

# Delayed diagnosis of salt wasting congenital adrenal hyperplasia, without complications of cortisol deficiency: a case report

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## Conclusion

We describe a salt wasting congenital adrenal hyperplasia (SW-CAH) patient who was not treated with glucocorticoids and mineralocorticoids during the first two years of life due to an initial misdiagnosis of secondary pseudohypoaldosteronism. Despite his severe enzymatic defect he had no clinically significant signs of cortisol deficiency despite undergoing surgery and suffering several illnesses. Based on the clinical observations in this patient and on *in vitro* studies showing that steroid precursors such as 17-hydroxyprogesterone and 21-deoxycortisol have an agonistic effect on the human glucocorticoid receptor (fig 1), we hypothesize that in untreated CAH patients elevated levels of steroid precursors may partially compensate cortisol deficiency.

## Background

Salt wasting congenital adrenal hyperplasia (SW-CAH) patients suffer from a deficiency of both cortisol and aldosterone and develop life-threatening salt wasting crises neonatally. Treatment consists of glucocorticoids, mineralocorticoids and salt supplementation. We present a case with a two years delayed diagnosis of SW-CAH.

## Case presentation

The patient was admitted to the hospital at the age of two weeks because of poor feeding, irritability and weight loss. Laboratory analysis showed a hyperkalemia (7.5 mmol/l; normal values 3.5-4.7 mmol/l) and hyponatremia (132 mmol/l; normal values 135-145 mmol/l) with inadequately high urinary sodium excretion (46 mmol/l). Based on an abdominal ultrasound and a voiding cystourethrogram the diagnosis of posterior urethral valves was made. Consequently, the electrolyte abnormalities were attributed to secondary pseudohypoaldosteronism (PHA). The posterior urethral valves were surgically resected and the hyponatremia was treated with sodiumchloride. No glucocorticoids were prescribed.

At the age of two years he still required high doses of oral sodiumchloride to prevent hyponatremia. Additional laboratory studies showed an increased 17-hydroxyprogesterone (309.80 nmol/l, normal values 0.2-7.4 nmol/l) and androstenedione (4.7 nmol/l, normal values 0.03-1.05 nmol/l). The diagnosis SW-CAH was made and confirmed with mutation analysis (homozygous R356W mutation, no residual enzyme activity). After initiation of treatment with hydrocortisone and fludrocortisone, he no longer needed sodiumchloride supplementation.

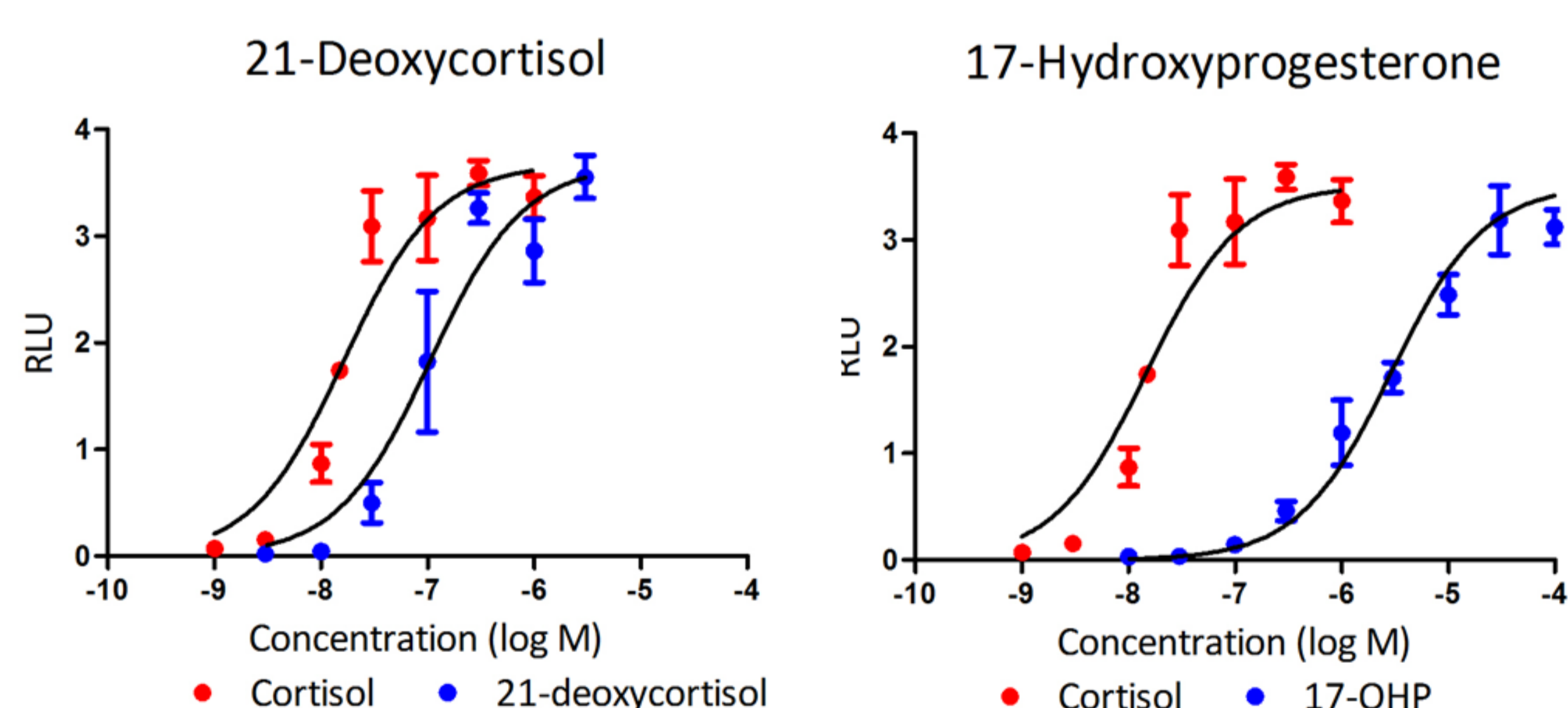


Figure 1. Transactivation of the hGR in COS-7 cells by various steroids, in comparison with the transactivation of the hGR by cortisol. Transactivation is measured using a dual luciferase assay after co-transfection of the cells with the hGR and the luciferase reporter vectors MMTV and pRL-TK.

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