CONDITION

Jansen disease

Jansen disease is a rare inherited condition that affects bone growth and raises blood calcium levels.

WHAT IT IS
Rare inherited bone-growth condition with abnormal calcium regulation

BODY SYSTEM
Bones, growth plates, kidneys, and mineral balance

MAIN SYMPTOMS
Short stature, bowed limbs, waddling gait, bone or joint pain

WHAT CAUSES IT
PTH1R gene change, usually new or autosomal dominant

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

MAIN TREATMENT
Calcium control, monitoring, rehabilitation, and orthopedic care

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

IS IT AN EMERGENCY?
Sometimes - severe hypercalcemia causing confusion or dehydration

LONG-TERM OUTLOOK
Lifelong condition shaped by skeletal and kidney effects

Overview

Jansen disease is more formally called Jansen metaphyseal chondrodysplasia . It is a rare genetic condition that affects the growth plates near the ends of long bones. These are the areas where bones lengthen during childhood.

The condition can cause short stature, bowed arms or legs, and changes in walking. It also disrupts the body's control of calcium and phosphate. Blood calcium may become too high, while blood phosphate may be low.

These mineral changes happen even though the parathyroid hormone level is usually low. Parathyroid hormone, or PTH, normally helps control calcium and phosphate. In Jansen disease, bone and kidney cells behave as if they are receiving a constant PTH signal.

High calcium may affect the kidneys, muscles, digestive system, and nervous system. Regular monitoring is therefore important even when you or your child feels well.

Jansen disease is not contagious. It is not caused by diet, exercise, or anything a parent did during pregnancy.

Symptoms

Symptoms usually become noticeable during infancy or childhood, although their timing and severity are not the same for everyone.

Changes in bone growth may include:

Some children have delayed motor skills because altered bone shape makes standing and walking harder. Muscle weakness caused by high calcium can add to these difficulties.

High blood calcium, called hypercalcemia , does not always cause obvious symptoms. When symptoms occur, they may include increased thirst, frequent urination, constipation, reduced appetite, nausea, tiredness, weakness, or difficulty concentrating.

Facial or jaw differences can occur, but they are not present in everyone. Adult teeth may develop or come through abnormally in some people.

Symptoms cannot confirm Jansen disease on their own. Other bone, hormone, and mineral conditions can cause similar findings.

- Short stature, especially from shortened arms and legs
- Bowing of the legs or arms
- Enlarged or irregular joints
- A waddling or uneven walking pattern
- Bone or joint pain
- Difficulty with movement or physical endurance

Causes and risk factors

Jansen disease 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 called PTH-related peptide.

The altered receptor stays active when it should be switched off. This disrupts normal growth-plate development and causes the kidneys and bones to handle calcium and

phosphate abnormally.

The condition follows an autosomal dominant inheritance pattern. This means one altered copy of the gene is enough to cause it. A person with the condition has a 1 in 2 chance of passing the altered gene to each child.

Many diagnosed people are the first person in their family to have the condition. In these cases, the genetic change occurred during the formation of an egg or sperm, or early after conception. This is called a de novo , or new, genetic change.

Having a new genetic change does not mean other family members caused or could have prevented it. Once someone has the change, however, it can be passed to their children.

Diagnosis

Diagnosis usually combines medical history, examination, blood and urine tests, skeletal X-rays, and genetic testing.

Blood tests commonly show:

These results are unusual because most conditions that act like excess parathyroid hormone involve a high hormone level. The combination of high calcium with low PTH helps narrow the possible causes.

Urine tests may measure calcium and assess how the kidneys are handling minerals. Kidney imaging may be used to look for stones or calcium deposits.

X-rays can show characteristic changes near the ends of long bones. The metaphyses-the wider areas next to the growth plates-may appear widened, irregular, cupped, or frayed. These findings can resemble rickets, but the blood-test pattern is different.

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

Because the condition is extremely rare, assessment may involve a clinical geneticist, pediatric or adult endocrinologist, metabolic bone specialist, and orthopedic specialist.

- High calcium
- Low phosphate
- Low or suppressed parathyroid hormone
- Other changes related to bone and mineral metabolism

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on controlling high calcium, protecting the kidneys, supporting movement, and treating skeletal problems.

Blood and urine tests are used to follow calcium, phosphate, kidney function, and other markers of bone health. Kidney ultrasound or other imaging may be repeated if there is concern about stones or calcium deposits.

Treatment for high calcium is based on its level, symptoms, and effects on the kidneys. Adequate fluid intake may be important, but the amount should be discussed with the clinical team if kidney or heart problems are present. Medicines that lower calcium or alter bone turnover may be considered by a metabolic bone specialist. Evidence is limited because so few people have Jansen disease.

Do not start calcium, phosphate, or vitamin D supplements unless the treating specialist recommends them. Low blood phosphate does not automatically mean that a phosphate supplement is safe. Likewise, severe dietary calcium restriction is not a substitute for medical care and could affect nutrition or bone health.

Physical and occupational therapy may help with strength, balance, mobility, daily activities, and safe use of mobility aids. Supportive footwear, braces, or other devices may improve comfort for some people.

