

Day one-presentation-session IV
Leslie Gordon

**Advancing Rare Disease Research:
The Intersection of Patient Registries, Biospecimen Repositories and Clinical Data
Bethesda, Maryland
January 11,12, 2010**

Abstract

**Essential Elements for Translational Research in Rare Diseases:
Progeria as a case study**

Leslie B. Gordon, MD, PhD

The Progeria Research Foundation; Warren Alpert Medical School of Brown University, Hasbro Children's Hospital, Harvard University, Children's Hospital Boston

Progeria is a rare, fatal, "premature aging" syndrome that afflicts children globally, who die of heart disease at an average age of 13 years. There are only 200 children living with Progeria in the world at any one time. In 1999, in the wake of her son's diagnosis of Progeria, Dr. Leslie Gordon co-founded The Progeria Research Foundation (PRF). Its mission is to discover the cause, treatment and cure for Progeria and its aging related disorders. Since that time, PRF has become a driving force behind the tremendous and rapid progress in the field of Progeria research. Within just 10 years, Progeria has gone from almost complete obscurity, to gene finding (2003) to Progeria drug treatment trial (2007).

Utilizing PRF as a case study, this talk will highlight key elements and opportunities for rare disease organizations to catalyze their mission efforts by:

- Developing a clear mission and staying on mission
- Creating an organized infrastructure
- Finding and involving patients
- Creating programs that inspire trust and serve your mission, which include patients, families, providers, and researchers.
- Finding and involving collaborators, while maintaining independence
- Balancing Basic and Clinical research
- Making rare disease research relevant to common conditions
- Identifying and overcoming challenges



Uniting Rare Diseases

Advancing Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories and Clinical Data

Session IV

Patient Participation & Outreach Activities/Patient Advocacy

Leslie Gordon, MD, PhD

Medical Director, The Progeria Research Foundation

*Essential Elements for Translational Research in Rare
Diseases: Progeria as a Case Study*



What is Progeria?

- **Premature “Aging Disease”**
- **Rare –affects 1 in 4 million (10-12 cases in USA)**
- **Uniformly Fatal**
 - **Heart Attacks/Strokes**
 - **Average Lifespan 13 years**



16 y.o.

1998 - There Was No Hope...



- **No Research Funding, No Non-profit Driving Research**
- **Was this a genetic disease? We did not know.**
- **No Central Source of Clinical Information**
- **No Treatment Prospects**
- **No Place for Families and Physicians to go for help**



1999: Founded PRF

Mission

- Cause
- Treatment
- Cure



Clear Mission- Avoiding Mission Creep

The Needs: Scientists, Physicians, Families

International
Registry/Medical
Database: 82



Cell/Tissue Bank: 121

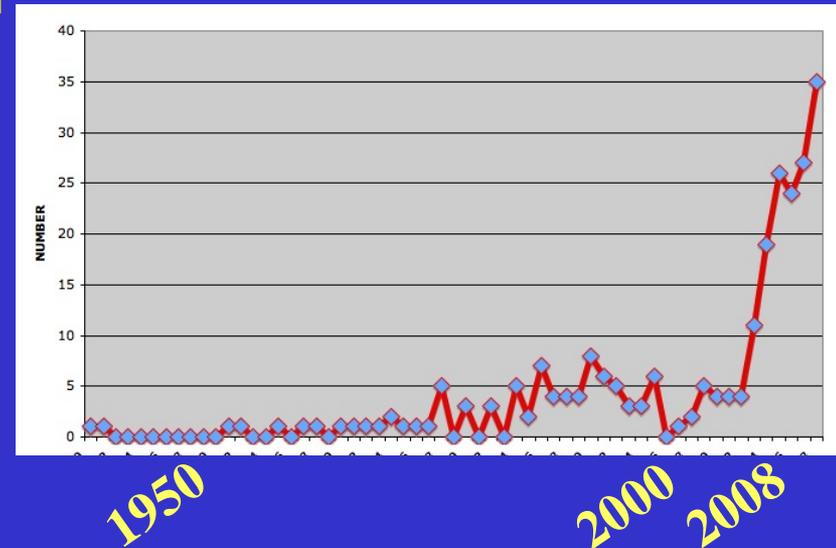


Research Funding: 27



Scientific Meetings: 5 (Apr. 2010)

HGPS Publications



Creating Programs That Inspire Trust and Involvement

International Progeria Registry

- **Online and Translated to Multiple Languages**
- **Basic contact, epidemiologic, and disease information (similar to a brief questionnaire)**
- **The first step in evaluating necessary assistance to the child/family/physician**
- **Prelude to diagnostics and database programs**

