Two cases of apparent mineralocorticoid excess due to novel mutations in \textit{HSD11B2} gene

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

\textbf{Objective.} We present 2 children with apparent mineralocorticoid excess.

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

\begin{tabular}{|c|c|c|}
\hline
\textbf{Biochemistry results} & \textbf{Basal} & \textbf{Rx, spironolactone (50 mg per day)} \\
\hline
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 \\
\hline
\end{tabular}

\textbf{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).

\begin{tabular}{|c|c|c|}
\hline
\textbf{Biochemistry results} & \textbf{Basal} & \textbf{Rx, spironolactone (50 mg per day)} \\
\hline
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 \\
\hline
\end{tabular}

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.

\textbf{Conclusion.} In the present study we described clinical and molecular genetic characterization of two patients with novel mutations in \textit{HSD11B2} gene.

\textbf{Methods.} \textit{HSD11B2} gene was analyzed by Sanger sequencing.

\textbf{Results.} Compound heterozygous p.G341S/p.H304R and a homozygous p.M234V mutations were found in Case 1 and Case 2, respectively. All mutations were novel.

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