Psychiatric Manifestations of Autoimmune Polyendocrine Syndrome in Adolescence - A Case Report

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INTRODUCTION

- Autoimmune polyendocrine syndrome (APS) is an autosomal recessive disorder caused by defects in autoimmune regulator (AIRE) gene. While the AIRE gene can only affect certain hormone-producing glands, the presentation can involve both endocrine and non-endocrine manifestations (1, 3).
- We describe a case of an adolescent girl with autoimmune encephalitis and myelopathy secondary to APS, manifesting with psychiatric symptoms.

DIVERSITY, EQUITY, INCLUSION

APS is most prevalent among certain ethnic groups, especially Iranian and Persian Jews in which there is a frequency of 1 case per 9,000 population. APS-1 is much less common in Western European countries. Women more commonly get sick with APS-1, with a ratio of up to 2.4:1, female to male ratio (1). Including and advancing awareness of illnesses that disproportionately affect historically marginalized groups can help clinicians more accurately diagnose and treat diverse populations.

CASE

- 14-year-old girl with history of hypoparathyroidism, hypothyroidism, primary adrenal insufficiency, positive diabetes antibodies, alopecia, subacute intermittent dysphagia.
- Presentation: New onset spells of paralysis, emotional lability, use of profanity, and homicidal threats.
- Consult Question: Could spells could be related to functional neurological symptom (conversion) disorder or another psychiatric condition?
- Clinical findings: Disorientation, agitation. Spells with cyanosis and O_2 desaturations. Bilateral ptosis, facial droop, and dysarthria. Symptoms did not resolve with administration of lorazepam.
- Diagnostic studies: Myelopathy autoimmune panel, positive for GAD65 Ab assay – indicating autoimmune encephalitis. AIRE gene variant, confirming diagnosis of APS, type 1.
- Results: Resolution of spells and autoimmune encephalitis with intravenous steroids and PLEX therapy.

AUTOIMMUNE POLYENDOCRINE SYNDROME-1 (APS-1)

FIGURE 1

Potential Neuropsychiatric Manifestations of APS-1 in a Pediatric Patient:

Delusions	Cognitive Difficulties
Hallucinations	Anxiety
Mood changes	Irritability
Sleep disturbances	Apathy

TABLE 1

Symptom	*Psychiatric Differential*	Autoimmune Polyendocrine Differential
Excoriation	*OCD-related Disorder (skin-picking)*	Eczema with delirious disinhibition
Spells	*Functional Spells (note: no desaturations or cyanosis)*	Autoimmune encephalitis with neurologic symptoms
Psychosis	*Primary psychotic disorder (schizophrenia)* *Mood disorder with psychosis*	Parathyroid pathology Pernicious anemia Autoimmune encephalitis
Insomnia	*Mania, sleep disorder*	Delirium
Disinhibition	*Primary psychotic disorder, mood disorder, substance use*	Delirium

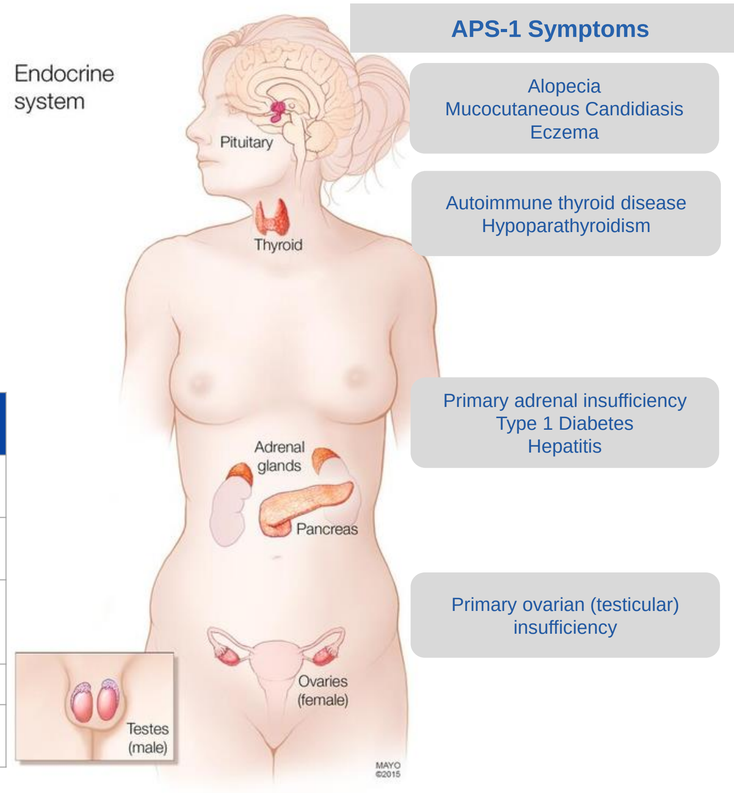
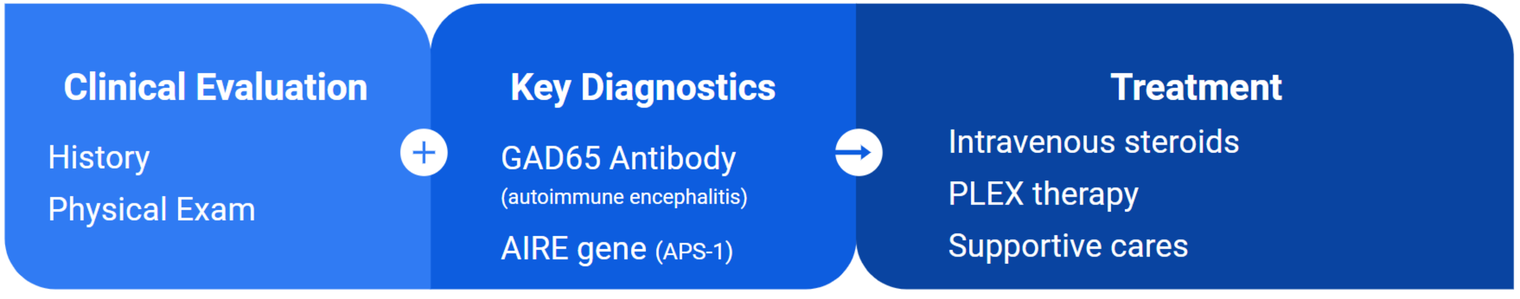


FIGURE 2



DISCUSSION

- This case initially presented with characteristics commonly associated with functional neurologic spells, including spontaneous full body paralysis with maintained awareness.
- Other symptoms were described in psychiatric terms of “OCD” behaviors, skin-picking, and anxiety.
- Psychiatric etiology less likely based on:
  - Cyanosis, oxygen desaturations, pharyngeal weakness are not features of functional disorders.
  - Phenomenology of the spells and behavioral changes were inconsistent with catatonia; IV Ativan did not help.
  - Skin-picking better explained by eczematic pruritis with delirious disinhibition
- Recommendations from consult-liaison psychiatry included delirium management and to continue search for neurological, rather than psychiatric, processes driving these spells.

CONCLUSION

The picture of abrupt onset of behavioral and cognitive changes, primarily around perseverative and disinhibited verbalizations and behavior in the context of a suspected underlying neurological process was most consistent with delirium being driven by the underlying autoimmune encephalitis. This was supported by laboratory results upon completion of thorough medical workup and effective treatment with intravenous steroids and PLEX therapy. While APS-1 typically initially manifests with endocrine, cutaneous and infectious symptoms, psychiatric presentations were observed in this case. Consult liaison psychiatrists must also be aware that patients with autoimmune conditions are prone to psychiatric symptoms.

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