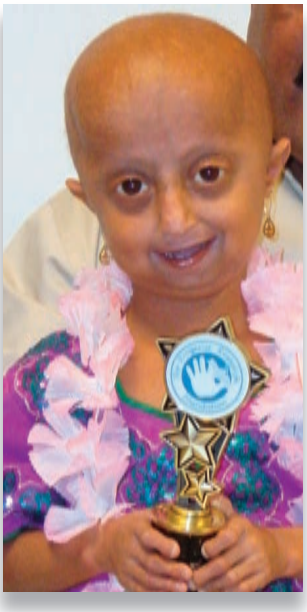
20. PRF Programs and Services

International Patient Registry
Diagnostic Testing Program
Medical & Research Database
The Weighing-in Program
Cell & Tissue Bank
Progeria Family Network
Research funding
Scientific workshops
Public awareness
Volunteers & fundraising



The Progeria Research Foundation (www.progeriaresearch.org) provides services for families and children with Progeria such as patient education and communication with other Progeria families. It serves as a resource for physicians and medical caretakers of these families via clinical care recommendations, a diagnostics facility, and a clinical and research database. It also provides funding for basic science and clinical research in Progeria and biological materials for the research, and brings researchers and clinicians together at scientific conferences.

This section describes the many programs and resources available through The Progeria Research Foundation.

The courage of children and families participating in PRF programs is the key to new discoveries and progress in the field of Progeria.

International Patient Registry

Progeria is a very rare condition. PRF's International Patient Registry has been established to provide services and information to families of children with Progeria, treating physicians, and researchers, and to better understand the nature and natural course of Progeria. Entering a child with Progeria into the Registry serves to improve communication of ideas among interested researchers, and assures rapid distribution of any new information that may benefit patients and/or their families.

Visit www.progeriaresearch.org/patient_registry.html for more information.

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Diagnostic Testing Program

The PRF Diagnostic Testing Program offers genetic testing for children with Progeria, provided at no cost to families. In previous years, with so little information available on Progeria, families often suffered for months or even years in fear and frustration as they tried to get an accurate diagnosis and appropriate medical treatments for their child. A genetic test means earlier diagnosis, fewer misdiagnoses and early medical intervention to ensure a better quality of life for the children.

The first step is for our medical director to look at a child's clinical history and photographs. Then we will be in touch with the family and home physicians about having this blood test done. All personal information is kept strictly confidential.

We provide genetic sequence testing by a CLIA-approved* laboratory for either Exon 11 of the LMNA gene (only the portion of the gene where the classical HGPS mutation is found) or full LMNA gene sequencing (for atypical types of Progeria called progeroid laminopathies).

Visit www.progeriaresearch.org/diagnostic_testing.html for information.

Medical & Research Database

The PRF Medical & Research Database is a collection of medical records and radiological tests such as X-rays, MRIs, and CTs from children with Progeria from all over the world. The data is rigorously analyzed to determine the best course of treatments to improve the quality of life. Analysis of these medical records has provided new insights into the nature of Progeria and into the nature of other diseases such as heart disease, which in turn will serve to stimulate the advancement of new research projects. The information is invaluable for the health care provider and families. PRF has used the information to provide new analyses of Progeria to the medical and research worlds. Our medical care recommendation sheets and this care handbook are products of the PRF Medical & Research Database.

PRF is privileged to work with top quality academic centers on the PRF Medical & Research Database: Brown University Center for Gerontology & Health Care Research and Rhode Island Hospital.

The highest level of confidentiality is maintained in this and all PRF programs. The PRF Medical & Research Database is approved by the Institutional Review Boards at Brown University and Rhode Island Hospital.

Visit www.progeriaresearch.org/medical_database.html for information.

The Weighing-in Program

Each child with Progeria has a consistent and slow weight gain. We have used this data to track baseline weight gain, and potentially to track improvements with treatment. We are using rate of weight gain as a substitute marker for general health, since we can easily and reliably track weight over time. When families participate in the weighing-in program, we send families a scale, log book, and instructions so that they can report weekly weights directly to PRF. This is part of the PRF Medical & Research Database program and consent is required to participate.

Rate of weight gain has been used to decide whether the treatment trial drugs are having a beneficial effect on the children who participate in the trials. To do this, pre-drug weights should be followed carefully for about 6 to 12 months or longer if the child is very young, since weight gain does not become reliable until about the age of 3 years in Progeria.

Visit www.progeriaresearch.org/medical_database.html for information.

