CONDITION

Nail patella syndrome

Nail patella syndrome is an inherited condition affecting the nails, joints, kidneys, and eyes.

WHAT IT IS
Inherited condition affecting nails, joints, kidneys, and eyes

BODY SYSTEM
Bones, joints, nails, kidneys, and eyes

MAIN SYMPTOMS
Unusual nails, small kneecaps, elbow stiffness, joint pain

WHAT CAUSES IT
Inherited LMX1B gene change

HOW IT IS DIAGNOSED
Examination, X-rays, and LMX1B genetic testing

MAIN TREATMENT
Symptom relief, physical therapy, kidney and eye care

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

IS IT AN EMERGENCY?
Sometimes - sudden vision loss needs emergency care

LONG-TERM OUTLOOK
Most live full lives with lifelong monitoring

Overview

Nail patella syndrome is a rare inherited condition that affects how several parts of the body develop. It is present from birth, although some features may not be noticed until later.

The name comes from two common features: unusual fingernails and small or missing patellas, which are the kneecaps. The condition can also affect the elbows, hips, kidneys, and eyes. The type and severity of symptoms can be very different, even among relatives with the same genetic change.

Nail and joint differences may affect comfort, grip, movement, or appearance. Kidney problems and glaucoma are less visible but more important to monitor. Glaucoma is damage to the nerve at the back of the eye, often linked to raised pressure inside the eye.

Nail patella syndrome is lifelong. There is no treatment that removes its genetic cause, but

many symptoms can be managed. Regular kidney and eye checks can find complications before they cause obvious symptoms.

Symptoms

The most recognisable changes usually involve the fingernails. Nails may be small, narrow, ridged, split, discoloured, or partly missing. The thumbnails are often affected most, while the little fingernails may look nearly normal. Toenails can also be affected, but usually less severely.

The pale half-moon at the base of a nail, called the lunula, may be triangular rather than rounded. This can be a useful clue during diagnosis.

Kneecaps may be small, unusually shaped, unstable, or absent. They can move out of position, which is called dislocation. This may cause knee pain, clicking, weakness, or a feeling that the knee is giving way. Some people have difficulty fully straightening the knee or develop wear-and-tear arthritis over time.

Elbow changes may limit the ability to straighten the arm or turn the palm upward. The elbow may dislocate, and the head of one forearm bone may be underdeveloped.

Many people have bony projections on both sides of the pelvis called iliac horns. These usually do not cause symptoms. They are often discovered on an X-ray and are strongly associated with nail patella syndrome.

Other possible symptoms include:

Kidney involvement may cause protein or blood to leak into the urine. This often causes no early symptoms. If kidney function becomes more affected, possible signs include swelling around the eyes or ankles, tiredness, high blood pressure, or changes in urination.

Glaucoma also tends to be silent at first. Without treatment, it can gradually reduce side vision and eventually affect central vision.

- Back, hip, foot, or ankle pain
- Reduced muscle bulk in the arms or legs
- Numbness, tingling, or altered sensation
- Constipation or irritable bowel-type symptoms
- Increased sweating, especially on the hands and feet
- Thin tooth enamel or other dental problems

Causes and risk factors

Nail patella syndrome is usually caused by a change in a gene called LMX1B . This gene helps guide the development of the limbs, nails, kneecaps, kidneys, and parts of the eye before birth.

The condition follows an autosomal dominant inheritance pattern. This means a person needs only one altered copy of the gene to have the condition. If a parent has nail patella syndrome, each child has a 1 in 2 chance of inheriting the altered gene.

Sometimes the genetic change occurs for the first time in a child. This is called a new, or de novo, genetic change. It is not caused by anything a parent did before or during pregnancy.

The same LMX1B change can cause mild features in one family member and more significant kidney, eye, or joint problems in another. Genetic testing cannot reliably predict how severely someone will be affected.

Nail patella syndrome is not caused by diet, exercise, infection, or an injury. These factors do not make the underlying genetic condition spread or pass from one person to another.

Diagnosis

A clinician may suspect nail patella syndrome after examining the nails, kneecaps, elbows, and hips. A family history of similar features can support the diagnosis, but the condition can still occur without a known family history.

X-rays can show small or absent kneecaps, changes around the elbows, or iliac horns on the pelvis. Iliac horns are not harmful by themselves, but they are a distinctive diagnostic clue.

A blood or saliva test can look for a disease-causing change in the LMX1B gene. Finding one can confirm the diagnosis. Genetic testing may also help relatives understand whether they carry the same change. However, a negative result does not always exclude nail patella syndrome when the physical features strongly fit the condition.

Assessment usually also includes blood pressure measurement and urine tests for blood or protein. Blood tests may be used to check how well the kidneys are filtering waste. An eye professional can measure eye pressure and examine the optic nerve for signs of glaucoma.

Treatment and management

There is no treatment that corrects the LMX1B gene change. Care is directed at relieving symptoms, protecting joints, and finding kidney or eye complications early.

Nail changes may not need treatment. Keeping nails short, moisturising the skin around them, and protecting the fingertips from repeated injury can reduce discomfort. A skin or nail specialist can assess painful splitting, infection, or other nail problems.

Physical therapy can help maintain strength, flexibility, balance, and joint stability. Low-impact activities may be more comfortable when the knees or elbows are affected. Braces, shoe inserts, walking aids, or other supports may help with instability.

An orthopaedic specialist may consider surgery for repeated kneecap dislocation, severe joint instability, restricted movement, or significant pain. The unusual bone anatomy can make planning more complex, so surgery is tailored to the specific joint problem.

