CONDITION

Murk jansen metaphyseal chondrodysplasia

A rare inherited condition that disrupts bone growth and the body's control of calcium and phosphate.

WHAT IT IS
Inherited condition affecting bone growth and mineral balance

BODY SYSTEM
Bones, joints, and kidneys

MAIN SYMPTOMS
Short stature, bowed limbs, enlarged joints, walking difficulty

WHAT CAUSES IT
A new or inherited PTH1R gene change

HOW IT IS DIAGNOSED
Blood tests, skeletal X-rays, and PTH1R genetic testing

MAIN TREATMENT
Mineral management, mobility support, and orthopedic care

CAN IT BE CURED?
No - the genetic change remains lifelong

IS IT AN EMERGENCY?
Sometimes - confusion or severe dehydration from high calcium

LONG-TERM OUTLOOK
Lifelong short stature; function shaped by bone and kidney complications

Overview

Murk Jansen metaphyseal chondrodysplasia is a rare genetic condition affecting bone growth. It is usually called Jansen metaphyseal chondrodysplasia.

The condition mainly affects the metaphyses. These are the wide areas near the ends of growing long bones, such as the thigh and shin bones. Changes in these areas can cause short limbs, bowed bones, enlarged joints, and difficulty walking.

The same genetic change also disrupts how the body manages calcium and phosphate. Calcium may become too high in the blood, while phosphate may become too low. This can affect the kidneys, muscles, energy levels, and bone health.

The condition usually becomes noticeable during infancy or childhood. Its effects differ

between people, even within the same family. Because it affects both bone growth and mineral balance, care commonly involves specialists in bone conditions, hormones, kidneys, rehabilitation, and orthopedic surgery.

Symptoms

Bone and joint changes are usually the most visible features. They may become more noticeable as a child grows.

Possible features include:

Some people have differences in facial or skull development, such as prominent brow areas or a small lower jaw. These features are not present in everyone.

High blood calcium, called hypercalcemia, may cause few symptoms when mild. Higher levels can cause thirst, frequent urination, constipation, nausea, tiredness, weakness, poor appetite, or difficulty concentrating.

Low phosphate can contribute to muscle weakness and poor bone mineral strength. Mineral changes may be found on blood tests before they cause clear symptoms.

- Short stature, especially from shortened arms and legs
- Bowing of the legs or arms
- Widened or enlarged-looking joints
- A waddling or uneven walking pattern
- Joint stiffness or reduced movement
- Bone or joint discomfort
- Muscle weakness and reduced stamina

Causes and risk factors

The condition is caused by a change in the PTH1R gene. This gene provides instructions for a receptor that responds to parathyroid hormone and a related protein. These signals help control bone growth and the handling of calcium and phosphate.

In this condition, the receptor stays active even when it has not received the usual signal. Growth plates do not develop normally, and the kidneys may lose too much phosphate. Blood calcium can rise even though the level of parathyroid hormone is low.

The condition has an autosomal dominant inheritance pattern. This means one changed copy

of the gene is enough to cause it. If a parent has the condition, each pregnancy has a 1 in 2 chance of inheriting the gene change.

Many affected people have a new, or de novo , gene change that was not inherited from either parent. Nothing a parent did during pregnancy caused that change.

Having the gene change is the main risk factor. Diet, physical activity, infection, and ordinary childhood injuries do not cause the condition.

Diagnosis

Diagnosis is based on the pattern of physical findings, blood and urine results, bone X-rays, and genetic testing.

Blood tests commonly show high calcium, low phosphate, and low or suppressed parathyroid hormone. Tests may also assess kidney function, vitamin D, and other markers of bone and mineral metabolism. Urine testing can show how much calcium and phosphate the kidneys are losing.

X-rays may show widened, irregular, cupped, or frayed metaphyses near the ends of long bones. These changes can resemble rickets, but Jansen metaphyseal chondrodysplasia has a different cause and a different pattern of blood results.

A genetic test that finds a disease-causing change in PTH1R can confirm the diagnosis. Testing the biological parents may show whether the change was inherited or occurred for the first time in the affected person.

Other bone-growth conditions can produce similar short stature or X-ray findings. The combination of skeletal changes, high calcium, low phosphate, and low parathyroid hormone helps distinguish this condition.

Treatment and management

There is no treatment that removes the underlying gene change. Care focuses on controlling mineral abnormalities, protecting the kidneys, supporting movement, and treating bone or joint problems.

A specialist may monitor blood calcium, phosphate, kidney function, and urine calcium. The timing of tests is based on age, previous results, symptoms, and treatment.

Treatment for high calcium may include careful attention to hydration and, in selected cases, medicines that reduce calcium levels or slow calcium release from bone. Bisphosphonates

have been used in some people, but their effects may be incomplete. There is no single medicine that is suitable for everyone with this condition.

