CONDITION

# Hutchinson-gilford syndrome

A rare genetic condition that causes poor growth and unusually rapid aging of the heart, blood vessels, skin, and bones.

**WHAT IT IS**
Rare genetic condition causing features of early aging

**BODY SYSTEM**
Heart and circulation, bones, skin, and growth

**MAIN SYMPTOMS**
Poor growth, hair loss, tight skin, stiff joints

**WHAT CAUSES IT**
Usually a new LMNA gene change

**HOW IT IS DIAGNOSED**
Clinical assessment and LMNA genetic testing

**MAIN TREATMENT**
Lonafarnib and coordinated supportive care

**CAN IT BE CURED?**
No - treatment can slow some effects

**IS IT AN EMERGENCY?**
Sometimes - chest pain or stroke signs

**LONG-TERM OUTLOOK**
Progressive condition with life-threatening cardiovascular complications

## Overview

Hutchinson-Gilford syndrome is a very rare genetic condition. It is often called Hutchinson-Gilford progeria syndrome, HGPS, or progeria.

Children with HGPS usually look healthy at birth. During the first one or two years, growth slows and characteristic changes in the skin, hair, face, joints, and bones begin to appear. These changes resemble some features of aging, but HGPS is not the same as ordinary aging.

The condition is especially important because it causes early hardening and narrowing of the arteries, called atherosclerosis. This can reduce blood flow to the heart or brain and may lead to a heart attack or stroke at a young age.

HGPS does not usually affect intelligence. Children can learn, communicate, attend school,

form relationships, and take part in family and community life. Physical adaptations may be needed as the condition progresses.

There is currently no cure. Treatment can reduce some effects, support comfort and independence, and address heart and circulation risks. Care is usually coordinated through a specialist team familiar with HGPS.

## Symptoms

The first sign is often slower-than-expected weight gain or growth during infancy. A child's head may continue growing while the rest of the body remains small.

Hair may become thin and then fall out, including scalp hair, eyebrows, and eyelashes. The skin may look thin, tight, shiny, or uneven in color. Veins can become more visible, particularly over the scalp.

Common physical features include:

Bone and joint changes can affect posture, walking, hand movement, and everyday tasks. The hips may be vulnerable to structural problems. The fingertips can become rounded, and the nails may be small or irregular.

Some children develop hearing loss, dry eyes, sensitivity to bright light, or difficulty closing their eyelids fully. Dental problems can occur because the jaw is small and teeth may come through late or in crowded positions.

Intellectual development is usually typical. HGPS also does not usually cause the memory loss seen in some older adults.

Symptoms and their timing are not identical for every child. Features generally become more noticeable over time.

- Short height and low body weight
- Loss of body fat and muscle
- Hair loss
- A small lower jaw and a narrow, beaked nose
- Prominent eyes
- Tight skin and reduced sweating
- Stiff joints and reduced movement
- A high-pitched voice

- Delayed or crowded teeth

## Causes and risk factors

HGPS is usually caused by a specific change in the LMNA gene. This gene provides instructions for making proteins that help support the nucleus, the control center inside a cell.

The genetic change causes cells to produce an abnormal protein called progerin . Progerin disrupts the structure and function of cells. Its effects are especially important in blood vessels, skin, bones, and other connective tissues.

In nearly all children with classic HGPS, the genetic change is new. This means it occurred in the egg, sperm, or very early embryo rather than being passed down from a parent. Nothing a parent did before or during pregnancy causes this change.

HGPS follows an autosomal dominant inheritance pattern. One altered copy of the gene is enough to cause the condition. However, because most cases result from a new genetic change, parents are usually unaffected.

The chance of the condition occurring again in the same family is generally low, but it is not always zero. Rarely, a parent may have the change in some egg or sperm cells without having it in the rest of the body. This is called germline mosaicism. A genetics professional can explain recurrence risk for a specific family.

## Diagnosis

A healthcare professional may suspect HGPS from its pattern of growth and physical features. Because children usually appear healthy at birth, the diagnosis is often considered only after characteristic changes develop.

The diagnosis is confirmed with a genetic blood or saliva test that looks for a condition-causing change in the LMNA gene. Genetic testing can also help distinguish classic HGPS from other rare conditions that cause poor growth or features of early aging.

An assessment may include measurements of height and weight, a physical examination, and a review of the child's development and medical history. Tests of the heart, blood vessels, bones, hearing, vision, and teeth help establish a baseline and identify complications that need attention. These tests support care but do not replace genetic confirmation.

Genetic counseling can help families understand the test result, inheritance, and reproductive options. Testing other relatives is not routinely needed when the change is confirmed to be

new, but recommendations may differ according to the family's results.

## Treatment and management

There is no treatment that removes the genetic change or completely stops HGPS. Management aims to slow some effects, protect heart and blood vessel health, maintain movement, and support daily life.

Lonafarnib is a medicine approved in the United States for eligible people with HGPS and certain related conditions. It is a farnesyltransferase inhibitor, meaning it blocks a chemical step needed for progerin to attach normally within cells. Clinical studies found that treatment was associated with longer survival, but it is not a cure.

Lonafarnib can cause side effects, particularly diarrhea, nausea, vomiting, reduced appetite, and tiredness. A specialist determines eligibility and monitors treatment, growth, nutrition, side effects, and interactions with other medicines.

Other care is tailored to the person's needs and may include:

A specialist may consider medicines to reduce blood-clot or cardiovascular risks. These decisions require an individual assessment because the possible benefits must be balanced against risks such as bleeding or side effects.

