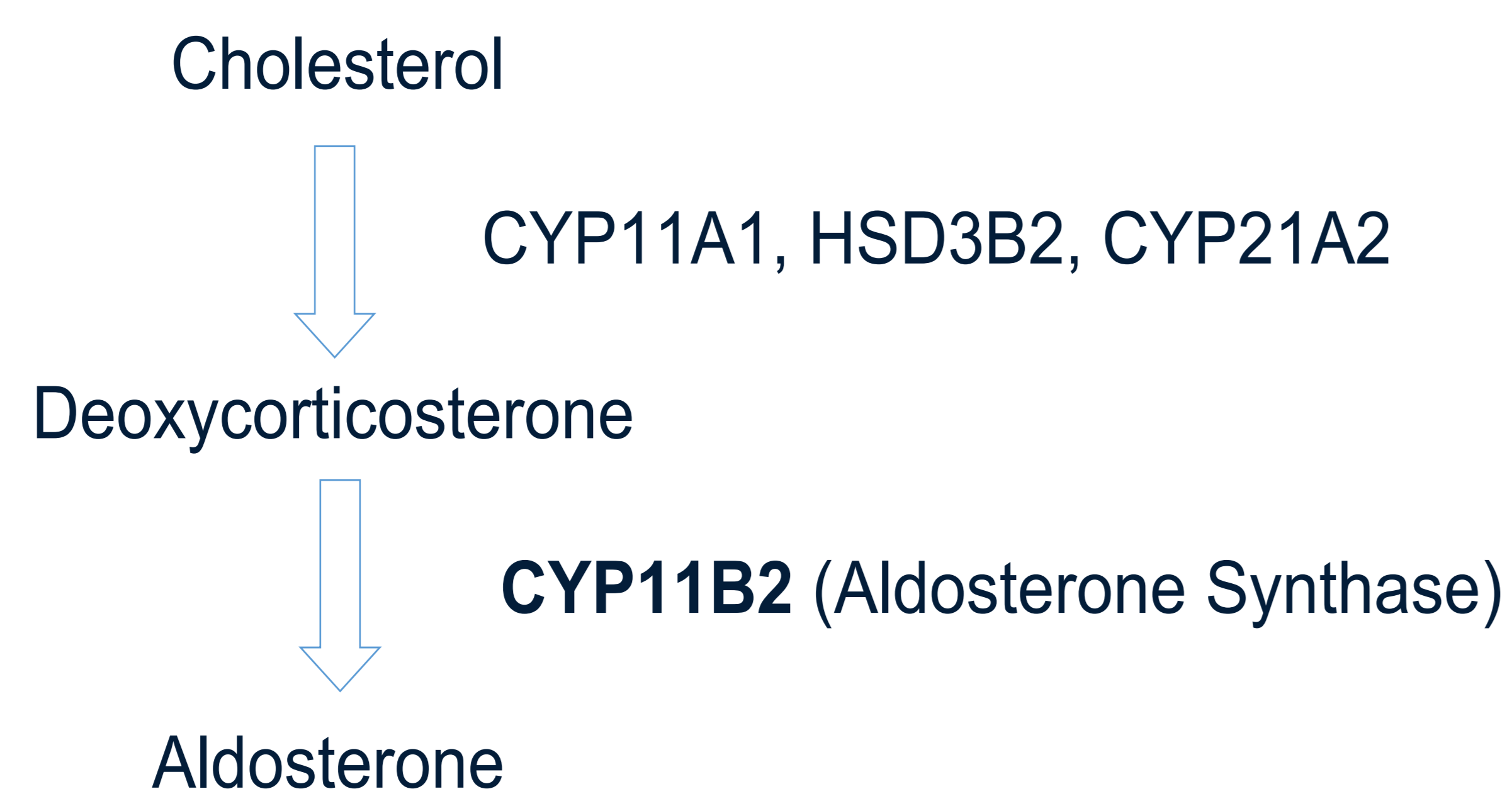


### Introduction

Failure to thrive (FTT) is a common entity encountered by pediatricians. It can be caused by inadequate energy intake or "organic causes" such as inborn errors of metabolism or disorders of the endocrine system. One cause that is rarely seen is a deficiency of aldosterone.



### Case presentation

- A set of 3-month-old monozygotic male twins presented with failure to thrive and dehydration.
- Pregnancy and delivery were uneventful; maternal serologies were negative. No family history of endocrine abnormalities.
- Throughout their first months of life, the twins' weight z-scores remained more than two standard deviations below the mean.
- Initially, they were evaluated for gastrointestinal anatomical causes that were ruled out.
- They were also found to have persistent hyponatremia and hyperkalemia despite rehydration, requiring admission to the pediatric intensive care unit.
- Endocrinology was consulted. Cortisol, corticotropin and 17-hydroxyprogesterone levels were normal.
- Aldosterone levels were **undetectable** in both twins, and plasma renin was normal.
- Gene sequencing was sent for both patients of CYP11B2, which codes for the enzyme aldosterone synthase (AS), and they were started on fludrocortisone 0.1 mg twice daily with sodium chloride supplementation, which resulted in rapid improvement of electrolyte imbalances and growth.
- They both had two different pathogenic variants of CYP11B2, confirming the diagnosis of AS deficiency.

Twin A	
Na	131 (135-142) mmol/L
K	6.5 (4.1-5.3) mmol/L
Cl	101 (100-107)mmol/L
HCO3-	15 (10-24) mmol/L
ACTH	25 (5-46) pg/ml
Cortisol	12.1 (3.7-19.4) mcg/dl
17-hydroxyprogesterone	337 (<110) ng/dl
Aldosterone	< 1 ng/dl
Renin	26 ng/ml/h
UNa	33

Twin B	
Na	128 (135-142) mmol/L
K	6.3 (4.1-5.3) mmol/L
Cl	99 (100-107) mmol/L
HCO3-	12 (10-24) mmol/L
ACTH	16 (5-46) pg/ml
Cortisol	12.4 (3.7-19.4) mcg/dl
17-hydroxyprogesterone	191 (<110) ng/dl
Aldosterone	<1 ng/dl
Renin	14 ng/ml/h
UNa	27 mmol/L

### Discussion

- Hyponatremia and hyperkalemia in the setting of failure to thrive should elicit consideration of primary adrenal insufficiency, which in infancy is most commonly due to congenital adrenal hyperplasia (CAH).
- Metabolic disorders were also on the differential however patients remained euglycemic throughout hospital course and urine organic acids, blood organic acids, and ammonia were unremarkable.
- Aldosterone deficiency and insensitivity may mimic the symptoms of CAH and should be considered as well.
- AS is only expressed in the zona glomerulosa of the adrenal cortex and is regulated by the renin-angiotensin system. It catalyzes the final three reactions of mineralocorticoid synthesis, which ultimately converts deoxycorticosterone to aldosterone.
- AS deficiency is inherited in an autosomal recessive manner. The most common defect is in the activity of the terminal enzyme in the aldosterone pathway: Aldosterone Synthase type I or II. This enzyme deficiency can be caused by a deletion or point mutation in chromosome 8.

### Conclusion

We present a unique case of monozygous twins with hypoaldosteronism due to AS deficiency that presented with FTT and dehydration. While the differential of FTT in infants can be broad, the importance of identifying and treating rare causes of FTT can ultimately be lifesaving. We aim to highlight that although aldosterone deficiency is rare, it can mimic the electrolyte abnormalities of CAH.

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