



HUTCHINSON-GILFORD PROGERIA SYNDROME FREQUENTLY ASKED QUESTIONS

WHAT IS PROGERIA?

Hutchinson-Gilford Progeria Syndrome (“Progeria”, or “HGPS”) is a rare, fatal genetic condition characterized by an appearance of accelerated aging in children. Its name is derived from the Greek and means "prematurely old." While there are different forms of Progeria*, the classic type is Hutchinson-Gilford Progeria Syndrome, which was named after the doctors who first described it in England; in 1886 by Dr. Jonathan Hutchinson and in 1897 by Dr. Hastings Gilford.

HOW COMMON IS PROGERIA?

Progeria has a reported incidence of about 1 in 4 - 8 million newborns. It affects both sexes equally and all races. In the past 15 years, children with Progeria have been reported all over the world, including in Algeria, Argentina, Australia, Austria, Canada, China, Cuba, England, France, Germany, Israel, Italy, Mexico, the Netherlands, Poland, Puerto Rico, South Africa, South America, South Korea, Switzerland, Turkey, the US, Venezuela, Vietnam and Yugoslavia.

WHAT ARE THE FEATURES OF PROGERIA?

Although they are born looking healthy, children with Progeria begin to display many characteristics of accelerated aging at around 18-24 months of age. Progeria signs include growth failure, loss of body fat and hair, aged-looking skin, stiffness of joints, hip dislocation, generalized atherosclerosis, cardiovascular (heart) disease and stroke. The children have a remarkably similar appearance, despite differing ethnic backgrounds. **Children with Progeria die of atherosclerosis (heart disease) at an average age of thirteen years** (with a range of about 8 – 21 years).

WHAT DOES PROGERIA HAVE TO DO WITH AGING?

Children with Progeria are genetically predisposed to premature, progressive heart disease. Death occurs almost exclusively due to widespread **heart disease, the leading cause of death worldwide**.⁺ As with any person suffering from heart disease, the common events for Progeria children are high blood pressure, strokes, angina (chest pain due to poor blood flow to the heart itself), enlarged heart, and heart failure, all conditions associated with aging.

Thus, there is clearly a tremendous need for research in Progeria. **Finding a cure for Progeria will not only help these children, but may provide keys for treating millions of adults with heart disease and stroke associated with the natural aging process.**

* Other progeroid syndromes include Werner’s syndrome, also known as “adult progeria” which does not have an onset until the late teen years, with a life span into the 40’s and 50’s.

⁺ American Heart Association, Journal Report: Sept. 24,2002

WHAT IS THE CAUSE OF PROGERIA?

HGPS is caused by a mutation in the gene called LMNA (pronounced, lamin – a). The LMNA gene produces the Lamin A protein, which is the structural scaffolding that holds the nucleus of a cell together. Researchers now believe that the defective Lamin A protein makes the nucleus unstable. That cellular instability appears to lead to the process of premature aging in Progeria.

PRF was the driving force behind finding the gene responsible for Progeria. A group of leading scientists from The Progeria Research Foundation's Genetics Consortium was able to isolate the Progeria gene in October 2002, and in April 2003, PRF led the announcement that Progeria is caused by a mutation of the gene LMNA, or Lamin A. This gene discovery was reported in the leading scientific journal *Nature*[♦].

The Progeria gene finding involved intensive collaboration between scientists including Dr. Leslie Gordon, PRF's Medical Director, Dr. W. Ted Brown, a world expert on Progeria and Chairman of New York's Institute of Basic Research in Developmental Disabilities' Department of Human Genetics, Dr. Tom Glover, a PRF grantee and Professor at University of Michigan's Department of Human Genetics, Dr. Francis Collins, Director of the National Human Genome Research Institute (responsible for mapping the human genome) and the senior author on the report, and first author Dr. Maria Eriksson, a postdoctoral fellow with Dr. Collins.

“Isolating the Progeria gene is a major achievement for the medical research community,” said Dr. Collins, “The discovery not only gives hope to children and families affected by Progeria, but also may shed light on the phenomenon of aging and cardiovascular disease.”

IS PROGERIA PASSED DOWN FROM PARENT TO CHILD? NO

Because neither parent carries or expresses the mutation, each case is believed to represent a sporadic (chance), new mutation that happens most probably in a single sperm or egg immediately prior to the time of conception. In other words, Hutchinson-Gilford Progeria Syndrome does not run in families.

There are other progeroid syndromes that may be passed down through families, but the classic HGPS is not. Therefore, genetic testing is crucial for accurate diagnosis of HGPS, so that children can receive proper care.

HOW IS PROGERIA DIAGNOSED?

Now that the gene mutation has been identified, **The Progeria Research Foundation has created a Diagnostics Testing Program.** We can now look at the specific genetic change, or mutation, in the Progeria gene that leads to HGPS. After an initial clinical evaluation (looking at the child's appearance and medical records), a sample of the child's blood will be tested for the Progeria gene. *For the first time ever, there is a definitive, scientific way to diagnose the children.* This will lead to more accurate and earlier diagnoses so that the children can receive proper care.

[♦] “Recurrent de novo point mutations in lamin A cause Hutchinson-Gilford Progeria Syndrome”, Vol. 423, May 15, 2003.

WHAT IS PRF DOING TO HELP CHILDREN WITH PROGERIA?

The Progeria Research Foundation **funds medical research** aimed at developing treatments and a cure for Progeria. PRF also has its own Cell & Tissue Bank that provides the biological materials researchers need to conduct their experiments. **The PRF Cell & Tissue Bank** was instrumental in the recent discovery of the Progeria gene. *Cell lines from the PRF Cell Bank were essential to the experiments that led to the Progeria gene discovery. These same cells and tissues will be essential to finding treatments and a cure for Progeria.* Additionally, PRF has established a **Medical & Research Database** to supply physicians and families with medical recommendations for cardiac care, nutrition and other medical issues to help the children have a better quality of life. We continue to analyze medical records of children with Progeria so that we can provide information on how best to medically help children with Progeria, and provide clues towards potential new treatments.

WHAT CAN YOU DO TO HELP CHILDREN WITH PROGERIA?

- *Make a financial contribution.* Donations are needed to continue the vital work of PRF. No donation is too little or too big – every dollar counts in our fight for a cure!
- *Donate your time.* Volunteers are also important to PRF's success. Hold a special event like a bake sale or letter writing campaign; translate documents for the families; help with a mailing – we'll find something for you to do that fits your schedule, location and talents!
- *Donate in-kind services or items.* Do you own a printing or office supply business? Do you have a background in non-profit development? These are just some of the many types of talents and connections PRF needs. The more tasks we can get accomplished on a pro bono basis, the more we can spend on research!
- *Spread the word and tap into your connections.* Do you know anyone who can do any of the above? Tell them about PRF and the amazing work we're doing! Many times, people are more likely to donate to an organization if they've gotten a recommendation from someone they know. So put in a good word for us and tell your friends, family and business colleagues to visit our web site at www.progeriaresearch.org, and contact us at info@progeriaresearch.org



The Progeria Research Foundation, Inc.

Together we *WILL* find a cure!