Finding and Involving Patients, Physicians

PRF Medical and Research Database (Detailed Registry) Design

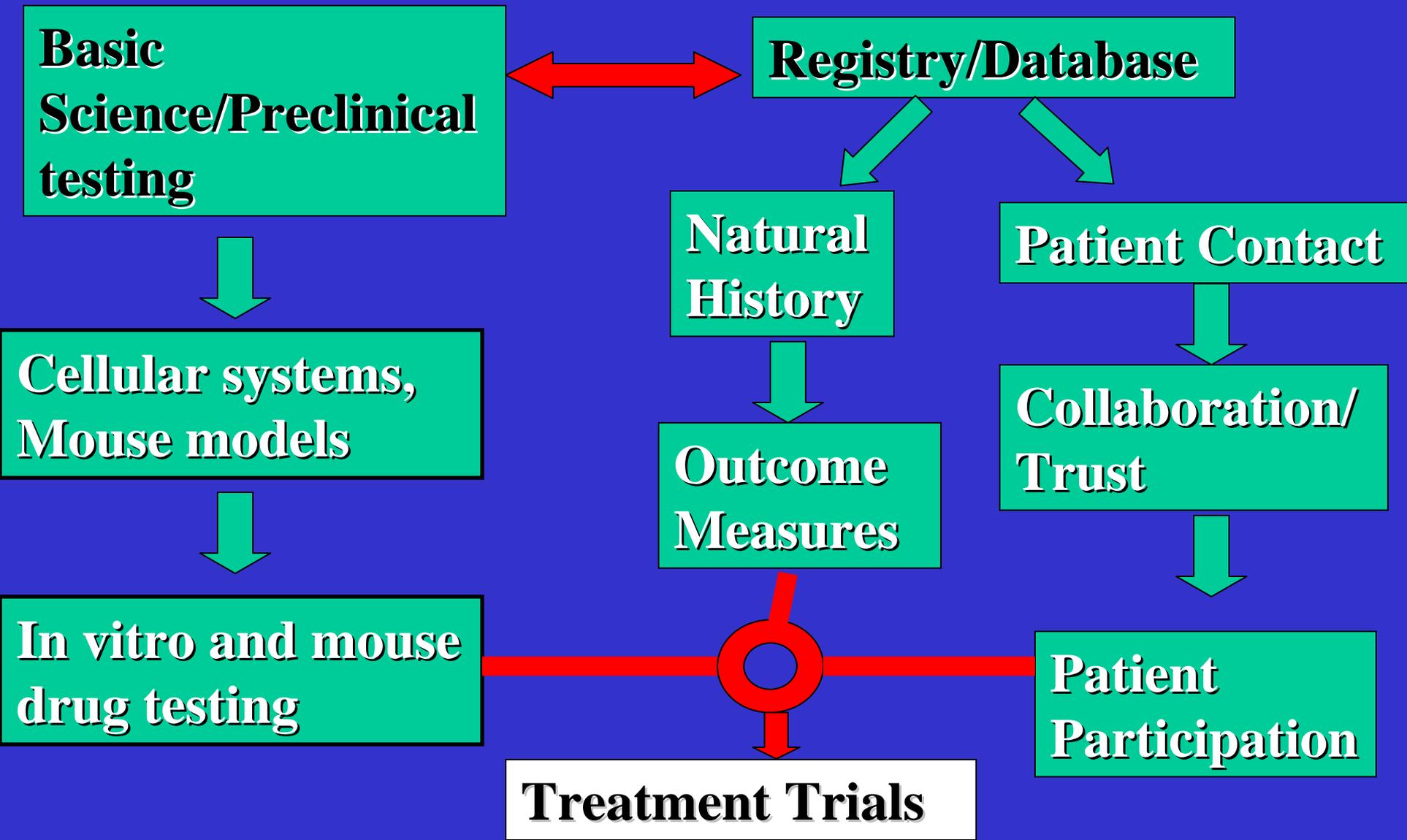


- **Collect all medical information, radiol. studies**
- **Abstract elements of interest – perpetual project**
- **Link to the PRF Cell and Tissue Bank specimens**

