Neonatal failure to thrive - not always a congenital adrenal hyperplasia...

L. Kasongo, R. Nicolescu
CHR Citadelle, Université de Liège

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a frequent cause of neonatal failure to thrive, with specific hormonal profile and rapid response to steroids. The disappointing clinical course and biological response under therapy, especially when the complete adrenal hormonal tests are not available, requires to enlarge the differential diagnosis to other adrenal disorders. Pseudohypoaldosteronism (PHA) is a rare entity inducing the same clinical appearance as CAH, but its biological diagnosis is highly specific.

METHODS

We report a case of 4-week-old boy presented to a general hospital with:

- failure to thrive (actual weight < birth weight)
- clinical dehydration (but no vomiting or diarrhoea)
- dyselectrolytemia (hyponatremia, hyperkalemia)
- metabolic acidosis.

There is no medical history around pregnancy. The baby is the second child of non-consanguineous parents. A paternal cousin had transient problems with salt in the neonatal period.

DIAGNOSIS - ADRENAL CRISIS

Treatment - Hydrocortisone iv

24 hours later: persisting dyselectrolytemia (hyponatremia and hyperkalemia).

RESULTS (1)

Extensive hormonal profile

- Cortisol ↑↑↑ under corticotherapy
- ACTH ↓↓↓
- Renin ↑↑
- Aldosterone ↑↑↑
- 17-OH-progesterone NORMAL!

Urinary analysis: sodium ↑↑↑ and potassium ↓↓↓

Sweat test: normal

RESULTS (2)

Clinical evolution, response to hydrocortisone and biological results were suggestive of PSEUDOHYPOADLOSTÉRONISME (genetic resistance to mineralocorticoid).

Hydrocortisone was stopped and normal salt was added into the milk.

The clinical and biological course was satisfactory (weigh gain of 50 g/d, stabilized sodium and normal potassium levels).

Considering the clinical spectrum, family history, favorable response to sodium supplementation and kidney restricted aldosterone resistance (no sodium loss through sweat) we assume that this PHA is a renal type 1.

A genetic analysis is under way.

CONCLUSIONS

Our goal describing this case is to underline the necessity to have in mind a differential diagnosis for clinical and biological appearance of congenital adrenal hyperplasia, notably early in the presentation, when the hormonal profile is not completely available.

PHA is a rare disease defined by resistance to mineralocorticoid that presents in the neonatal period with failure to thrive, salt-wasting nephropathy, hyperkalemia and metabolic acidosis.

Two forms are described: autosomal dominant renal form and autosomal recessive systemic form.