HYPERCALCAEMIA AS AN INDICATION OF ADRENAL INSUFFICIENCY IN A PATIENT WITH AUTOIMMUNE POLYENDOCRINOPATHY-CANDIDIASIS-ECTODERMAL DYSTROPHY (APECED)

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Introduction

Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), is a rare inherited disease of childhood, caused by the mutation of the AIRE gene on chromosome 21. It is characterized by 3 main diseases: chronic mucocutaneous candidiasis (CMC), chronic hypoparathyroidism (HP) and Addison's disease (AD), and can be associated with other autoimmune diseases and/or manifestations of ectodermal dystrophy.

Case presentation

An 8 year old girl, who was known to have hypoparathyroidism and she was treated with calcium and calcitriol, presented at the endocrinology department, complaining of fatigue and abdominal pain for five days, without vomiting, nor fever.

Physical examination

On physical examination she was pale with poor skin turgor, low normal blood pressure and mildly tachycardic.

Laboratory findings

<u>The initial blood investigation revealed</u>:
1.hypercalcaemia 11.6 mg/dl
2.hyponatremia 125 m Eq/l
3.hyperkalemia 5.8 m Eq/l

which suggested the possibility of adrenal failure

Further investigation

Further laboratory investigation demonstrated:
1.elevated ACTH levels (3465 pg/ml)
2.decreased cortisol production (5.84 μg/dl)
3.anti-adrenal antibodies were present.
4.PRA: 148.2 ng/ml/h

Which confirmed the diagnosis of adrenal failure. The patient was treated with hydrocortisone and she was rapidly improved.

Follow up

The patient was followed regularly, treated with hydrocortisone, calcium and calcitriol, having normal electrolytes and low normal calcium. Eight months later she presented in our endocrinology department for her scheduled follow up and a candidiasis scalp lesion was detected.
Molecular genetic analysis is anticipated to confirm the diagnosis of APECED

AIRE gene mutations (a) and functional domains of corresponding proteins (b)

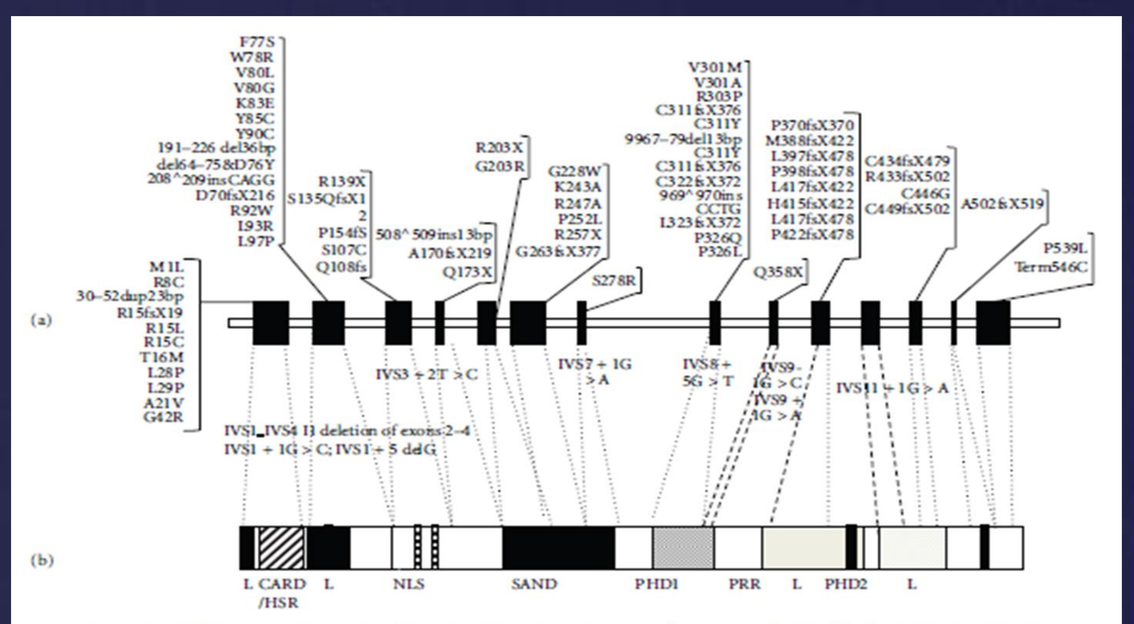

FIGURE 1: *AIRE* gene mutations (a) and functional domains of corresponding protein (b). Modified by Meloni et al [68].

Clinical features of AIRE deficiency/APECED

<u>The classic triad</u> is mucocutaneous candidiasis, hypoparathyroidism, and adrenal failure

<u>Mechanisms proposed to explain the causal relationship of adrenal insufficiency to hypercalcemia</u>.

1.The elevated calcium levels are due to an increase of calcium released from bone and is supported by the presence of marked hypercalciuria and urinary hydroxyproline excretion, unrelated to bone remodeling process.

2. The adrenal gland produces a paracrine hormone called stanniocalcin, which reduces circulating calcium. Thus, in a state of adrenal insufficiency, blood calcium may be elevated due to decreased stanniocalcin production

3. Endogenous glucocorticoids decrease intestinal calcium absorption opposing the effects of vitamin D and increase urinary calcium excretion

Conclusion

The presence of abdominal pain and hypercalcaemia in a patient with hypoparathyroidism should raise the suspicion of adrenal insufficiency which has to be investigated and treated on an emergency basis. Moreover, as the timing of the appearance of the individual disorders varies, a high level of suspicion regarding the development of associated endocrinopathies in particular adrenal failure, as well as informing parents of the possible symptoms is of outmost importance

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