CONDITION

Fanconi syndrome

Fanconi syndrome is a kidney-tubule condition that causes important nutrients and minerals to be lost in urine.

WHAT IT IS
Faulty reabsorption in the kidney's filtering tubes

BODY SYSTEM
Kidneys and urinary system

MAIN SYMPTOMS
Excess urination, thirst, weakness, bone pain

WHAT CAUSES IT
Inherited disorders, medicines, toxins, or abnormal blood proteins

HOW IT IS DIAGNOSED
Blood and urine tests showing characteristic losses

MAIN TREATMENT
Treating the cause and replacing lost substances

CAN IT BE CURED?
Sometimes - reversible causes can resolve

IS IT AN EMERGENCY?
Sometimes - severe dehydration, confusion, fainting, or abnormal heartbeat

LONG-TERM OUTLOOK
Often manageable, but kidney and bone damage can progress

Overview

Fanconi syndrome is a problem with the proximal tubules, which are tiny tubes inside the kidneys. These tubes normally return useful substances from filtered fluid to the blood. In Fanconi syndrome, they do not reabsorb these substances properly, so too much is lost in urine.

The substances lost may include glucose, phosphate, bicarbonate, potassium, uric acid, amino acids, and small proteins. Bicarbonate helps control the body's acid balance, while phosphate and other minerals support healthy bones and muscles.

Fanconi syndrome is not the same as Fanconi anemia, a separate inherited condition affecting bone marrow, physical development, and cancer risk.

Fanconi syndrome can begin in childhood or adulthood. In children, it is often related to an inherited condition. In adults, it may be caused by a medicine, toxin, or disorder involving abnormal blood proteins.

Ongoing losses can cause dehydration, abnormal blood mineral levels, weak bones, poor growth in children, and sometimes chronic kidney disease. Early recognition and treatment may limit these problems.

Symptoms

Symptoms depend on which substances are lost, how severe the losses are, and how long they continue. Some people have few symptoms at first, and the condition is found through blood or urine tests.

Possible symptoms include:

Children may develop rickets, in which growing bones become soft and may bend or develop an unusual shape. Adults can develop osteomalacia, meaning mature bones become poorly mineralised and painful.

Low potassium can cause muscle weakness and, when severe, abnormal heart rhythms. Loss of bicarbonate can cause metabolic acidosis, meaning the blood becomes too acidic. This may lead to tiredness, faster breathing, poor growth, or bone problems.

Glucose may appear in urine even when blood glucose is normal. This differs from diabetes, where urine glucose is usually caused by high blood glucose.

- Passing large amounts of urine and feeling unusually thirsty
- Tiredness, muscle weakness, cramps, or reduced stamina
- Bone pain, difficulty walking, or fractures after minor injuries
- Poor growth or delayed development in children
- Reduced appetite, nausea, or weight loss
- Signs of dehydration, such as a dry mouth or dizziness

Causes and risk factors

Fanconi syndrome is a pattern of kidney-tubule dysfunction rather than one disease. It may be inherited or acquired later in life.

Inherited causes include cystinosis, Wilson disease, Dent disease, Lowe syndrome, hereditary

fructose intolerance, galactosemia, and some mitochondrial disorders. Cystinosis is an important cause in children. In this condition, cystine crystals build up inside cells and damage organs, including the kidneys.

Acquired Fanconi syndrome can be associated with:

Medicine-related risk depends on the drug, length of exposure, kidney health, and other treatments. Most people taking a medicine associated with Fanconi syndrome do not develop the condition.

Sometimes no clear cause is found.

- Certain medicines, including some antiviral like acyclovir or oseltamivir , chemotherapy, and antiseizure medicines
- Heavy-metal exposure, such as lead or cadmium
- Multiple myeloma and related conditions that produce abnormal antibody fragments called light chains
- Severe vitamin D deficiency in some settings
- Kidney injury following transplantation or other major illness

Diagnosis

Diagnosis usually requires blood and urine tests. No single symptom confirms Fanconi syndrome.

Blood tests may show low phosphate, potassium, bicarbonate, or uric acid. Kidney function is checked using creatinine and an estimated filtration rate. Blood glucose helps determine whether glucose in urine is due to kidney-tubule loss or diabetes.

Urine testing may find glucose despite normal blood glucose, excess phosphate or uric acid, amino acids, bicarbonate, and small proteins. Detailed tests can compare blood and urine levels to show that the kidneys are losing substances they would normally retain.

Not everyone has every abnormality. Results may change with hydration, treatment, and the stage of the condition.

Further testing looks for the cause. This may include reviewing medicines and toxin exposure, testing for abnormal blood proteins, or arranging genetic tests. Children with possible cystinosis may have specialised tests for cystine in white blood cells and an eye examination for cystine crystals.

Bone X-rays or a bone-density scan may be used for bone pain, poor growth, deformity, or

concern about fractures.

Treatment and management

Treatment has two main goals: addressing the cause when possible and replacing substances lost through the kidneys.

Replacement treatment may include fluids, phosphate, potassium, and alkali such as bicarbonate or citrate. Alkali reduces excess acidity in the blood. Vitamin D may be needed when bone mineralisation is affected. Treatment is guided by repeated blood and urine results because too little or too much replacement can cause problems.

If a medicine is suspected, the prescriber may adjust, replace, or stop it after considering why it is needed and what alternatives are available. Do not stop a prescribed medicine on your own, especially antiviral, chemotherapy, or antiseizure treatment.