Procedures requiring anesthesia need careful planning. HGPS can affect the airway, joints, blood vessels, and ability to maintain body temperature. The surgical and anesthesia teams should know about the condition in advance.

- Regular heart and blood vessel assessments
- Physical and occupational therapy to maintain movement
- Nutrition support for adequate energy and hydration
- Dental care for delayed, crowded, or fragile teeth
- Hearing and eye assessments
- Footwear, padding, or mobility equipment
- Skin care and protection from pressure injuries
- School adjustments for physical access and fatigue

## Complications

The most serious complications involve the heart and blood vessels. Progerin damages artery walls, encouraging early atherosclerosis. Narrowed arteries can limit blood supply and lead to chest pain, a heart attack, a transient ischemic attack, or a stroke.

Other cardiovascular complications can include high blood pressure, heart valve problems, an abnormal heartbeat, or reduced heart function.

Bone and joint changes may lead to pain, stiffness, hip problems, altered walking, or fractures. Loss of body fat can make sitting or lying in one position uncomfortable and can increase the risk of pressure injuries.

Low body weight gives children fewer reserves during vomiting, diarrhea, fever, or hot weather. Dehydration can therefore become significant more quickly than expected.

Hearing loss, dental problems, dry eyes, reduced eyelid closure, and vision difficulties can also affect comfort and participation. Regular monitoring helps identify these concerns before they cause avoidable limitations.

## Prevention

HGPS cannot currently be prevented through lifestyle choices, nutrition, medicines, or actions during pregnancy. The genetic change usually happens by chance.

After a genetic diagnosis, counseling can explain the chance of HGPS occurring in another pregnancy. If the family's specific LMNA change is known, prenatal testing or testing embryos created through in vitro fertilization may be possible. These are personal reproductive options, not treatments or guarantees.

Nothing about the diagnosis suggests that a parent caused the condition.

## Living with it and what to expect

HGPS is progressive, meaning its physical effects increase over time. Growth remains limited, and joint, skin, bone, and cardiovascular changes may become more pronounced through childhood and adolescence. Treatment may slow some effects but does not stop the condition.

Children with HGPS usually continue to develop intellectually. Many attend school with children of the same age and enjoy friendships, hobbies, travel, and family activities. Seating changes, extra time between classes, adapted physical activities, or mobility aids may help preserve participation and independence.

Energy levels and physical abilities can change. Regular therapy can help maintain flexibility without forcing joints through painful movements. Plans often need to be adjusted as the child grows and new needs appear.

Eating enough can be challenging because of a small jaw, dental problems, low appetite, or the body's high energy needs. Nutrition support focuses on practical ways to maintain intake rather than expecting typical growth.

The outlook is serious because cardiovascular complications can occur in childhood or adolescence. Life expectancy is substantially shortened, although individual outcomes cannot be predicted precisely. Lonafarnib and careful supportive care may improve survival and quality of life.

Emotional support matters for both the child and family. Honest, age-appropriate conversations can help a child take part in decisions. Counseling, peer connections, palliative care, and practical family support can be used alongside active treatment. Palliative care is not limited to the end of life; it can help with symptoms, stress, and planning at any stage.

## When to get medical help

Get emergency medical help immediately for possible heart attack or stroke symptoms, including:

Children may not describe chest symptoms in the same way as adults. Sudden unexplained distress, sweating, extreme tiredness, or breathing difficulty also needs urgent assessment.

Seek urgent medical advice for repeated vomiting or diarrhea, inability to drink, much less urination than usual, unusual sleepiness, or other signs of dehydration.

Routine follow-up is important even when the child seems well. Contact the regular care team about increasing joint pain, reduced movement, new walking difficulties, hearing or vision changes, dental problems, skin sores, or trouble eating. These concerns are not usually emergencies, but earlier support may reduce their effect on daily life.

- New chest pain, pressure, or tightness
- Sudden trouble breathing
- Sudden weakness or numbness, especially on one side
- A drooping face or new difficulty speaking
- Sudden loss of vision, severe dizziness, or loss of balance
- Collapse, unresponsiveness, or a seizure

## Common questions

Is progeria inherited from a parent?

Usually not. Most children with HGPS have a new LMNA gene change that was not found in either parent. The condition can technically be inherited, but parent-to-child transmission is unusual. Genetic counseling can provide information based on the family's actual test result.

Is there a cure for Hutchinson-Gilford syndrome?

No. Current treatment cannot correct the genetic change or remove all progerin from the body. Lonafarnib can interfere with progerin's harmful effects and has been associated with improved survival. Supportive care addresses mobility, nutrition, comfort, and cardiovascular risks.

Does HGPS affect the brain or intelligence?

Intelligence and learning ability are usually not affected. A stroke can injure the brain and cause new neurological problems, but this is a complication of damaged blood vessels rather than a typical developmental effect of HGPS itself.

Can someone develop progeria as an adult?

Classic HGPS begins in early childhood, even though the signs may not be visible at birth. It does not suddenly begin in an otherwise unaffected adult. Other genetic conditions can cause aging-like features later in life and require separate testing.

Is progeria the same as normal aging?

No. HGPS shares some features with aging, particularly artery, skin, hair, and bone changes. However, it is caused by a specific genetic change and does not reproduce every part of ordinary aging. For example, intellectual decline is not a usual feature.

How long can someone with HGPS live?

HGPS substantially shortens life, most often because of a heart attack, stroke, or related cardiovascular complication. Exact survival cannot be predicted for one person. Treatment with lonafarnib and specialist care may extend life, and outcomes continue to be studied.

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