CONDITION

Bartter syndrome

Learn how Bartter syndrome affects kidney salt balance, including its symptoms, genetic causes, diagnosis, treatment, complications, and follow-up care.

WHAT IT IS
Group of rare inherited kidney salt-wasting conditions

BODY SYSTEM
Kidneys and urinary system

MAIN SYMPTOMS
Frequent urination, strong thirst, dehydration, muscle weakness

WHAT CAUSES IT
Inherited genetic changes

HOW IT IS DIAGNOSED
Blood and urine tests, genetic testing

MAIN TREATMENT
Salt and electrolyte replacement

CAN IT BE CURED?
No, inherited transport problem persists

IS IT AN EMERGENCY?
Sometimes, severe dehydration or irregular heartbeat

LONG-TERM OUTLOOK
Lifelong management with possible chronic kidney disease

Overview

Bartter syndrome is a group of rare inherited conditions that affect how the kidneys handle salt. Normally, the kidneys return much of the filtered sodium and chloride to the bloodstream. In Bartter syndrome, part of this process does not work properly, so too much salt passes into the urine.

Losing salt causes the kidneys to release more water. It also activates hormones that try to preserve blood volume. These changes can lead to low potassium, changes in the body's acid-base balance, increased thirst, and frequent urination. Blood pressure is often normal or low even though the hormones renin and aldosterone are elevated.

Some forms begin before birth and cause excess amniotic fluid around the fetus, known as polyhydramnios. Other forms first become noticeable during infancy, childhood, or, less often,

adulthood. The severity varies widely, including among people with changes in the same gene.

Bartter syndrome matters because ongoing losses of salt, water, and potassium can affect hydration, growth, muscles, heart rhythm, and kidney health. Treatment does not repair the inherited kidney transport problem, but it can replace losses, improve symptoms, support growth, and reduce complications.

Symptoms

Symptoms depend on the specific form of Bartter syndrome, the person's age, and how much salt and water the kidneys lose.

Before birth, severe forms may cause polyhydramnios because the fetus produces unusually large amounts of urine. This can increase the chance of premature birth. After birth, an affected infant may have high urine output, dehydration, difficulty gaining weight, fever without infection, vomiting, or marked thirst.

Children with a later-onset form may have poor growth, frequent urination, bed-wetting, strong thirst, salt cravings, constipation, muscle weakness, or painful muscle cramps. Some people experience tiredness, headaches, or tingling sensations when potassium or other electrolytes are low. Electrolytes are minerals, such as potassium and chloride, that help regulate nerves, muscles, and fluid balance.

Kidney calcium deposits, called nephrocalcinosis, may occur without obvious symptoms. Some people develop kidney stones, which can cause pain or blood in the urine.

Certain genetic forms also cause sensorineural hearing loss. This means the inner ear or the nerve pathways involved in hearing do not function normally. Hearing loss may be present from birth.

Symptoms can overlap with many more common conditions. Thirst, muscle cramps, or a low potassium result alone does not establish a diagnosis of Bartter syndrome.

Causes and risk factors

Bartter syndrome is caused by a disease-related change in a gene needed for salt transport in the kidney. The affected transport proteins are mainly located in the thick ascending limb of the loop of Henle, a section of each kidney's filtering tubes.

Several genes can be involved. Examples include `SLC12A1`, `KCNJ1`, `CLCNKB`, `BSND`, and combined changes affecting `CLCNKA` and `CLCNKB`. Changes in `MAGED2` can cause a

temporary antenatal form that usually improves during infancy. Genetic categories are sometimes described as Bartter syndrome types 1 through 5, but naming systems differ and continue to evolve.

The genetic form influences the expected pattern:

Most forms follow an autosomal recessive inheritance pattern. This means a child inherits one altered copy of the gene from each biological parent. The parents usually carry the genetic change without having Bartter syndrome themselves. When both parents carry changes in the same gene, each pregnancy has its own chance of being affected.

The `MAGED2`-related form follows an X-linked pattern because the gene is on the X chromosome. A genetics professional can explain inheritance for a specific test result.

The main risk factor is having an inherited disease-causing gene change. A family history increases suspicion, but many affected people have no previously diagnosed relative. Bartter-like laboratory findings can also result from other conditions or medicines; these are not inherited Bartter syndrome.

- Some forms cause severe salt and water loss before or soon after birth.
- `CLCNKB`-related disease often appears during childhood but can range from mild to severe.
- `BSND` or combined `CLCNKA` and `CLCNKB` changes can cause both kidney symptoms and hearing loss.
- Urine calcium may be high in some forms and normal or lower in others.

Diagnosis

Diagnosis begins with a medical history, physical examination, and blood and urine testing. The clinician may ask about growth, thirst, urine output, vomiting, medicine use, pregnancy complications, hearing, and relatives with similar problems.

Typical blood findings can include low potassium and chloride. The blood may also be more alkaline than normal, a change called metabolic alkalosis. Renin and aldosterone are often elevated because the body is responding to salt and fluid loss. Magnesium may be low in some people, while kidney function can be normal or reduced depending on the clinical situation.

Urine testing helps show whether the kidneys are losing chloride, potassium, calcium, or other minerals. A high urine chloride level during metabolic alkalosis can support kidney salt wasting, although results must be interpreted in context. Urine calcium varies among genetic forms and can help guide the evaluation.