Treating an underlying condition is also important. For example, cysteamine is used in cystinosis to reduce cystine buildup and slow organ damage. Disorders involving abnormal light chains may need specialist blood-cancer treatment. Removing an ongoing source of heavy-metal exposure may prevent further injury.

Children may need support from kidney, hormone, nutrition, bone, and growth specialists. Adults with bone disease or abnormal blood proteins may also need care from several specialties.

Follow-up commonly includes blood pressure checks, growth measurements in children, blood and urine tests, kidney-function monitoring, and bone-health assessment. Treatment often needs adjustment over time.

Complications

Untreated or poorly controlled Fanconi syndrome can affect several parts of the body.

Loss of phosphate and changes in vitamin D activity can cause rickets in children or osteomalacia in adults. Possible effects include bone pain, bowed legs, reduced height, difficulty walking, and fractures.

Ongoing bicarbonate loss can lead to chronic metabolic acidosis, worsening muscle loss, poor growth, and bone disease. Potassium loss may cause weakness or abnormal heart rhythms.

Large urine losses can cause repeated dehydration. This is especially important during vomiting, diarrhoea, fever, hot weather, or illness that makes drinking difficult.

Some people develop chronic kidney disease. Progression is not inevitable and depends on the underlying cause, extent of kidney damage, and response to treatment.

Complications may also come from the condition causing Fanconi syndrome. Cystinosis can affect the eyes, thyroid, muscles, and other organs. Multiple myeloma requires treatment beyond managing kidney losses.

Prevention

Inherited Fanconi syndrome cannot currently be prevented. Genetic counselling may help families understand inheritance and testing options.

Not every acquired case is preventable. When a medicine can injure proximal tubules, kidney and urine monitoring may identify changes before severe symptoms develop. Monitoring is particularly important when other kidney risks are present.

Following workplace and household safety measures can reduce exposure to lead, cadmium, and other kidney-toxic substances. Unregulated remedies or supplements may contain undisclosed ingredients.

If you take a medicine linked with kidney-tubule injury, continue it as prescribed unless the prescriber changes the plan. The benefits may be substantial, and stopping suddenly can be harmful.

Living with it and what to expect

The course of Fanconi syndrome is closely tied to its cause. When an acquired cause is found and removed early, kidney-tubule function may partly or fully recover over weeks or months. Recovery is not guaranteed, and some losses may continue.

Inherited forms usually require long-term management. Treatment can correct many blood abnormalities, support growth, protect bones, and reduce symptoms, but may not restore normal tubule function. Some inherited conditions also affect organs outside the kidneys.

Daily life may involve taking replacement treatments at set times, having regular blood and urine tests, and maintaining hydration. Children may need frequent dose adjustments as they grow. School, exercise, work, and travel are often possible, although treatment routines may require planning.

New bone pain, increasing thirst or urination, reduced growth, or worsening fatigue should be discussed with the care team rather than waiting for the next scheduled test. Regular

monitoring can detect changes before they become serious.

Some people maintain stable kidney function for years. Others develop worsening chronic kidney disease and may eventually need advanced kidney care. Outlook depends on the cause, existing kidney damage, and response to treatment.

When to get medical help

Arrange a routine medical appointment if you or your child has persistent excessive thirst or urination, unexplained muscle weakness, bone pain, poor growth, or repeated abnormal urine results. These symptoms have many possible causes and do not establish Fanconi syndrome by themselves.

If Fanconi syndrome has been diagnosed, contact the usual care team promptly for vomiting, diarrhoea, reduced drinking, or a clear increase in urination. These can disturb fluid and mineral levels and may require earlier testing.

Seek urgent medical help for:

Call emergency services for loss of consciousness, a seizure, severe breathing difficulty, or chest pain. These symptoms are not specific to Fanconi syndrome, but they need immediate assessment.

- Severe weakness, worsening drowsiness, or confusion
- Fainting or inability to stand because of dizziness
- A fast, irregular, or pounding heartbeat
- Very little urine, despite being unwell or dehydrated
- Inability to keep fluids or prescribed replacement treatment down
- A suspected fracture or sudden severe bone pain

Common questions

Is Fanconi syndrome the same as Fanconi anemia?

No. Fanconi syndrome affects reabsorption within the kidney's proximal tubules. Fanconi anemia is an inherited disorder that mainly causes bone-marrow failure, physical differences, and increased cancer risk. The similar names are historical.

Can Fanconi syndrome go away?

Some acquired cases improve after the cause is removed, particularly when kidney injury is

recognised early. Recovery may be incomplete or take months. Inherited forms are usually long term, although treatment can control losses and reduce complications.

Why is there sugar in my urine if I do not have diabetes?

Healthy proximal tubules return filtered glucose to the blood. In Fanconi syndrome, some glucose remains in urine even when blood glucose is normal. Blood testing is needed to distinguish this from diabetes and other causes.

Does Fanconi syndrome cause kidney failure?

It can contribute to chronic kidney disease, but kidney failure is not inevitable. Risk depends on the underlying cause, existing damage, and how well treatment controls the losses and cause.

Is there a special diet for Fanconi syndrome?

There is no single diet that corrects the tubule problem. Nutritional needs depend on phosphate, potassium, bicarbonate, kidney function, growth, and the underlying condition. Large dietary changes or mineral supplements can be harmful if they are not matched to blood results.

Can I exercise with Fanconi syndrome?

Many people can remain active. Exercise may need adjustment for significant muscle weakness, bone pain, rickets, osteomalacia, or fracture risk. Hydration and treatment schedules may also need planning around activity.

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