Changes to calcium, phosphate, or vitamin D intake should be made only with specialist guidance. Ordinary supplements can affect blood and urine mineral levels, and treatment needs to be matched to test results.

Physical and occupational therapy can help maintain joint movement, muscle strength, balance, and independence. Mobility aids, supportive footwear, or changes at school or work may reduce strain and make daily activities easier.

An orthopedic surgeon may assess severe bowing, joint alignment, pain, or walking difficulty. Surgery can sometimes improve alignment or function, but it does not correct the genetic cause. Further procedures may be needed as bones grow.

Pain management should be tailored to the cause of the pain and its effect on daily life. Emotional support and practical help may also be useful when short stature, visible differences, or mobility limits affect confidence or participation.

Complications

High urine calcium can lead to kidney stones or nephrocalcinosis , which means calcium deposits within the kidneys. Persistent mineral abnormalities may eventually affect kidney function, so kidney monitoring is important even when there are no urinary symptoms.

Progressive bowing and poor joint alignment can contribute to pain, stiffness, early joint wear, and reduced mobility. Muscle weakness may add to walking difficulty.

Complications are not the same for everyone. Regular follow-up is intended to find changes before they cause greater problems.

Prevention

Murk Jansen metaphyseal chondrodysplasia cannot be prevented through diet, exercise, pregnancy care, or lifestyle changes. It is caused by a genetic change.

If the specific PTH1R change is known, a genetics professional can explain inheritance and available reproductive testing options. Choosing whether to have testing is personal.

Kidney and mobility complications cannot always be prevented, but monitoring and timely treatment may reduce their effects.

Living with it and what to expect

The condition is lifelong. Skeletal differences often become more apparent during childhood while the bones are growing. Bowing, joint alignment, and walking ability may change over time. Adult height is usually substantially shorter than average.

Once growth is complete, the growth-plate changes no longer progress in the same way. However, established bone shape, short stature, joint strain, and mineral abnormalities may remain. Adults may continue to need kidney monitoring, blood tests, pain care, rehabilitation, or orthopedic review.

Some people walk independently, while others use braces, walking aids, or a wheelchair for certain distances or activities. Needs can change after surgery, during periods of pain, or as joint wear develops.

School and work adjustments may include accessible seating, extra time between locations, reduced lifting, or equipment placed within reach. These adjustments address the physical environment rather than a person's ability to learn or make decisions.

Routine follow-up may involve an endocrinology or metabolic bone clinic, kidney assessment, orthopedic review, and rehabilitation support. The exact schedule is based on symptoms, mineral levels, kidney findings, and stage of growth.

When to get medical help

Arrange routine medical review if there is increasing bone or joint pain, worsening bowing, a change in walking, reduced joint movement, or new difficulty with everyday activities. Increased thirst, frequent urination, constipation, nausea, or unusual tiredness can be signs that calcium levels need checking.

Prompt medical assessment is appropriate for symptoms of a kidney stone, such as strong pain in the side or back, pain when urinating, or blood in the urine. Sudden pain after a fall or an inability to bear weight also needs assessment for an injury.

Seek emergency help for confusion, extreme drowsiness, repeated vomiting with inability to keep fluids down, fainting, severe weakness, or signs of marked dehydration. These symptoms can occur with severe hypercalcemia or another urgent problem.

Routine follow-up is still important when you feel well because high calcium, urinary calcium, or kidney deposits may not cause early symptoms.

Common questions

Is Jansen metaphyseal chondrodysplasia a form of rickets?

No. Its X-ray appearance can resemble rickets because both conditions affect growing bone near the metaphyses. However, Jansen metaphyseal chondrodysplasia is caused by an activating PTH1R gene change and usually has a characteristic pattern of high calcium, low phosphate, and low parathyroid hormone.

Can it be cured?

There is currently no cure that corrects the gene change throughout the body. Treatment can address high calcium, kidney risks, pain, limb alignment, and mobility. Research into treatments that act more directly on the abnormal receptor is ongoing, but these are not established cures.

Will my child keep getting shorter?

A child does not usually lose height. Instead, the long bones grow more slowly and unevenly, so the difference from average height may increase during childhood. A specialist can track growth without promising a specific adult height.

Does everyone with the gene change have severe symptoms?

No. The condition can differ in severity. The exact gene change, mineral levels, bone alignment, kidney effects, and response to treatment can all influence daily function. Even relatives with the same condition may be affected differently.

Can someone with this condition have children?

Yes. Fertility is not necessarily affected. A person with the condition has a 1 in 2 chance of passing the changed gene to each child. Genetic counseling can explain testing options and support personal reproductive decisions.

Should calcium and vitamin D be avoided?

Not automatically. Both too much and too little can be harmful, and needs differ according to blood and urine results. Supplements, fortified products, and major dietary restrictions should be discussed with the specialist managing mineral levels.

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