Liddle Syndrome

Template:Infobox - genetická choroba

It is a genetic kidney disease with autosomal dominant inheritance, which usually manifests itself in adolescence or adulthood. The symptoms are the same as in primary hyperaldosteronism, but the aldosterone level is not elevated. That is why we are talking about **pseudohyperaldosteronism**.

Pathogenesis

The cause is a mutation in the gene for the subunits of **sodium channels** on the luminal side of the cells of the renal collecting duct (so-called ENaC channel). There are more channels in the renal tubules and they are hyperactive due to the mutation. Thus, more Na + is absorbed from the urine as in the case of Aldosterone.

Clinical Picture

Manifestations of the disease include hypertension, hypokalemia, metabolic alkalosis, hypernatremia, polyuria, thirst, failure to thrive. From laboratory tests we find reduced or normal levels of aldosterone and renin.

Treatment

We supply the patient with potassium in the form of potassium salts. The main drug is **triamterene**, a potassium-sparing diuretic, that inhibits Na^+ channel activity or amiloride. Spironolactone has no effect here. Salt restriction is recommended.

References

Related articles

- Aldosterone
- Primary hyperaldosteronism
- Hypokalemia

Source

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