CONDITION

Fong disease

Fong disease is an inherited condition that affects the nails, bones and joints, kidneys, and eyes.

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

BODY SYSTEM
Nails, bones and joints, kidneys, and eyes

MAIN SYMPTOMS
Abnormal nails, unstable kneecaps, joint pain, limited elbow movement

WHAT CAUSES IT
Inherited changes in the LMX1B gene

HOW IT IS DIAGNOSED
Clinical examination, bone X-rays, and LMX1B genetic testing

MAIN TREATMENT
Joint support, symptom treatment, and kidney and eye monitoring

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

IS IT AN EMERGENCY?
No - sudden vision loss needs emergency assessment

LONG-TERM OUTLOOK
Usually manageable; kidney or eye complications can be serious

Overview

Fong disease is another name for nail-patella syndrome , a lifelong inherited condition affecting how certain body parts develop. The nails and kneecaps are most often affected, but changes can also involve the elbows, pelvis, kidneys, and eyes.

The name describes two characteristic features: unusual fingernails and small or missing kneecaps. The medical word for a kneecap is patella . However, the condition varies widely, even among members of the same family.

Some people have only mild nail changes. Others experience unstable joints, chronic pain, kidney disease, or glaucoma. Glaucoma damages the nerve at the back of the eye and is often associated with increased pressure inside the eye.

Kidney and eye complications may not cause symptoms at first. Regular monitoring is therefore important, even when nail and joint changes are mild.

Symptoms

Features are usually present from birth, although some are not noticed until later.

Nail changes commonly affect the thumbnails and index fingernails most strongly. Nails may be small, split, ridged, unusually shaped, or partly or completely absent. The pale half-moon at the nail base may be triangular rather than curved. Toenails can also be affected.

Kneecaps may be small, irregularly shaped, or absent. They can move out of position, making the knee feel unstable or causing it to give way. Pain may occur during walking, running, climbing stairs, or sitting with bent knees. Muscles around an affected knee may be weaker or smaller.

Elbow changes can limit straightening of the arm or rotation of the forearm. This may cause pain, stiffness, or difficulty turning the palm up and down.

X-rays may show pointed pieces of bone called iliac horns on the pelvis. These are strongly associated with nail-patella syndrome and usually do not cause symptoms.

Other possible features include:

Kidney involvement ranges from small amounts of protein in the urine to long-term loss of kidney function. Laboratory tests are needed because protein leakage may not be visible.

Glaucoma can also develop without early symptoms. Untreated optic nerve damage may gradually cause permanent vision loss.

- Back, hip, foot, or ankle pain
- Early joint wear, called osteoarthritis
- Reduced sensation to pain or temperature in parts of the limbs
- Protein or blood in the urine
- High pressure inside the eye or glaucoma

Causes and risk factors

Fong disease is caused by a harmful change, or pathogenic variant, in the LMX1B gene. This gene helps guide development of the limbs, nails, kneecaps, kidneys, and parts of the eye before birth.

The condition has an autosomal dominant inheritance pattern. A person needs only one changed copy of the gene to have it. If a parent carries the variant, each child has a 1 in 2 chance of inheriting it.

Sometimes the variant begins for the first time in an affected person. This is called a de novo variant. That person can still pass it to their children.

Symptoms in a child cannot be predicted from the affected parent's symptoms. One relative may have mainly nail changes, while another has more significant joint or kidney problems.

Fong disease is not caused by diet, exercise, infection, lifestyle choices, or something that happened during pregnancy.

Diagnosis

Diagnosis begins with a medical and family history and an examination of the nails, knees, elbows, and other joints. A clinician may look for triangular nail half-moons, small kneecaps, limited elbow movement, and similar findings in relatives.

X-rays can show small or absent kneecaps, elbow changes, and iliac horns on the pelvis. A blood or saliva genetic test can identify a pathogenic variant in the LMX1B gene, confirming the diagnosis and allowing targeted testing of relatives.

A clinical diagnosis may still be possible when physical and X-ray findings are characteristic but genetic testing does not identify a variant.

Assessment for complications may include:

Kidney and eye findings are not required for diagnosis. Normal results at one examination do not guarantee that complications will never develop.

- Blood pressure measurement
- Urine tests for blood, protein, and albumin
- Blood tests to estimate kidney function
- A complete eye examination, including eye pressure and optic nerve assessment
- Joint assessment for pain, instability, or restricted movement

Treatment and management

There is no treatment that removes the LMX1B gene change. Care aims to maintain movement, control symptoms, treat complications, and monitor the kidneys and eyes.

Physiotherapy can improve strength, balance, and joint control while reducing strain on unstable knees or elbows. Braces, supports, shoe inserts, or other aids may help. An occupational therapist can recommend adjustments for school, work, or daily activities.

Pain treatment depends on its cause and the person's kidney health. Some medicines, including non-steroidal anti-inflammatory drugs such as ibuprofen, can affect kidney function when used frequently. Ask a clinician or pharmacist what is suitable before using these regularly.

