CLINICAL AND HORMONAL EVOLUTION OF ALDOSTERONE SYNTHASE DEFICIENCY: IS COMPLETE REMISSION POSSIBLE?

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BACKGROUND
Aldosterone synthase deficiency (ASD);
- Autosomal recessive
- Bilaleic mutations of the CYP11B2 gene
- The patients are presented with symptoms of severe salt-wasting
- The need for treatment decreases with the increasing age
- Adult patients are usually asymptomatic without having mineralocorticoid therapy
- Data are scarce regarding clinical and biochemical outcomes

OBJECTIVE
Assessment of the growth and steroid profiles of patients at the time of diagnosis and after discontinuation of treatment.

DESIGN AND METHOD
- Children with clinical diagnosis of ASD
- Multicenter study
- Growth and treatment characteristics were recorded
- Plasma adrenal steroids were measured using LC/MS/MS
- Genetic diagnosis was confirmed by CYP11B2 gene sequencing and in silico analyses

RESULTS
- Sixteen patients from 12 families were included (8 females; median age at presentation: 3.1 months, range: 0.4-8.1)
- The most common symptom was poor weight gain (56.3%)
- Median age of onset of fludrocortisone treatment was 3.6 months (0.9-8.3)
- Catch-up growth was achieved at median 2 months (0.5-14.5) after treatment.

CONCLUSIONS
- Fludrocortisone treatment is associated with a rapid catch-up growth and control of electrolyte imbalances in ASD
- Decreased mineralocorticoid requirement over time can be explained by the development of physiological adaptation mechanisms rather than improved aldosterone synthase activity
- As complete biochemical remission cannot be achieved, a long-term surveillance of these patients is required