Adrenal hypoplasia seemingly first as a primary hypoaldosteronism

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THE CASE

Medical history:

- Male full-term newborn from non-consanguineous parents, caucasian ethnicity
- SGA (small for gestational age) for weight (IUGR during the last month of pregnancy)
- Spontaneous delivery, no complications at birth
- Hospitalization at 11 days of life for ineffective breastfeading, discharged after registering weigh growth (blood and urine tests were normal)

Age	Na+ (n.r. 136-146 mEq/l)	K+ (n.r. 3,5-5,30 mEq/l)	Aldosteron (n.r. 50-300 pg/ml)	Renin (n.r. 4,4-46,1 µU/ml)	ACTH (n.r. 4,3-52 pg/ml)	Cortisol (n.r. 6,7-22,6 µU/dl)
18-days-old	**110** (natriuria 16 mEq/l)	**7.5**	**38.6**	**44100**	91.4	13.7
5-months-old	134	4.9	55.9	181.2	124.9	4.4
7-months-old	139	5.0	45.2	/	/	5.2
9-months-old	139	4.2	21.5	123.6	**300.6**	**7.4** (urinary cortisol 20ug/24h, n.r. 58-403 µg/24h)
12-months-old	138	4.4	<10	30.7	89.9	22.8

Fludrocortisone 100mcg/die + NaCl 3mEq/kgdie

Hydrocortisone 13.16mg/mq/die

DIFFERENTIAL DIAGNOSIS

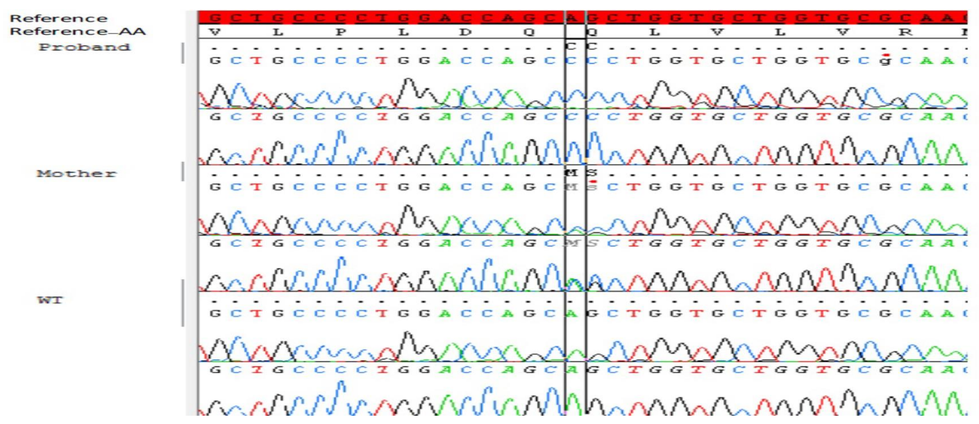
1)Primary Hypoaldosteronism? → genetic analysis of CYP11B2 gene (encoding aldosynthase) sequencing did not confirm the hypothesis

2)Congenital adrenal hyperplasia (CAH)? → normal 17-OH-progesterone levels

3) Autoimmune adrenal insufficiency? → Adrenal antibodies negative

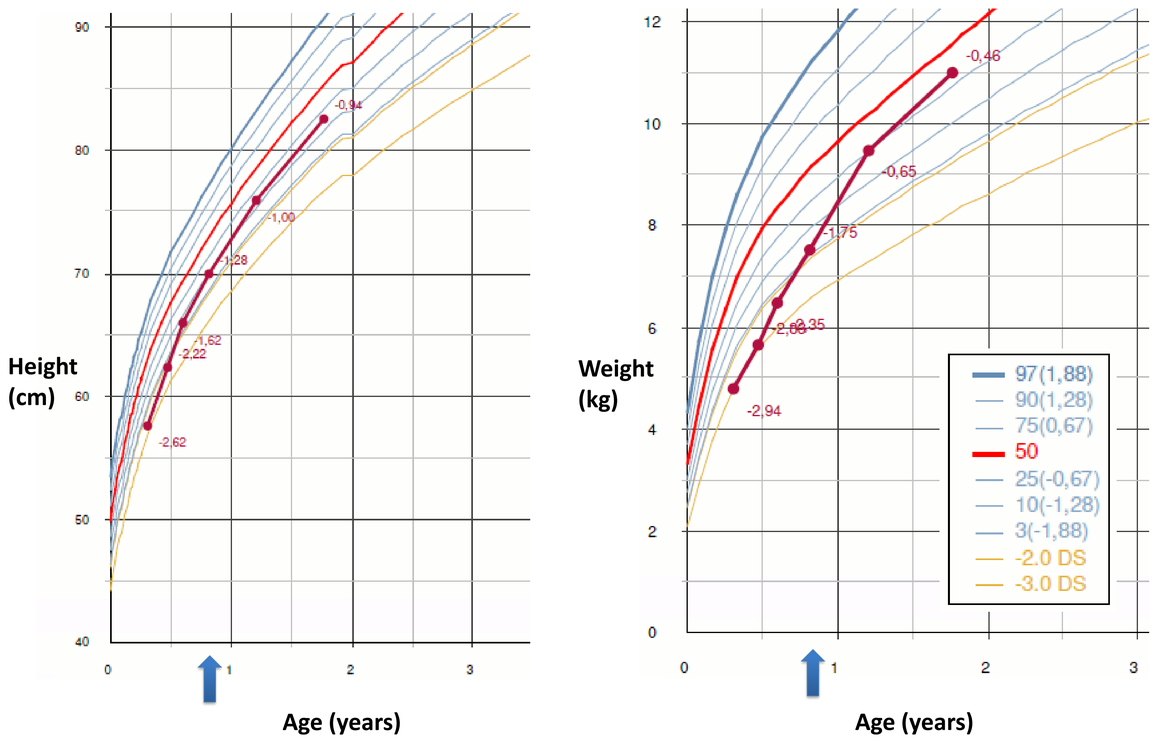
4)Adrenoleukodystrophy? → VLCFA negative

5)Adrenal hypoplasia congenita?



DNA analysis performed by Sanger sequencing identified a novel in frame indel mutation in DAX-1 gene 109 (c.848_849delinsCC or p.(Gln283Pro)), confirming the diagnosis of AHC. As expected the mutation was carried by the mother.

EFFECT OF THERAPY



(WHO, 2007)

TAKE HOME MESSAGES

- X-linked Adrenal Hypoplasia Congenita (AHC) is a **congenital disorder** characterized by **adrenal insufficiency** sometimes associated with **hypogonadotropic hypogonadism** (HHG).
- The estimated incidence is **1 in 12500 births**.
- Unspecific symptoms and silent family history can hide a life-treating condition
- Pay attention to presence of dehydration, hyponatremia and hyperkaliemia in a vomiting patient without other signs
- Isolated mineralocorticoid deficiency is a rare first manifestation of AHC
- Hyponatriemia is always present at the onset, K+ levels can be high or normal
- A tightened endocrinological follow-up allows to discover a glucocorticoid deficiency with subsequent starting of supplementation therapy with hydrocortisone before any adrenal crisis

References

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