CONDITION

NPS

NPS is an inherited condition that mainly affects the nails, kneecaps, elbows, hips, kidneys, and eyes.

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

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

MAIN SYMPTOMS
Abnormal nails, unstable kneecaps, elbow stiffness, joint pain

WHAT CAUSES IT
Inherited change in the LMX1B gene

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

MAIN TREATMENT
Symptom management, monitoring, therapy, and surgery when needed

CAN IT BE CURED?
No, it is a lifelong genetic condition

IS IT AN EMERGENCY?
Sometimes, with severe breathlessness or sudden vision loss

LONG-TERM OUTLOOK
Usually manageable, but kidney complications can become serious

Overview

NPS usually means nail-patella syndrome . It is a rare genetic condition that affects how certain tissues develop before birth. Its main features involve the nails, kneecaps, elbows, and pelvis. It can also affect the kidneys, eyes, and nerves.

The pattern is different for each person. Some people have only mild nail changes. Others have unstable kneecaps, pain, limited joint movement, kidney disease, or glaucoma. Glaucoma is damage to the nerve at the back of the eye, often linked to raised pressure inside the eye.

NPS is present from birth, although some features may not be noticed until later. Nail or bone changes do not usually spread through the body. However, joint symptoms may become more noticeable with activity or age, and kidney or eye problems can develop without obvious early symptoms.

For that reason, long-term kidney and eye checks matter even when you feel well.

Symptoms

Nail changes are often the first visible sign. They usually affect the thumbnails most strongly, although other fingernails and toenails can also be involved. Nails may be:

The pale half-moon at the base of a nail may also look triangular. Nail changes are usually permanent but do not always cause pain or interfere with hand use.

The kneecaps, also called patellae, may be small, unusually shaped, out of position, or absent. This can cause knee pain, clicking, weakness, or a feeling that the knee may give way. A kneecap can sometimes slip partly or completely out of place.

Elbow development may also be affected. You may have difficulty fully straightening, bending, or turning an arm. The elbows can look different on X-rays even when movement is only mildly limited.

Many people with NPS have small bony projections on the pelvis called iliac horns . These usually cause no symptoms. They are often found on an X-ray and are strongly associated with NPS.

Other possible features include:

Kidney involvement may first appear as protein or blood in the urine. This often causes no early symptoms. More advanced kidney problems can cause ankle swelling, tiredness, nausea, breathlessness, or high blood pressure.

Glaucoma may also be silent at first. If it progresses, it can gradually reduce side vision. Rarely, a sudden painful red eye, blurred vision, or rainbow-like rings around lights can indicate an acute eye-pressure problem.

- Small, narrow, split, ridged, or partly missing
- Thin or discoloured
- Triangular at the base instead of rounded
- Hip, foot, ankle, back, or shoulder pain
- Tight muscles or reduced joint movement
- Scoliosis, meaning a sideways curve of the spine
- Numbness, tingling, or altered pain and temperature sensation
- Constipation or irritable bowel-type symptoms

- Thin muscles in some areas of the arms or legs

Causes and risk factors

NPS is caused by a disease-causing change in the LMX1B gene. This gene helps control development of the limbs, nails, kidneys, eyes, and parts of the nervous system before birth.

The condition follows an autosomal dominant inheritance pattern. This means a person with NPS usually has one changed copy of the gene and one working copy. Each child of an affected parent has a 1 in 2 chance of inheriting the changed copy.

Some people inherit NPS from a parent. In others, the genetic change begins for the first time in the egg, sperm, or early embryo. This is called a new, or de novo , genetic change.

People in the same family can be affected very differently. A mildly affected parent may have a child with more noticeable joint or kidney problems, or the reverse. The gene result alone usually cannot predict how severe the condition will be.

Nothing a parent did during pregnancy causes NPS.

Diagnosis

A healthcare professional may suspect NPS from its characteristic nail and joint features and from family history. Examination often focuses on the nails, knees, elbows, hips, feet, spine, blood pressure, and eyes.

X-rays can show small or absent kneecaps, elbow changes, and iliac horns. Iliac horns are especially useful in supporting the diagnosis because they are unusual outside NPS.

A blood or saliva test can look for a disease-causing change in the LMX1B gene. Finding one can confirm the diagnosis and may help relatives understand their own risk. A negative genetic test does not always rule out NPS if the physical features are convincing.

Tests used to check the condition's effects may include:

Genetic counselling can help you understand test results, inheritance, and testing choices for relatives.

- Urine testing for protein and blood
- Blood tests of kidney function
- Blood pressure measurement

- A complete glaucoma assessment, including eye-pressure and optic-nerve checks
- Imaging or nerve tests when symptoms suggest another complication

Treatment and management

There is no treatment that removes the genetic cause of NPS. Care is aimed at protecting kidney and eye health, improving movement, reducing pain, and treating specific complications.

Regular kidney monitoring may include blood pressure checks and urine tests. Blood tests are added when kidney involvement is suspected or already known. If protein is found in the urine or kidney function changes, a kidney specialist may recommend medicines and closer follow-up.

Routine eye examinations are important because glaucoma may not cause early symptoms. If glaucoma develops, treatment may include pressure-lowering eye drops, laser treatment, or surgery. Treatment aims to prevent further sight loss; it cannot usually restore vision already lost to glaucoma.

Joint and mobility care may involve:

Surgery requires careful planning because the bones, joints, muscles, nerves, and blood vessels may differ from usual anatomy. Results can differ between people, so the expected benefits and limits should be discussed beforehand.