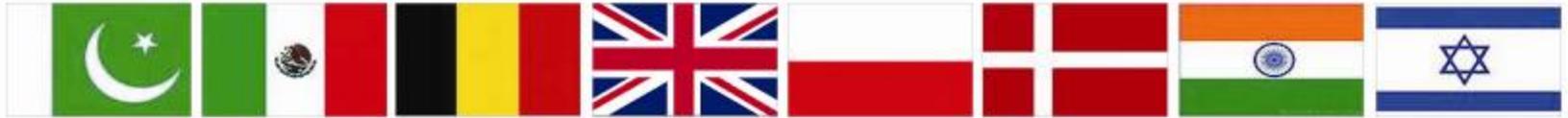
PRF Database Outcomes

- **Define/redefine the disease through publications**
 - Autopsy & neurovascular studies
- **Discovered disease marker that became the primary outcome parameter for the trials**
 - Weighing-in Program – retrospective to prospective
- **Provide records for clinical trials**
- **Recruitment at lightening speed**
 - All came through PRF
 - Stimulus Grant –
 - reviewers all commented on ability to recruit

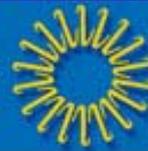
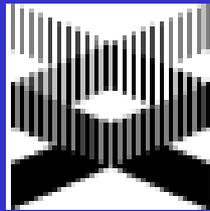
Our Journey Thus Far: A Marriage of Basic and Clinical Research



May, 2007 the First-ever Treatment Trial Begins

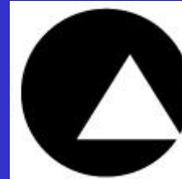


28 children from 16 countries now participating in a first-ever Progeria clinical drug trial



Rhode Island Hospital
A Lifespan Partner

Families



SPECTRUM SCIENCE
COMMUNICATIONS

Partnerships create powerful forces for change!



Children's Hospital Boston



BROWN



Chapters,
Individual Donors
and Volunteers



Schering-Plough

PreventionGenetics



*Private
Foundations &
Corporations*

Grass Roots Foundations are Essential to Rare Disease Progress



- **The link between patient community and scientists, physician-researchers**
- **Collaboration with ongoing influence on data access and utilization**
- **Programs are key to this link**

Thank you!

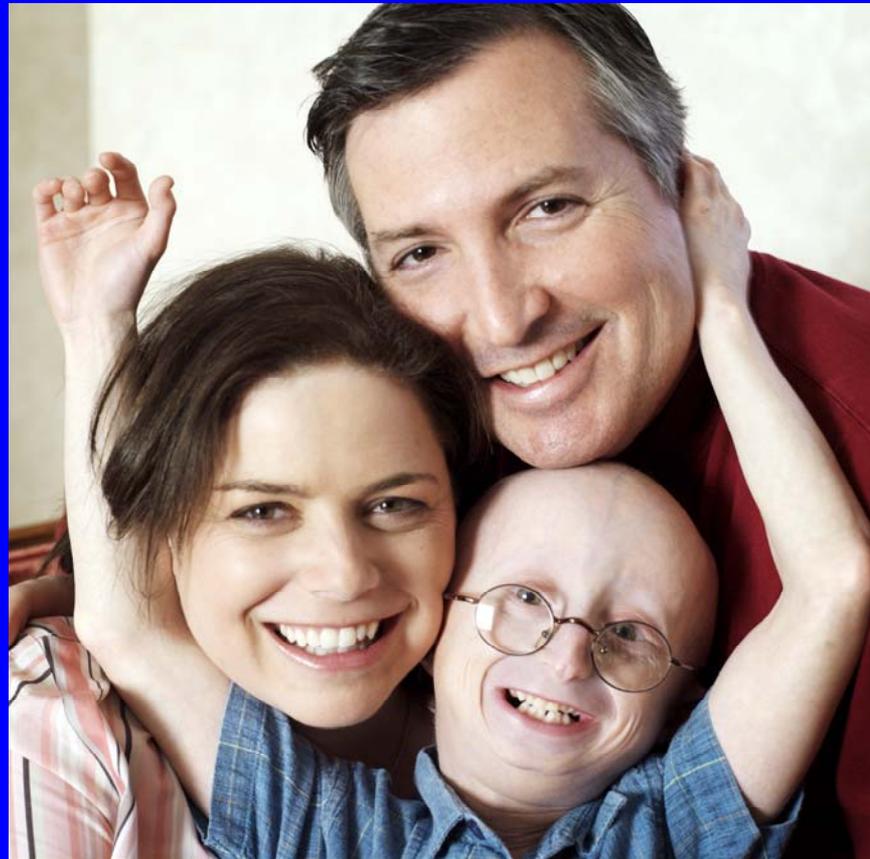
Susan Campbell

**Nancy
Grossman**

**Nancy Wolf-
Jensen**

Lorraine Fast

Audrey Gordon



**Yaffa
Rubenstein
Steve Groft
ORD staff**



Together, we WILL find the cure.

www.progeriaresearch.org