Hutchinson-Gilford progeria syndrome

Hutchinson-Gilford progeria syndrome is a genetic condition characterized by the dramatic, rapid appearance of aging beginning in childhood. Affected children typically look normal at birth and in early infancy, but then grow more slowly than other children and do not gain weight at the expected rate (failure to thrive). They develop a characteristic facial appearance including prominent eyes, a thin nose with a beaked tip, thin lips, a small chin, and protruding ears. Hutchinson-Gilford progeria syndrome also causes hair loss (alopecia), aged-looking skin, joint abnormalities, and a loss of fat under the skin (subcutaneous fat). This condition does not affect intellectual development or the development of motor skills such as sitting, standing, and walking.

People with Hutchinson-Gilford progeria syndrome experience severe hardening of the arteries (arteriosclerosis) beginning in childhood. This condition greatly increases the chances of having a heart attack or stroke at a young age. These serious complications can worsen over time and are life-threatening for affected individuals.

Frequency

This condition is very rare; it is reported to occur in 1 in 4 million newborns worldwide. More than 130 cases have been reported in the scientific literature since the condition was first described in 1886.

Causes

Mutations in the *LMNA* gene cause Hutchinson-Gilford progeria syndrome. The *LMNA* gene provides instructions for making a protein called lamin A. This protein plays an important role in determining the shape of the nucleus within cells. It is an essential scaffolding (supporting) component of the nuclear envelope, which is the membrane that surrounds the nucleus. Mutations that cause Hutchinson-Gilford progeria syndrome result in the production of an abnormal version of the lamin A protein. The altered protein makes the nuclear envelope unstable and progressively damages the nucleus, making cells more likely to die prematurely. Researchers are working to determine how these changes lead to the characteristic features of Hutchinson-Gilford progeria syndrome.

Inheritance Pattern

Hutchinson-Gilford progeria syndrome is considered an autosomal dominant condition, which means one copy of the altered gene in each cell is sufficient to cause the disorder. The condition results from new mutations in the *LMNA* gene, and almost always occurs in people with no history of the disorder in their family.

Other Names for This Condition

- HGPS
- Hutchinson-Gilford syndrome
- progeria
- progeria of childhood

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Hutchinson-Gilford progeria syndrome, atypical
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C4016241/
- Genetic Testing Registry: Hutchinson-Gilford progeria syndrome, childhood-onset
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750285/
- Genetic Testing Registry: Hutchinson-Gilford syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0033300/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22hutchinson-gilford+progeria+syndrome%22

Other Diagnosis and Management Resources

- GeneReview: Hutchinson-Gilford Progeria Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1121
- MedlinePlus Encyclopedia: Progeria
  https://medlineplus.gov/ency/article/001657.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Progeria
  https://medlineplus.gov/ency/article/001657.htm
- Health Topic: Atherosclerosis
  https://medlineplus.gov/atherosclerosis.html
- Health Topic: Metabolic Disorders
  https://medlineplus.gov/metabolicdisorders.html

Genetic and Rare Diseases Information Center

- Progeria
  https://rarediseases.info.nih.gov/diseases/7467/progeria

Additional NIH Resources

- National Human Genome Research Institute
  https://www.genome.gov/Genetic-Disorders/Progeria

Educational Resources

- MalaCards: hutchinson-gilford progeria syndrome
  https://www.malacards.org/card/hutchinson_gilford_progeria_syndrome
- Merck Manual Professional Version
  https://www.merckmanuals.com/professional/pediatrics/miscellaneous-disorders-in-infants-and-children/progeria
- Orphanet: Hutchinson-Gilford progeria syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=740

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/hutchinson-gilford-progeria/
- Progeria Research Foundation, Inc.
  https://www.progeriaresearch.org
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/progeria.html

Clinical Information from GeneReviews

- Hutchinson-Gilford Progeria Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1121

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Progeria%5BMAJR%5D%29+AND+%28Hutchinson-Gilford%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- HUTCHINSON-GILFORD PROGERIA SYNDROME
  http://omim.org/entry/176670

Medical Genetics Database from MedGen

- Hutchinson-Gilford progeria syndrome, childhood-onset
  https://www.ncbi.nlm.nih.gov/medgen/442435
- Hutchinson-Gilford syndrome
  https://www.ncbi.nlm.nih.gov/medgen/46123

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