Cell & Tissue Bank

The PRF Cell & Tissue Bank provides medical researchers with genetic and biological material from Progeria patients and their families, so that research on Progeria and other aging-related diseases can be performed to bring us closer to finding the cure. Thanks to the participation of courageous children and their families, PRF provides over 100 cell lines and tissues from affected children and their immediate relatives. This includes cells from blood, skin biopsies, teeth, hair, autopsy tissue and more. These essential research tools are provided worldwide. This helps assure not only that research into Progeria is maximized, but that children do not have to be asked to donate blood and skin biopsies multiple times. Researchers can simply apply to the PRF Cell & Tissue Bank for the biological materials they need to ask key questions about Progeria.

PRF is privileged to work with top quality academic centers and collaborators on the PRF Cell & Tissue Bank: Rhode Island Hospital, Brown University, and Rutgers University Cell & DNA Repository.

The highest level of confidentiality is maintained in this and all PRF programs. We remove names and all other identifying information and code all samples. The PRF Cell & Tissue Bank is approved by the Institutional Review Board of Rhode Island Hospital.

Visit www.progeriaresearch.org/cell_tissue_bank.html for information.

*Clinical Laboratory Improvement Amendments (CLIA) is a body of industry regulations ensuring quality laboratory testing.

Progeria Family Network

Because Progeria is so rare, it is unlikely that families will be located close to one another. Yet, it is essential that families share feelings and advice, and give each other emotional support. To help families connect, PRF has created a private message board website. This on-line tool helps the families get to know each other, and develop a support network of people with whom they can share concerns and ideas on how best to care for their children. PRF also provides contact information to families privately, so that they can exchange emails, phone calls, and even meet in person.

Visit www.progeriaresearch.org/chat_room.html for information.

Research funding

PRF's grants of up to $100,000 over two years, have allowed innovative new research in Progeria to thrive through research projects performed throughout the USA and the world. Proposals are carefully evaluated by PRF's Medical Research Committee and Board of Directors. PRF solicits proposals worldwide in a continuing effort to encourage researchers to work in this intriguing and ever-growing field.

> The PRF Medical Research Committee:

Bryan P. Toole, PhD, Chair
Professor of Cell Biology and Anatomy,
Medical University of South Carolina

W. Ted Brown, MD, PhD
Director; New York State Institute for Basic Research
in Developmental Disabilities

Judith A. Campisi, PhD
Senior Staff Scientist, Lawrence Berkeley National Laboratory

Thomas Glover, PhD
Professor of Human Genetics, University of Michigan

Leslie Gordon, MD, PhD
Medical Director, The Progeria Research Foundation
Associate Professor of Pediatric Research,
Alpert Medical School of Brown University

Christine Harling-Berg, PhD
Assistant Professor of Pediatrics,
Alpert Medical School of Brown University
Memorial Hospital of Rhode Island

Monica Kleinman, MD
Clinical Director, Medical-Surgical ICU, Children's Hospital Boston

Paul Knopf, PhD (Retired 2009)
Professor of Medical Science, Emeritus, Brown University

Frank Rothman, PhD
Professor of Biology and Provost, Emeritus, Brown University

Scientific workshops

PRF organizes successful scientific conferences every two years. These meetings have brought together scientists and clinicians from all over the world to collaborate, sharing ideas and contributing their expertise in this lethal disease. The workshops are a cornerstone of inspiration for those in the scientific and medical communities who seek to understand Progeria and its relationship to aging and heart disease, and search for treatments and cure. Many generous organizations have co-sponsored these meetings, including the National Institutes of Health's Office of Rare Diseases; National Heart, Lung, and Blood Institute; National Cancer Institute; National Human Genome Research Institute; and National Institute on Aging, The Ellison Medical Foundation, Celgene Corporation, The Max and Victoria Dreyfus Foundation, and the American Federation on Aging Research.

Visit www.progeriaresearch.org/scientific_meetings.html for information.

Public awareness

Before PRF was formed, Progeria was virtually unknown to the general public and to many healthcare workers. Information about Progeria and our far-reaching message – that finding a cure may help those with heart disease and other aging-related conditions – has reached millions through PRF's web site, newsletters, educational materials, and the media. PRF's story has appeared on CNN, BBC, "Primetime", "Dateline", "Discovery", in *Time* and *People* magazines, *The New York Times*, *The Wall Street Journal*, and dozens of other widely-read media outlets. As awareness continues to spread throughout the world, more children come to PRF for diagnostic testing; more researchers apply to PRF for grant funding and cells to support their research; more scientists participate in PRF's scientific workshops; and more volunteers offer needed support.

Visit www.progeriaresearch.org/press_room.html for information.

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Volunteers & fundraising

PRF relies on its chapters and other volunteers to help spread the word and raise funds for medical research. With the exception of the small staff, everyone involved with PRF, including its Board of Directors, committee members, and corporate officers generously give their time, energy, and talents to PRF for free so that we can spend less on administrative costs and more on raising awareness and finding a cure for Hutchinson-Gilford Progeria Syndrome.

Please visit www.progeriaresearch.org/get_involved.html to find out how you can be part of PRF's efforts.