Additional assessment may include:

Genetic testing can confirm many cases, but a negative result does not always exclude the condition. Test panels may not detect every type of genetic change, and new causes may still be identified.

Clinicians also rule out more common explanations for the laboratory pattern. These include ongoing vomiting, diuretic medicines, laxative misuse, and some forms of cystic fibrosis that cause heavy salt loss through sweat. Gitelman syndrome is another inherited kidney salt-wasting condition. It usually affects a different part of the kidney tube, but its findings can overlap with Bartter syndrome.

In a pregnancy with polyhydramnios, Bartter syndrome is only one of several possible causes. Evaluation should be coordinated by maternal-fetal medicine, newborn, kidney, and genetics specialists when appropriate.

- Kidney ultrasound to look for nephrocalcinosis or stones.
- Hearing tests when a form associated with hearing loss is possible.
- Growth measurements and nutrition assessment in children.
- Genetic testing to identify the cause and help clarify inheritance.

Treatment and management

Treatment is tailored to the form of Bartter syndrome, the person's age, symptoms, laboratory results, and kidney function. A pediatric or adult kidney specialist often coordinates care.

Replacing salt and potassium is a central part of management. Infants and children may need extra sodium chloride because of ongoing urinary salt loss. Potassium is commonly replaced with a chloride-containing preparation. The amount must be individualized because needs can change with growth, diet, illness, kidney function, and other medicines. Supplements should not be started or adjusted without clinical guidance.

Some people also need magnesium replacement. Nutrition support may be useful when high fluid needs, poor appetite, or premature birth makes growth difficult. Children should have growth tracked over time rather than relying on a single measurement.

Bartter syndrome is associated with increased production of prostaglandins, substances that can worsen salt and water loss. A clinician may consider a nonsteroidal anti-inflammatory drug like ibuprofen or naproxen , or NSAID, to reduce prostaglandin production. Indomethacin, ibuprofen, and other NSAIDs have been used, but the choice depends on age and clinical circumstances.

NSAIDs require careful supervision. They can irritate the stomach, cause bleeding, affect kidney function, raise blood pressure, and contribute to cardiovascular problems in some people. The potential benefit must be balanced against these risks, particularly during dehydration or long-term treatment. NSAID use during pregnancy requires separate specialist assessment because it can affect the fetus and pregnancy.

Medicines that reduce aldosterone activity, conserve potassium, or block the renin-angiotensin system are occasionally considered in selected cases. However, they can worsen low blood pressure, salt loss, dehydration, or kidney function. They are not appropriate for everyone and require close monitoring.

People with hearing loss may benefit from hearing aids, cochlear implants, school support, or communication services. Treatment should involve an audiology and ear, nose, and throat team.

During vomiting, diarrhea, fever, surgery, or poor fluid intake, usual losses can become harder to replace. A clinician may provide an illness plan covering fluid intake, supplements, laboratory testing, and when to seek care. Severe dehydration or electrolyte imbalance may require intravenous fluids and hospital monitoring.

Complications

Poorly controlled salt and water loss can cause repeated dehydration and interfere with weight gain and childhood growth. Persistent low potassium may lead to weakness, cramps, constipation, or, when severe, an abnormal heart rhythm.

High calcium levels in the urine can contribute to nephrocalcinosis and kidney stones. Over time, some people develop chronic kidney disease. The risk may reflect several factors, including repeated dehydration, calcium deposits, the underlying genetic condition, and the effects of long-term treatment.

Complications can also include:

Not everyone develops these complications. Regular monitoring is intended to identify changes early and adjust treatment safely.

- Premature birth related to severe polyhydramnios.
- Hearing loss in specific genetic forms.
- Delayed development associated with prematurity, severe early illness, or hearing impairment.
- Stomach, kidney, bleeding, or cardiovascular problems related to NSAID treatment.

Follow up

Bartter syndrome usually requires long-term follow-up. The schedule depends on age, disease severity, treatment, and whether findings are stable.

Routine visits may include measurement of blood pressure, weight, height in children, hydration status, and symptoms. Blood tests commonly monitor potassium, sodium, chloride, bicarbonate, magnesium, and kidney function. Urine tests and kidney imaging may be repeated when there is concern about excess calcium, nephrocalcinosis, or stones.

People taking an NSAID need monitoring for kidney effects, stomach symptoms, bleeding, and blood pressure changes. Hearing should be assessed when the genetic form is associated with hearing loss or when there are concerns about speech or response to sound.

A genetics consultation can help families understand test results, recurrence chances, and options for testing relatives. Before pregnancy, people with Bartter syndrome or a known family variant may wish to discuss reproductive and prenatal testing options.

Contact the treating clinician promptly for a clear increase in thirst or urine output, worsening cramps or weakness, poor growth, repeated vomiting or diarrhea, difficulty taking supplements, new hearing concerns, or possible medicine side effects. These concerns usually need timely assessment but are not always emergencies.

Seek urgent medical care if the person cannot keep down fluids or supplements, appears significantly dehydrated, or has much less urine than usual. Emergency assessment is appropriate for fainting, severe weakness or paralysis, confusion, a seizure, chest discomfort, or a fast or irregular heartbeat, especially when an electrolyte problem may be present.

Sources

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