Hutchinson-Gilford Progeria Syndrome: Phenotype and Course

Results Provide Insight into HGPS, General Aging Process

[Boston, MA – February 7, 2008] – A study published in the New England Journal of Medicine (NEJM) identified several clinical characteristics found in all children with the rare, premature aging syndrome known as Hutchinson-Gilford Progeria Syndrome (Progeria or HGPS). Detailed study results were published in today’s issue of NEJM.

Between February 2005 and March 2006, fifteen unrelated children with Progeria were enrolled in a comprehensive clinical protocol involving a number of assessments to help characterize the disease process. The study examined cardiovascular health, growth, nutrition, musculoskeletal, neurological, oral health, hearing, speech and language, eye and blood tests. The fifteen children, ages 1-17, who took part in the study represent a third of the worlds’ known HGPS population.

The study results confirmed five clinical characteristics consistent in children with Progeria, including sclerotic skin, joint contractures, bone abnormalities, alopecia and growth impairment. The study also uncovered previously unknown information about the onset of low frequency hearing loss, prolonged prothrombin times (clotting tendency of blood), elevated platelet counts and serum phosphorus levels, and abnormal range of motion in wrist, hip and ankle joints. In addition to providing information on Progeria, the study may offer additional information on the normal aging process that affects everyone.

“This study is significant,” said Leslie Gordon, MD, PhD, co-Investigator and co-author on the study, and Medical Director of The Progeria Research Foundation. “The more information we have about Progeria, the better armed we are to treat and eventually cure this disease.”

PRF is the only non-profit organization established to find the cause, treatment and cure for Progeria – a rapid aging disease that causes children to die from heart disease or stroke at an average age of 13. In just eight years, PRF has done a tremendous amount to help save these children, and in 2003, PRF accomplished the first part of its mission by identifying the gene that causes Progeria. Recent studies have also uncovered a drug that may treat Progeria, and PRF is now working to raise funds for a first-ever clinical drug trial at Children’s Hospital Boston, which began in May 2007. Please visit www.ProgeriaResearch.org for more information.

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