Orthopaedic surgery may be considered for repeated kneecap dislocation, severe instability, deformity, or loss of function. Because bone shape can be unusual, surgery requires careful planning. Not everyone needs an operation.

Protecting nails from repeated injury and wearing gloves for wet or manual work may reduce splitting and irritation. Redness, warmth, swelling, or discharge around a nail may indicate infection and needs assessment.

Kidney treatment can include blood pressure control, medicines that reduce protein loss, and specialist kidney care. Advanced kidney failure may require dialysis or a transplant, although this is not the usual outcome for everyone.

Glaucoma may be treated with pressure-lowering eye drops, laser treatment, or surgery. Early treatment offers the best chance of protecting vision.

Complications

Joint instability can cause falls, repeated dislocations, chronic pain, reduced activity, and osteoarthritis. Limited elbow movement may affect lifting, dressing, or tasks involving rotation of the hand.

Kidney disease is the most important internal complication. Protein leakage may remain mild, but some people develop high blood pressure or progressive kidney scarring. Rarely, kidney function declines enough to require dialysis or transplantation.

Increased eye pressure and glaucoma can permanently damage sight. Early glaucoma is often painless and may not immediately affect vision, so symptoms alone are not a reliable warning.

People who are pregnant or planning pregnancy may need closer monitoring if they have kidney disease or high blood pressure. They should discuss kidney health, medicines, and inheritance with their healthcare team.

Prevention

Fong disease cannot be prevented in someone who inherits the LMX1B variant. Nail, bone, and joint differences develop before birth and are not caused by something a parent did.

Regular urine, blood pressure, kidney function, and eye checks may identify complications early, when treatment can help protect kidney function or vision.

Genetic counselling can explain the chance of passing the condition to a child. If the family variant is known, prenatal testing and testing of embryos created through in vitro fertilisation may be possible. These are personal options, not requirements.

Appropriate strengthening, joint protection, and treatment of instability may reduce injuries and help preserve movement. No single exercise or diet prevents every complication.

Living with it and what to expect

Fong disease remains present throughout life, but not every feature worsens. Nail appearance and basic bone shape are usually stable. Joint symptoms may change with growth, activity, muscle strength, injuries, and age.

Knee instability can become more noticeable during growth or demanding activity. Osteoarthritis and chronic pain may develop over time, particularly in joints with abnormal alignment. Physiotherapy, activity adjustments, supports, and sometimes surgery can help people remain active.

Kidney or eye problems may first appear later, even after normal earlier checks. Ongoing routine follow-up is intended to find these silent changes before they become serious.

Many people with Fong disease attend school, work, exercise, have relationships, and raise families. Some need adjustments for pain, limited movement, or kidney or visual complications. Long-term outlook depends mainly on the severity of kidney disease and whether glaucoma is detected and treated early.

Keep a record of genetic results, X-rays, kidney tests, eye examinations, operations, and medicines. This can help when changing healthcare teams, and relatives may benefit from knowing the confirmed family variant.

When to get medical help

Arrange routine kidney tests, blood pressure checks, and complete eye examinations even if you feel well. Discuss persistent joint pain, repeated giving way, reduced movement, or

symptoms that interfere with sleep or daily activities.

Seek prompt medical advice for:

Get emergency help for sudden vision loss, severe eye pain with nausea, or a serious joint injury with obvious deformity. A limb that becomes cold, pale, blue, numb, or very weak after an injury also needs emergency assessment.

- New blood in the urine, persistent foamy urine, or swelling
- A clear reduction in urine output
- New blurred vision, halos around lights, or worsening side vision
- Repeated kneecap dislocations or rapidly increasing joint pain
- Redness, swelling, warmth, or discharge around a nail
- New high blood pressure readings or severe headaches

Common questions

Is Fong disease the same as nail-patella syndrome?

Yes. Fong disease is an older name for nail-patella syndrome. Hereditary onycho-osteodysplasia is another historical term.

Can Fong disease be cured?

No. The inherited gene change cannot currently be removed. Treatment can improve joint function, manage pain, and control kidney or eye complications.

Will I develop kidney failure?

Not necessarily. Some people never develop kidney disease, while others have protein in the urine or reduced kidney function. Severe kidney failure is possible but not inevitable. Regular urine, blood pressure, and kidney blood tests are important.

Can I pass it to my children?

Yes. A person with an LMX1B pathogenic variant has a 1 in 2 chance of passing it on in each pregnancy. The chance is the same for children of any sex, and severity cannot be predicted from the parent's symptoms.

Can I exercise with nail-patella syndrome?

Many people can exercise. The best activities depend on joint stability, pain, and previous injuries. Lower-impact exercise and strengthening may be easier on unstable knees. A physiotherapist can help adapt activities safely.

Why do I need eye checks if my vision is normal?

Glaucoma can damage the optic nerve before you notice a change in sight. Eye examinations can detect increased pressure or early nerve damage so treatment can begin sooner.

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