Pain medicines may help, but kidney health can affect which medicines are suitable. In particular, frequent use of non-steroidal anti-inflammatory drugs, such as ibuprofen or naproxen, may be unsuitable when kidney disease is present. A clinician or pharmacist can check whether a medicine is safe for you.

Nail treatment is usually cosmetic or protective. Keeping rough edges smooth, avoiding repeated trauma, and using gloves for irritating work may reduce splitting or discomfort.

- Physiotherapy to support strength, balance, and safe movement
- Occupational therapy for daily tasks, school, or work
- Braces, supportive footwear, or mobility aids
- Pain-management strategies
- Orthopaedic surgery for severe instability, deformity, or loss of function

Complications

Kidney disease is one of the most important possible complications. It can range from small amounts of protein in the urine to long-term loss of kidney function. A minority of affected people develop severe kidney failure. Early monitoring allows changes to be addressed before symptoms become obvious.

High blood pressure can occur with kidney involvement and can place further strain on the kidneys and circulation.

Glaucoma can cause permanent sight loss if it is not detected and treated. The risk is why routine eye checks remain important even when your vision seems normal.

Joint complications can include repeated kneecap dislocation, early wear of joint surfaces, chronic pain, weakness, and reduced mobility. Falls may become more likely if a knee is unstable.

Nerve-related differences can increase sensitivity to pain or reduce awareness of temperature in some people. Reduced temperature sensation may make burns harder to notice.

Pregnancy does not usually change the inherited condition itself. However, existing kidney disease, protein in the urine, or high blood pressure can increase pregnancy-related risks. Additional kidney, blood pressure, and maternity monitoring may be advised.

Prevention

NPS cannot be prevented because it results from a genetic change that affects development before birth. Diet, exercise, vaccinations, and pregnancy behaviour do not prevent the condition.

Some complications may be detected early or reduced through regular monitoring. Kidney checks, blood pressure measurement, glaucoma screening, and appropriate joint care can help protect health and function.

If you have NPS and are considering pregnancy, genetic counselling can explain the chance of passing it on. It can also provide information about reproductive testing options. These are personal choices, and counselling should support rather than direct your decision.

Living with it and what to expect

NPS is lifelong. Nail and skeletal differences usually remain, although their effect on daily life may change. Children may need support with running, stairs, sports, handwriting, or activities

that require full elbow movement. Many people attend mainstream school, work, have relationships, and live independently.

Joint pain or instability may become more troublesome over time, especially after injury or with repeated strain. Strengthening, pacing activities, supportive equipment, and changes to work or exercise can help. Staying active within comfortable limits generally supports muscle strength and joint stability.

Kidney and eye complications do not affect everyone, and their course cannot be predicted from nail or knee changes. Someone with mild-looking physical features can still develop kidney disease or glaucoma. Regular follow-up should therefore continue through adulthood.

Monitoring is usually routine rather than urgent. It may include periodic blood pressure and urine checks, kidney blood tests when indicated, and complete eye examinations. The timing is based on your age, symptoms, previous results, and local specialist advice.

Emotional effects also matter. Visible nails, an unusual walk, pain, or uncertainty about inheritance may affect confidence. Practical support, counselling, workplace adjustments, or contact with a rare-condition support group may be useful.

When to get medical help

Arrange a routine appointment if you develop new joint pain, repeated kneecap slipping, reduced movement, numbness, swelling, or changes in your ability to walk or manage daily activities. Also seek follow-up if routine kidney or eye checks are overdue.

Contact a healthcare professional promptly if you notice:

Seek urgent medical help for a kneecap or other joint that has moved out of place, especially if the limb looks deformed, becomes numb, changes colour, or cannot bear weight. Do not try to force it back into position.

Seek emergency care for severe breathlessness, confusion, fainting, chest pain, or very little urine with rapidly increasing swelling. A sudden painful red eye, sudden loss of vision, or severe blurred vision with nausea also needs emergency eye assessment.

- Blood in your urine
- New ankle or facial swelling
- Persistently foamy urine
- A clear reduction in urination
- New headaches or blurred vision

- Worsening blood pressure readings

Common questions

Is NPS the same as nail-patella syndrome?

Yes. In this context, NPS is the usual abbreviation for nail-patella syndrome. The name highlights two common features, but the condition can also involve the elbows, pelvis, kidneys, eyes, and nerves.

Does everyone with NPS have missing kneecaps?

No. Kneecaps may be absent, but they can also be small, misshapen, or positioned differently. Some people have significant instability, while others have little knee trouble.

Can NPS affect my kidneys even if I feel well?

Yes. Protein or blood can appear in the urine before kidney disease causes symptoms. Routine urine, blood pressure, and sometimes blood testing help find changes early.

Will my child inherit NPS?

If you have an LMX1B change that causes NPS, each pregnancy has a 1 in 2 chance of passing it on. The severity in a child cannot be predicted from the parent's symptoms.

Can I exercise with NPS?

Many people can exercise, but the best activities depend on joint stability, pain, and previous injuries. Low-impact exercise and guided strengthening may be easier on unstable knees. Repeated dislocation, swelling, or increasing pain should be assessed.

Does NPS get worse with age?

The underlying genetic condition does not spread, and nail or bone differences are generally stable. Joint wear, pain, kidney disease, or glaucoma may develop or progress over time. Regular monitoring helps identify treatable changes.

Sources

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