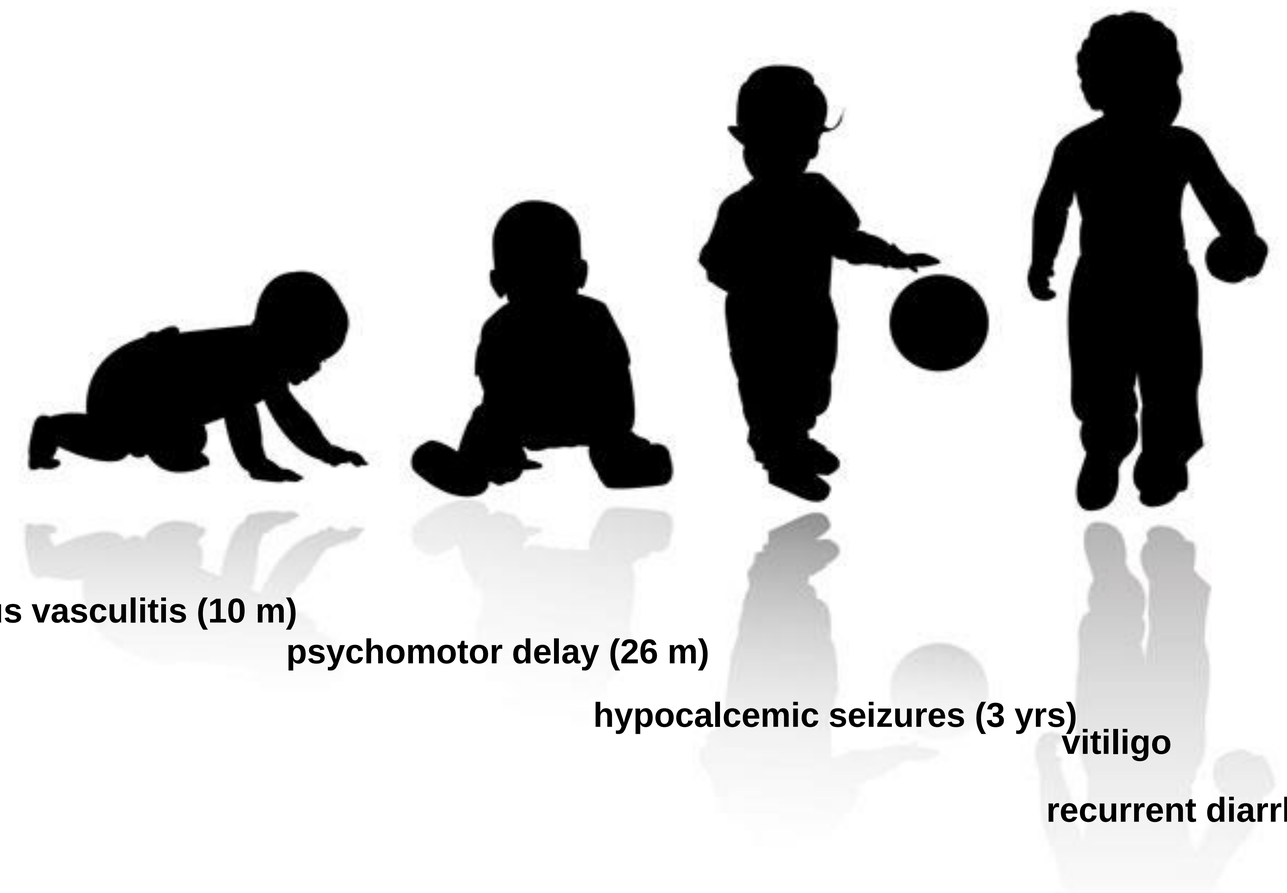
Background: Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED) or autoimmune polyendocrinopathy type 1 (APS-1) is a rare monogenic autosomal recessive disease due to pathogenic variants in the *AIRE* gene. APECED usually begins during early childhood with chronic mucocutaneous candidiasis (CMC), followed by hypoparathyroidism (HP) and Addison's disease (AD); however, other endocrine and non-endocrine components may occur with a different prevalence.

Clinical case We report on a boy affected by APS-1 with unusual clinical presentation.



Diagnostic suspicion

Autoimmune vasculitis

Autism

Hypoparathyroidism

Chr 22q deletion: negative

CGH array: negative

Autoimmune hypoparathyroidism

Polyendocrinopathy

Work up

Autoantibody profile (OS and NOS):	negative	
PTH	<1	
Magnesium	1.66	
Vitamin 25 OH D	20.2	
Autoantibodies		
Parathyroid:	negative	
TPO/TG:		**positive**
21 OH hydroxylase:		**positive**
Cortisol		148
ACTH		15
Na		141
K		5,1

Treatment

calcium	1 g/d
magnesium	1.5 g/d
Calcifediol	0,8 mcg/d
Teriparatide	2 U/d

AIRE gene analysis

The *AIRE* gene analysis showed a compound heterozigosis with a frameshift **(c.967_979del13)** and a potential causative missense mutations **(c.47C>T)** inherited from non consanguineous parents.

Discussion:

Our case showed uncommon features that anticipated common signs of APS 1 :

1. rashes, with the histological characteristics of vasculitis

2. neurological alterations interpreted as autism spectrum disorders,

After the substitutive treatment the patient did not repeat hypocalcemic seizures and showed a significant improvement of his neuromotor and behavior development

Conclusion: The clinical picture of APS-1 may be characterized by rare or atypical isolated or immune-mediated autoimmune manifestations , even years before the beginning of the classical components of the disease.