The PRF Diagnostic Testing Program Information Sheet

After an intense scientific search, the gene for Hutchinson-Gilford Progeria Syndrome (HGPS), or Progeria, 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, Leslie Gordon, MD, PhD, 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 normal 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 causes the LMNA gene to make an abnormal disease-causing protein called progerin. Progerin compromises 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. For parents who have never had a child with HGPS, the chances of having a child with Progeria are 1 in 4 – 8 million.

Rarely, HGPS can be passed down from a parent. This is due to a condition called "mosaicism", where a parent has multiple egg or sperm cells (or other cell types) with the HGPS mutation, but does not exhibit the HGPS disease characteristics. To date, we know of two families in which this has occurred. Therefore, for parents who have already had a child with Progeria, the chances of it happening again to those parents could be about 3%. Prenatal testing is available to look for the *LMNA* genetic change.

The Progeria Research Foundation Genetic Testing Program

The Progeria Research Foundation is pleased to be able to provide a genetic test for Progeria through The PRF Diagnostic Testing Program. This test looks at the DNA that we get from a blood sample. The test results, along with the clinical information, tell us whether a child has Progeria.

How do I get this test done?

The first step is for our medical director to look at the clinical history of the child. If Progeria is a possible diagnosis, we will be in touch with the parents, legal guardians and/or physician of the participating child about having this blood test done.

- PRF pays for genetic testing
- The test is performed at a qualified clinical laboratory
- The final test report will be sent to the child's physician who will explain the results to the participant's parents or legal guardians.

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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