An orthopedic specialist can monitor limb alignment, joint function, and walking. Surgery may be considered for severe bowing, pain, loss of function, or other major deformity. The timing and type of surgery are individualized, particularly while a child is still growing.

Dental assessment can help identify problems with tooth development or alignment. Genetic counseling can explain inheritance, testing options, and what results may mean for relatives.

Complications

Long-term high calcium can increase the amount of calcium passing through the kidneys. This may lead to kidney stones or nephrocalcinosis , which means calcium deposits within kidney tissue. Kidney function can be affected in some people.

Skeletal complications may include increasing limb deformity, difficulty walking, joint strain, chronic pain, and reduced mobility. Abnormal joint loading over many years may contribute to early joint wear.

Falls and fractures are possible, especially when weakness, poor balance, or substantial bone

deformity is present. However, fracture risk is not identical for everyone with the condition.

Marked hypercalcemia can cause dehydration, vomiting, confusion, severe weakness, or changes in heart rhythm. These problems need prompt assessment, but they are not the everyday course for every person with Jansen disease.

Emotional and social effects also matter. Short stature, visible skeletal differences, repeated appointments, pain, or mobility limits can affect school, work, independence, and self-confidence. Psychological support, school adjustments, workplace accommodations, and disability services may be useful when needed.

Prevention

Jansen disease cannot be prevented through diet, exercise, pregnancy care, or lifestyle changes. The genetic change is either inherited or occurs spontaneously.

Complications may sometimes be reduced through regular monitoring and early treatment. This includes checking calcium levels and kidney health, following bone growth, assessing mobility, and reviewing new symptoms.

If you have Jansen disease or a confirmed PTH1R change, genetic counseling can explain the chance of passing it on. Counseling can also provide information about reproductive testing options without directing you toward a particular decision.

Living with it and what to expect

Jansen disease is lifelong. Bone-growth differences are usually most active during childhood, when the growth plates are open. Short stature and established changes in bone shape generally remain in adulthood, even though the pattern may change after growth is complete.

Some people need repeated orthopedic reviews or procedures as they grow. Others manage with rehabilitation, mobility support, pain care, and periodic monitoring. The course cannot be predicted from the gene result alone.

High calcium may continue after childhood, so adults still need follow-up. Feeling well does not always mean the calcium level or kidneys are unaffected. Routine appointments may include blood tests, urine tests, blood pressure checks, kidney assessment, and review of pain and mobility.

Everyday activities can often be adapted around physical needs. Low-impact movement may help maintain strength and joint function when it is comfortable and approved for the person's

skeletal situation. School or workplace adjustments can reduce unnecessary physical strain while supporting independence.

Pregnancy may require coordinated care because calcium balance can change during pregnancy and breastfeeding. A person with Jansen disease who is planning a pregnancy can discuss monitoring with an endocrinology, genetics, and maternity team.

Most long-term planning is based on the individual's skeletal changes, calcium control, kidney health, pain, and mobility rather than the name of the condition alone.

When to get medical help

Arrange routine medical review if there is increasing thirst, frequent urination, constipation, reduced appetite, persistent tiredness, new bone or joint pain, or a change in walking. These symptoms may have many causes, but they can justify checking calcium and reviewing bone health.

Seek prompt medical advice for symptoms that could indicate a kidney stone, such as strong pain in the side or back, blood in the urine, pain when urinating, or repeated vomiting.

Get urgent or emergency help for:

These symptoms do not prove that calcium is dangerously high, but they need urgent assessment.

- New confusion, unusual drowsiness, or difficulty waking
- Repeated vomiting with inability to keep fluids down
- Signs of severe dehydration, such as very little urine or fainting
- Sudden severe weakness
- Chest pain, collapse, or a new irregular or very fast heartbeat
- A serious fall, visible limb deformity, or inability to bear weight

Common questions

Is Jansen disease the same as rickets?

No. Both can cause bowed limbs and abnormal-looking growth plates on X-rays. Jansen disease has a distinct genetic cause and typically produces high calcium, low phosphate, and low PTH. Rickets has several other causes and a different biochemical pattern.

Why is calcium high if parathyroid hormone is low?

The altered PTH1R receptor sends a continuous signal inside certain cells. The body then responds as though parathyroid hormone were present, even when the actual hormone level is low.

Can Jansen disease be cured?

There is currently no cure that corrects the genetic change throughout the body. Treatment can address high calcium, kidney risks, mobility problems, pain, and significant bone deformity.

Will I pass it on to my children?

A person with the altered gene has a 1 in 2 chance of passing it to each child. The outcome of one pregnancy does not change the chance for the next. A genetic counselor can explain testing and reproductive options.

Does everyone with Jansen disease need surgery?

No. Surgery is considered when bone deformity causes major pain, reduced function, difficulty walking, or other significant problems. Some people are managed with monitoring, therapy, mobility support, and treatment of calcium-related effects.

Is Jansen disease cancer?

No. It is an inherited bone-growth and mineral-balance condition, not a form of cancer. The word "chondrodysplasia" describes abnormal cartilage and bone development at the growth plates.

Sources

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