Two cases of apparent mineralocorticoid excess due to novel mutations in *HSD11B2* gene

Nina Makretskaya[1], Irina Kostrova[2], Anatoly Tiulpakov[1]
[1]Endocrinology Research Centre, Moscow, Russian Federation
[2]Children's Republic hospital, Makhachkala, Russian Federation

Background. Human HSD11B2 metabolizes active cortisol into cortisone and protects the mineralocorticoid receptor from glucocorticoid occupancy. Loss of function mutations in *HSD11B2* gene cause a rare autosomal recessive disorder, apparent mineralocorticoid excess, resulting in low-renin hypertension and hypokalemia.

Objective. We present 2 children with apparent mineralocorticoid excess.

Case 1: a boy presenting at 11 years with growth retardation (SD, -2.8), polyuria, polydipsia, hypertension (160/110-170/140 mm Hg).

Biochemistry results	Basal	Rx, spironolactone (50 mg per day)
Potassium, mmol/l	2.2-2.7	4.0-4.2
Sodium, mmol/l	140-142	140-143
Plasma renin activity, ng/ml•h	0.14	5.5
Serum aldosterone, pmol/l	<30.0	63.1

Case 2: a girl presenting at the age of 6 years with polyuria, high blood pressure (120/85-130/90 mm Hg) and hypokalemia (2.4 mmol/l).

Biochemistry results	Basal	Rx, spironolactone (50 mg per day)
Potassium, mmol/l	1.5-2.4	3.5-3.7
Sodium, mmol/l	142-144	140-142
Plasma renin activity, ng/ml•h	<0.1	4.3
Serum aldosterone, pmol/l	32.3	54.1

Therapy with spironolactone (50 mg per day) was started. At present the children show normal electrolytes and PRA, and blood pressure 100/70-110/80 mm Hg.

Conclusion. In the present study we described clinical and molecular genetic characterization of two patients with novel mutations in *HSD11B2* gene.

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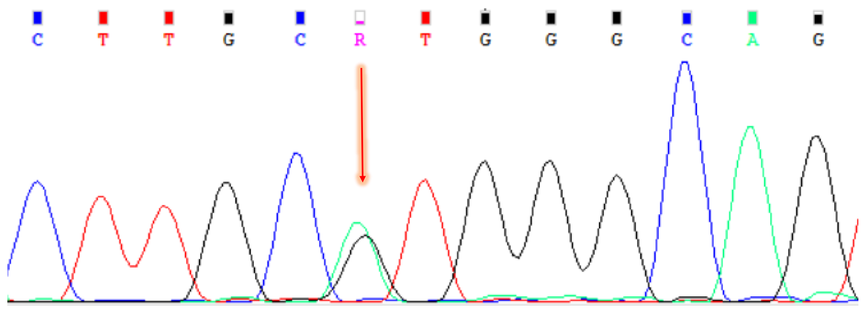
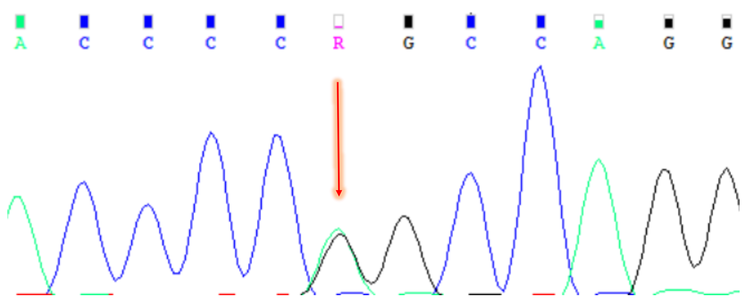
Methods. *HSD11B2* gene was analyzed by Sanger sequencing.

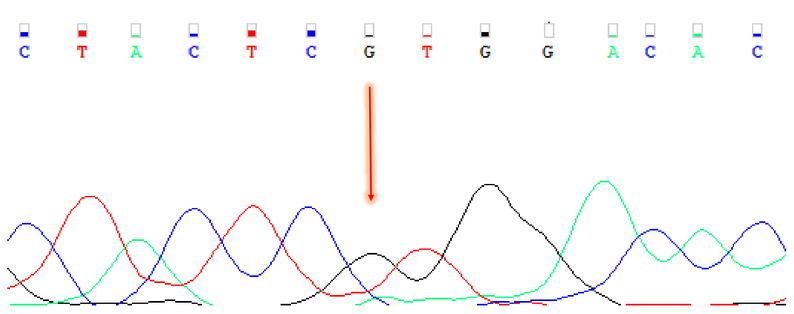
Results. Compound heterozygous p.G341S/p.H304R and a homozygous p.M243V mutations were found in Case 1 and Case 2, respectively. All mutations were novel.



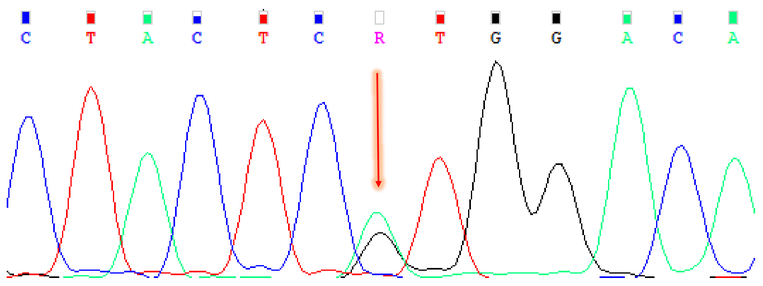
c.911A>G p.H304R (case 1)



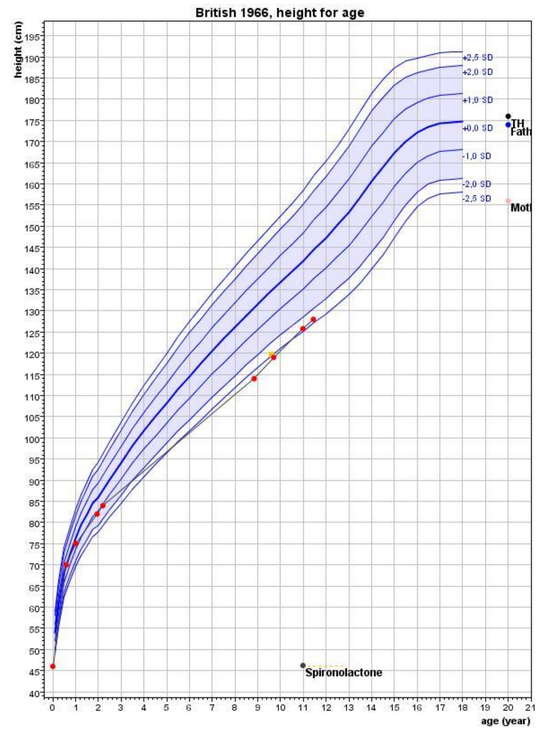
c.1021G>A p.G341S (case 1)



c.727A>G p.M234V (case 2) - homozygous



c.727A>G p.M234V (patient's parent) - heterozygous



Case #1 Growth Chart