CONDITION

Incontinentia pigmenti

Incontinentia pigmenti is an inherited condition that affects the skin and may also affect the teeth, eyes, hair, nails, and nervous system.

WHAT IT IS
Inherited condition affecting skin and other developing tissues

BODY SYSTEM
Skin, teeth, eyes, hair, nails, and nervous system

MAIN SYMPTOMS
Blistering rash, swirling pigmentation, dental changes, sparse hair

WHAT CAUSES IT
Usually an inherited or new IKBKG gene change

HOW IT IS DIAGNOSED
Characteristic skin changes, genetic testing, sometimes skin biopsy

MAIN TREATMENT
Skin care, eye monitoring, and treatment of specific complications

CAN IT BE CURED?
No - the genetic change cannot be removed

IS IT AN EMERGENCY?
Sometimes - seizures, sudden weakness, or sudden vision loss

LONG-TERM OUTLOOK
Often good, but eye or brain complications may persist

Overview

Incontinentia pigmenti, often shortened to IP, is a genetic condition that affects tissues developing from the outer layer of an embryo. These include the skin, teeth, eyes, hair, nails, and parts of the nervous system.

The skin changes often follow curved or swirling lines called Blaschko lines . These invisible developmental patterns become noticeable when certain skin cells differ from nearby cells. The name "incontinentia pigmenti" comes from the way skin pigment appears under a microscope.

IP occurs mainly in girls and women. It is uncommon in boys because the genetic change usually has more serious effects when there is only one X chromosome. However, boys can occasionally have IP.

Skin changes are often the first sign. They may begin at birth or during the first few weeks of life. The rash itself usually improves over time, but changes involving the eyes, teeth, or nervous system can have longer-lasting effects. Early eye assessment is especially important because retinal problems may be present before vision changes are obvious.

IP is not contagious. It cannot spread through touching the rash, sharing household items, or spending time with someone who has the condition.

Symptoms

The skin findings are traditionally described in four stages. These stages can overlap, appear out of order, or be so mild that one is missed.

Blistering usually settles during infancy, although it can occasionally return during a fever or illness. The darker pigmentation often fades during childhood or adolescence. Pale or thin areas may remain into adulthood.

Dental differences are common. Teeth may be missing, unusually small, cone-shaped, widely spaced, or slow to appear. Both baby teeth and permanent teeth can be affected. These changes may affect chewing, speech, appearance, and how the upper and lower teeth meet.

Hair may be sparse, coarse, or absent in small patches. Some people have a patch of scarring hair loss on the scalp. Nails may be ridged, thickened, misshapen, or slow growing. Rarely, painful growths form beneath a nail.

Eye changes usually involve the retina , the light-sensitive tissue at the back of the eye. Abnormal retinal blood vessels can leak, scar, or pull the retina away from the back of the eye. This is called retinal detachment. An affected baby may not show an obvious eye symptom, which is why specialist examinations matter.

Possible nervous system symptoms include seizures, muscle weakness, reduced coordination, developmental delay, learning difficulties, or a smaller-than-expected head size. Many people with IP do not have nervous system involvement.

Some people also sweat less in affected skin. This can make it harder to stay comfortable in hot weather.

- Blistering stage: Groups of small blisters, bumps, or red areas, often arranged in lines on the arms, legs, or scalp
- Warty stage: Raised, rough, or crusted areas that may follow the earlier blisters
- Pigmented stage: Brown, grey, or blue-grey streaks and swirls, especially on the trunk and limbs

- Pale or thin-skin stage: Faint, hairless, or slightly indented streaks, often on the lower legs

Causes and risk factors

IP is usually caused by a harmful change in the IKBKG gene, also called NEMO . This gene helps regulate inflammation, immune responses, and cell survival during development.

The gene is located on the X chromosome, one of the sex chromosomes. IP is therefore described as an X-linked condition. A girl or woman usually has two X chromosomes. In each cell, one X chromosome is naturally switched off. This creates groups of cells using different X chromosomes and helps explain the streaked skin pattern.

A boy usually has one X chromosome. A severe IKBKG change can prevent normal development in a pregnancy with only one X chromosome. For this reason, many affected male pregnancies end in miscarriage. Boys who survive may have an extra X chromosome, a genetic change present in only some cells, or a less disruptive IKBKG variant.

The genetic change may be inherited from a parent or may arise for the first time in the affected person. A parent can have subtle signs, such as a few missing teeth or faint skin streaks, and may not have been diagnosed previously.

An affected person can pass the altered gene to a child. The chance and possible outcomes depend on the specific genetic finding and the chromosomes inherited by the pregnancy. A genetics specialist can explain these possibilities using the family's test results.

Diagnosis

A clinician may suspect IP from the pattern and timing of the skin changes, particularly when they follow Blaschko lines. Dental, hair, nail, eye, and family findings can support the diagnosis.

A genetic blood test can look for changes in the IKBKG gene. Testing commonly begins by looking for a recurring deletion involving part of the gene, followed by other testing if needed. Finding a disease-causing IKBKG variant can confirm the diagnosis and help with family testing.

A negative blood test does not always exclude IP. Some genetic changes may be difficult to detect, and a change present in only some cells may not be found in blood. A genetics team may discuss additional testing when the clinical signs remain convincing.

A skin biopsy may be used when the diagnosis is uncertain. During a biopsy, a small skin sample is removed with local numbing medicine and examined under a microscope. The findings differ

according to the rash stage.

Eye examinations, brain imaging, an electroencephalogram for seizures, hearing checks, or developmental assessments do not by themselves confirm IP. They are used to look for associated complications.

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on protecting vision, supporting development, managing symptoms, and treating complications.

