The Progeria Research Foundation

Diagnostic Testing Program

Dear Parent,

After an intense scientific search, the gene for Hutchinson-Gilford Progeria Syndrome (HGPS) was found in April 2003 by a group of researchers working together through The Progeria Research Foundation (PRF) Genetics Consortium. Among them were PRF Medical Director and prominent researchers from institutions throughout the United States, including the National Institutes of Health.

This information sheet is intended to answer the most common questions about the gene finding, the diagnostic test, and what this means for children with HGPS.

What is the Gene for HGPS?

The gene responsible for HGPS is called LMNA (pronounced "lamin-a"). Within this gene there is a change in one element of DNA that is responsible for HGPS. This type of gene change is called a point mutation. The LMNA gene makes a protein called Lamin A, which is an important protein for most cells of our bodies. Lamin A is found in the cell nucleus, and helps maintain the integrity of the cell. The point mutation compromises that cell integrity, causing HGPS.

Is This Disease Passed Down?

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. This means that, for a family with one child with HGPS, non-twin siblings have the same chance of having HGPS as any other child in any other family – approximately one in 4-8 million. In very rare instances, about once every 100 cases of HGPS (or frequency of about 1 in 400 million births), HGPS can be passed down within a family.

What is the test all about?

The Progeria Research Foundation is pleased to be able to provide a genetic test for Progeria through The PRF Diagnostic Testing Program. Previously we could only diagnose HGPS using clinical information such as an overall look and X-rays. Now we have a genetic test that can more definitively identify HGPS.

How do I get this test done?

The first step is for our medical director to look at the child's clinical history. Then, if HGPS is a possible diagnosis, we will be in touch with you and your physicians about having this blood test done. The test is of no cost to you or your physicians. *We uphold the highest standards of medical care, so that all information is kept strictly confidential.*

For further assistance, questions or problems, please contact **Dr. Leslie Gordon** at The Progeria Research Foundation at info@progeriaresearch.org, or call (978) 535-2594.

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