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Bartter syndrome

Bartter syndrome is a group of rare conditions that affect the kidneys.

Causes

There are five gene defects known to be associated with Bartter syndrome. The condition is present at birth (congenital).

The condition is caused by a defect in the kidneys' ability to reabsorb sodium. People affected by Bartter syndrome lose too much sodium through the urine. This causes a rise in the level of the hormone aldosterone, and makes the kidneys remove too much potassium from the body. This is known as potassium wasting.

The condition also results in an abnormal acid balance in the blood called hypokalemic alkalosis, which causes too much calcium in the urine.

Symptoms

This disease usually occurs in childhood. Symptoms include:

- Constipation
- Less weight gain than other children of similar age and sex (growth failure)
- Frequent urination (urinary frequency)
- Low blood pressure
- Kidney stones
- Muscle cramping and weakness

Exams and Tests

Bartter syndrome is usually suspected when a blood test finds a low level of potassium in the blood. Unlike other forms of kidney disease, this condition does not cause high blood pressure. There is a tendency toward low blood pressure. Laboratory tests may show:

- High urine potassium, calcium, and chloride levels
- High blood levels of the hormones, renin and aldosterone
- Low blood chloride level

- Metabolic alkalosis

These same signs and symptoms can also occur in people who take too many diuretics (water pills) or laxatives. Urine tests can be done to rule out other causes.

An ultrasound of the kidneys may be done.

Treatment

Bartter syndrome is treated by eating foods rich in potassium or taking potassium supplements.

Many people also need salt and magnesium supplements. Medicine may be needed that blocks the kidney's ability to get rid of potassium. High doses of nonsteroidal anti-inflammatory drugs (NSAIDs) may also be used.

Outlook (Prognosis)

Infants who have severe growth failure may grow normally with treatment. Over time, some people with the condition will develop kidney failure.

When to Contact a Medical Professional

Contact your health care provider if your child is:

- Having muscle cramps
- Not growing well
- Urinating frequently

Alternative Names

Potassium wasting; Salt-wasting nephropathy

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Review Date 12/31/2023

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Learn how to cite this page



Health Content
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06/01/2028

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