



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 affects approximately 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	Cuba	Ireland	Peru	Sweden
Argentina	Denmark	Israel	Philippines	Switzerland
Australia	Dominican Republic	Italy	Poland	Turkey
Austria	Egypt	Japan	Portugal	United States
Belgium	England	Libya	Puerto Rico	Venezuela
Brazil	France	Mexico	Romania	Vietnam
Canada	Germany	Morocco	South Africa	Yugoslavia
China	India	Netherlands	South Korea	
Columbia		Pakistan	Spain	



WHAT ARE THE FEATURES OF PROGERIA?

Although they are born looking healthy, most children with Progeria begin to display many characteristics of Progeria within the first year of life. 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.

* 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.

♦ For a map of where living children reside, please go to www.progeriaresearch.org/meet_the_kids.html

[†] The World Health Organization Fact Sheet No. 310, “The Top Ten Causes of Death” (November 2008 update)

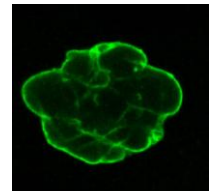
Thus there is clearly a tremendous need for research in Progeria. **Finding a cure for Progeria will not only help these children, but may provide keys for treating millions of adults with heart disease and stroke associated with the natural aging process.**

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 leads 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 PRF’s Genetics Consortium was able to isolate the Progeria gene, and in April 2003, PRF led the announcement reported in the leading scientific journal *Nature*[❖].

“Isolating the Progeria gene is a major achievement for the medical research community,” said Dr. Francis Collins, Director of the National Institutes of Health and co-discoverer of the Progeria gene, *“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.”*



Progeria cell nucleus

IS PROGERIA PASSED DOWN FROM PARENT TO CHILD?

HGPS is not usually passed down in families. The gene change is almost always a chance occurrence that is extremely rare. Children with other types of “progeroid” syndromes which are not HGPS may have diseases that are passed down in families. However, HGPS is a “sporadic autosomal dominant” mutation – sporadic because it is a new change in that family, and dominant because only one copy of the gene needs to be changed in order to have the syndrome. For parents who have never had a child with Progeria, the chances of having a child with Progeria are 1 in 4 – 8 million. But for parents who have already had a child with Progeria, the chances of it happening again to those parents is much higher – about 2-3%. Why the increase? This is due to a condition called “mosaicism”, where a parent has the genetic mutation for Progeria in a small proportion of their cells, but does not have Progeria. Prenatal testing is available to look for the LMNA genetic change.

HOW IS PROGERIA DIAGNOSED?

Now that the gene mutation has been identified, The Progeria Research Foundation has 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. ***Now there is a definitive, scientific way to diagnose the children.*** This leads to more accurate and earlier diagnoses so the children can receive proper care.

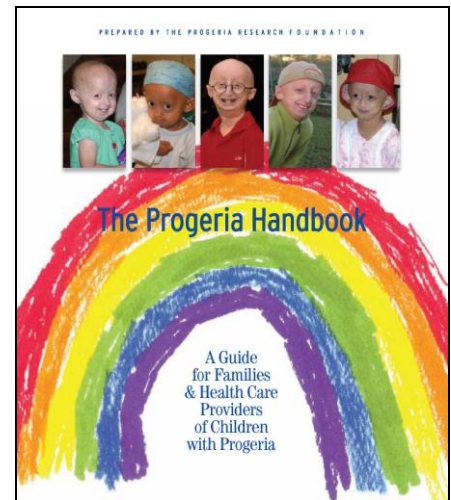
WHAT IS PRF DOING TO HELP CHILDREN WITH PROGERIA?

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. Additionally, PRF has established a **Medical & Research Database** to supply physicians and families with medical recommendations for cardiac care,

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

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. In April 2010, PRF published **The Progeria Handbook**, for families and doctors. From basic health facts to daily care recommendations to extensive treatment guidelines, the handbook helps answer many questions for children with Progeria throughout the world.

PRF is also involved in **Progeria clinical drug trials**, testing potential treatments. To date, PRF has funded and co-coordinated three clinical trials: the first began in May 2007 and all patients have completed their visits as of December 2009; the second was a one-month trial in March 2009 to determine if we could move forward with a larger population, which we did and that newest trial began August 2009. Please refer to www.progeriaresearch.org/clinical_trial for more details.



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 quest for a cure!
- ***Donate your time.*** Volunteers are also important to PRF’s success. Hold a special event like a bake sale or road race; 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? Are you a computer whiz that can help with our Twitter, FaceBook, web site and/or other on-line presence? Do you have a business 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 money we can devote to research!
- ***Spread the word and tap into your connections.*** Do you know anyone who can help? Tell your friends, family and colleagues 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 everyone to visit our web site at www.progeriaresearch.org, and contact us at info@progeriaresearch.org