Kidney care depends on urine findings, blood pressure, and kidney function. Medicines that lower blood pressure may also help reduce protein loss through the kidneys. More advanced kidney problems may need specialist treatment, and kidney failure can be treated with dialysis or transplantation.

Some anti-inflammatory painkillers like ibuprofen or naproxen can affect kidney function. If you have kidney involvement, check whether an over-the-counter pain medicine is suitable before using it regularly.

Glaucoma is treated with eye drops, laser treatment, surgery, or a combination of these. Treatment cannot restore vision already lost to glaucoma, but it can reduce the chance of further damage.

Ongoing monitoring commonly includes kidney tests, blood pressure checks, and regular eye examinations. The timing should reflect age, previous results, symptoms, and family history.

Complications

Kidney involvement ranges from small amounts of protein or blood in the urine to chronic kidney disease. Only some people develop major loss of kidney function, but progression can occur without early symptoms. High blood pressure may be both a sign and a cause of further kidney damage.

Glaucoma can permanently damage vision if it is not detected and treated. Because early glaucoma usually causes no pain or noticeable sight change, routine eye examinations remain important even when vision seems normal.

Joint instability can lead to repeated dislocations, falls, long-term pain, or earlier arthritis. Limited elbow movement may affect dressing, lifting, or turning the hand. Muscle weakness and altered sensation can add to mobility problems.

Pregnancy does not usually change the underlying condition, but kidney involvement or high blood pressure can increase the need for closer monitoring. A person with unusual pelvic anatomy or previous joint surgery may also need individual planning for labour and anaesthesia.

Prevention

Nail patella syndrome cannot be prevented through lifestyle changes because it is genetic. Nothing done during pregnancy causes the condition.

Complications cannot always be prevented either, but monitoring can lower the chance of avoidable harm. Kidney tests can identify protein leakage before kidney function declines, while eye examinations can find glaucoma before noticeable vision loss.

If an LMX1B change has been identified in a family, genetic counselling can explain inheritance and testing options. Prenatal testing and testing of embryos created through in vitro fertilisation may be possible. These are personal choices rather than required steps.

Joint protection, suitable exercise, and early treatment of instability may reduce pain or injury, but they do not change the genetic condition itself.

Living with it and what to expect

Nail and bone differences are generally lifelong. Nail appearance may remain fairly stable, while joint pain or stiffness can change with activity, injury, and age. Some people need little treatment beyond monitoring. Others use physical therapy, mobility supports, pain management, or surgery.

Kidney and eye complications may appear later, even if childhood tests were normal. This is why follow-up usually continues throughout life. Abnormal urine results do not always lead to kidney failure, but they may require more frequent testing or kidney specialist care.

Most people can attend school, work, exercise, have relationships, and raise families. Activities may need adjustment when kneecaps are unstable or elbows have limited movement. An occupational therapist can suggest practical changes for writing, dressing, cooking, work tasks, or other daily activities.

Visible nail differences can affect confidence. Clear information for teachers, employers, or family members may reduce misunderstandings. Nail patella syndrome is not contagious, and unusual nails are not necessarily a sign of poor hygiene or infection.

It can be useful to keep copies of genetic test results, kidney results, eye reports, and previous X-rays. This may prevent repeated investigations and help new healthcare professionals understand your anatomy.

When to get medical help

Arrange routine follow-up for blood pressure, urine testing, kidney function, and glaucoma screening, even if you feel well.

Seek a prompt medical assessment if you develop:

A dislocated joint needs urgent assessment, especially if it looks deformed, cannot move, or the limb becomes numb, pale, or cold. Do not try to force it back into place yourself.

Seek emergency care for sudden loss of vision, severe eye pain with a red eye, or rapidly worsening breathlessness with marked swelling. These symptoms have several possible causes and need immediate assessment.

- New swelling around the eyes, hands, legs, or ankles
- Visible blood in the urine or a major change in urination
- New or worsening knee instability, joint pain, or repeated dislocation
- Persistent numbness, weakness, or loss of function
- Gradual loss of side vision or other new visual changes

Common questions

Is nail patella syndrome a type of arthritis?

No. It is a genetic developmental condition. However, unstable or unusually shaped joints can develop arthritis over time, causing pain and stiffness.

Will my kidney problems get worse?

Not everyone with nail patella syndrome develops significant kidney problems. Protein or blood in the urine may remain mild, but some people develop chronic kidney disease. Regular testing is the best way to identify change early; genetic test results alone cannot predict the course.

Can nail patella syndrome skip a generation?

A person who inherits the altered LMX1B gene will usually have some features, but they may be subtle. This can make the condition appear to have skipped a generation. A person who did

not inherit the family's gene change cannot pass that change to a child.

Can I exercise with small or unstable kneecaps?

Many people can exercise, but the most comfortable activities differ. Low-impact exercise such as swimming, cycling, or controlled strength work may place less stress on unstable knees. A physical therapist can help adapt exercise to your joint structure and symptoms.

Are the iliac horns dangerous?

Usually not. Iliac horns are bony projections on the pelvis that often cause no pain or movement problem. Their main importance is that they can help confirm the diagnosis on an X-ray.

Does nail patella syndrome affect life expectancy?

Many people have a typical lifespan. Serious kidney complications can affect long-term health, and untreated glaucoma can damage vision. Lifelong kidney and eye monitoring helps identify problems that can be treated.

Sources

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