The Progeria Research Foundation, Inc.

P. O. Box 3453
Peabody, MA 01961-3453
Tel: (978) 535-2594
Fax: (978) 535-5849
Email: progeria@netzero.net
www.progeriaresearch.org

Embargoed For Release 2 pm Eastern Time, Wednesday, April 16, 2003

Contact: Liza Morris
202-955-6222 x2524
lmorris@spectrumscience.com

Pamela Lippincott
202-955-6222 x2551
plippincott@spectrumscience.com

IDENTIFICATION OF GENE GIVES HOPE TO CHILDREN WITH PROGERIA; MAY SHED LIGHT ON PHENOMENON OF AGING

PRF Plays Key Role in Gene Discovery

[Boston, MA – April 16, 2003] – The Progeria Research Foundation (PRF), along with the National Institutes of Health (NIH), today announced the discovery of the gene that causes Hutchinson-Gilford Progeria Syndrome (HGPS or Progeria), a rare, fatal genetic condition characterized by an appearance of accelerated aging in children.

"Isolating the Progeria gene is a major achievement for the medical research community," said Francis Collins, MD, PhD, director, National Human Genome Research Institute and the senior author on the report, which appears today in *Nature*. "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."

Children with Progeria die from complications of cardiovascular disease or arteriosclerosis at an average age of 13. Researchers now believe finding the gene that causes Progeria may lead to answers surrounding the natural aging process and cardiovascular disease. Heart disease and stroke are the first and third leading causes of death in the United States, accounting for more than 40 percent of all deaths.

Within just one year of beginning the research, a group of leading scientists from the PRF Genetics Consortium were able to isolate the Progeria gene. Key aspects of the finding include the fact that Progeria is not inherited and that mutations to the gene LMNA (Lamin A) cause Progeria. The Lamin A protein is the structural scaffolding that holds the nucleus together and has been studied for years. 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.

-more-

In March 1999, Scott D. Berns, MD, MPH and Leslie Gordon, MD, PhD together with family, friends and colleagues formed the Progeria Research Foundation after their son Sam was diagnosed with Progeria at 21 months. They discovered a tremendous lack of medical information on Progeria and few research projects being conducted to find the cause, treatment or cure. Gordon, a physician-scientist, gave up her medical career path to devote her life to finding answers about Progeria.

Gordon is now the medical director of PRF. The non-profit organization is dedicated to helping families and children with Progeria through medical research. Gordon's sister, Audrey Gordon, Esq., serves as president and executive director of the foundation. PRF was the driving force behind the discovery of the gene.

"Members of PRF recruited all of the geneticists central to finding the gene," said Leslie Gordon, who was also a key investigator on the study. "In addition to the vision and commitment of the researchers, the blood donations from children with Progeria and their parents played an essential part in finding the gene."

In 2001, PRF began working in partnership with major institutes of the National Institutes of Health (NIH), including the National Institute on Aging and the Office of Rare Diseases, to co-host a joint workshop. PRF and NIH brought together leading scientists from around the world to identify promising areas of research in Progeria. This workshop led to funding for Progeria research and the formation of the PRF Genetics Consortium, a group of twenty scientists whose common goal was to find the genetic cause, treatment and cure for Progeria.

Furthermore, PRF will soon provide a diagnostic test for the disease where there was no test before. The finding offers new starting points for researchers and equips them with a significant foundation for future studies that may also help lead to treatment options. Currently, there is no treatment for Progeria.

About one in four to eight million newborns have Progeria and, although they are born looking healthy, they 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 disease and stroke.

The mission of PRF is to discover the cause, treatment and cure for Progeria through research and education. As the only organization in the world solely dedicated to finding treatments and a cure for Progeria, PRF has developed groundbreaking initiatives that seek to find explanations to the many unanswered questions surrounding this syndrome. PRF has funded basic science research aimed at discovering the biological basis of disease in Progeria, including finding the gene. They also launched a cell and tissue bank, developed a clinical and research database to supply physicians and families with medical recommendations, and created a website to make information accessible to all those interested. Find PRF at www.progeriaresearch.org.

###