The skin lesions usually heal without specific treatment. Gentle cleansing, avoiding unnecessary friction, and protecting open areas can reduce irritation. Antibiotics like amoxicillin or doxycycline are used only when there is evidence of a bacterial infection; they do not treat IP itself. Persistent, painful, or unusual skin and nail lesions may need review by a dermatologist.

A baby diagnosed with or strongly suspected of having IP should have a prompt examination by an ophthalmologist experienced in children or retinal conditions. Retinal changes can be present even when the eyes look normal. Follow-up frequency is based on age and examination findings.

Abnormal retinal blood vessels may be treated with laser therapy or freezing treatment to reduce the risk of bleeding, scarring, and retinal detachment. Retinal surgery may be needed if detachment occurs. These treatments aim to protect remaining vision, but they cannot always restore sight already lost.

A dentist familiar with developmental tooth differences can monitor tooth eruption and plan care over time. Treatment may include restoring small teeth, replacing missing teeth, orthodontic care, or dental implants when growth and development make them appropriate.

Seizures, weakness, developmental differences, and learning needs are managed according to the symptoms present. Care may involve medicines for seizures, physiotherapy, occupational therapy, speech and language therapy, educational support, or other rehabilitation services.

Complications

Retinal disease is one of the most important possible complications. Without timely detection and treatment, abnormal blood vessel growth can lead to retinal detachment and permanent sight loss. One eye may be affected more than the other.

Neurological complications can include seizures, stroke-like brain injury, movement problems, developmental delay, or intellectual disability. These complications are not present in everyone and may range from mild to severe.

Missing or misshapen teeth can affect nutrition, speech, confidence, and jaw development. Coordinated dental care can improve function as well as appearance.

Damaged skin may develop a bacterial infection, although infection is not part of IP itself. Signs can include increasing pain, warmth, swelling, pus, fever, or redness spreading beyond the original rash.

Reduced sweating may increase discomfort in hot conditions. Nail changes can cause repeated irritation or pain, particularly if a growth develops beneath a nail.

Prevention

IP itself cannot usually be prevented. It results from an inherited or newly occurring genetic change, not from anything done during pregnancy or after birth.

Genetic counselling can help people understand test results, inheritance, and reproductive choices. Options may include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal decisions, and their availability differs by location.

Although the condition cannot be prevented, some complications may be reduced through early monitoring. Prompt retinal examinations are particularly important. Regular dental, developmental, and neurological follow-up can also identify needs before they cause greater difficulty.

Living with it and what to expect

The skin appearance often changes most during infancy and early childhood. Blisters and raised lesions usually settle, while the swirling pigmentation may remain for years before fading. Pale, thin, or hairless streaks can persist into adult life.

Dental differences do not usually resolve without treatment. Care often continues in stages as baby teeth are replaced, the jaw grows, and permanent dental options become suitable.

The risk of serious retinal change is greatest early in life, but the timing is not identical for everyone. Eye follow-up should continue for as long as the ophthalmology team recommends. Vision already lost from retinal damage may be permanent.

Neurological outcomes are broad. A child without seizures, weakness, developmental

concerns, or abnormal brain findings may continue without major neurological problems. When brain involvement is present, some effects may be long term, although therapy, rehabilitation, and educational support can improve function and independence.

Many people without severe eye or nervous system complications have an active life and a usual lifespan. Ongoing concerns may centre on dental treatment, vision in an affected eye, skin appearance, heat tolerance, or family planning rather than worsening illness.

Routine follow-up may involve ophthalmology, dentistry, dermatology, genetics, and developmental care. Not every person needs every specialist indefinitely; follow-up should reflect the features actually present.

When to get medical help

Arrange prompt medical assessment if a newborn develops unexplained linear blisters or a swirling rash. Newborn rashes have several possible causes, including infections that may need rapid treatment.

A new diagnosis or strong suspicion of IP should lead to prompt specialist eye assessment, even when the baby appears to see normally. This is urgent follow-up rather than usually an emergency visit.

Seek same-day medical advice for:

Call emergency services for:

Routine concerns such as fading pigmentation, tooth shape, nail changes, school support, or family-planning questions can usually be discussed at a scheduled appointment.

- Increasing skin pain, warmth, swelling, pus, or spreading redness
- Fever or poor feeding with a blistering rash
- New staring spells, unusual movements, or possible brief seizures
- A new squint, abnormal eye movement, or concern about visual attention
- A seizure lasting several minutes or repeated seizures without recovery
- Sudden weakness, facial drooping, loss of balance, or reduced responsiveness
- Sudden loss of vision or a major sudden change in sight
- Breathing difficulty, blue or grey colouring, or difficulty waking

Common questions

Is incontinentia pigmenti contagious?

No. IP is caused by a genetic change. The skin lesions cannot spread to another person through touch, fluid from a blister, or shared objects.

Will the skin rash go away?

The blistering and raised stages usually settle during infancy. Dark streaks often fade through childhood or adolescence. Pale, thin, or hairless streaks may remain, and the exact course differs between people.

Can boys have incontinentia pigmenti?

Yes, but it is rare. Surviving boys may have an extra X chromosome, the genetic change in only some cells, or a less disruptive form of the IKBKG change.

Does incontinentia pigmenti affect intelligence?

Not always. Many people have no major brain involvement. Others may have seizures, developmental delay, learning difficulties, or intellectual disability, particularly when IP has affected the brain.

Can I pass incontinentia pigmenti to my children?

An affected person may pass the altered X chromosome to a child. The possible outcomes are different for pregnancies inheriting an XX or XY chromosome pattern. Genetic counselling can provide information based on your specific test result and family history.

Is there a cure for incontinentia pigmenti?

There is no cure that removes the genetic change. Skin findings often improve naturally, while eye, dental, neurological, and other complications can be monitored and treated.

Sources

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