

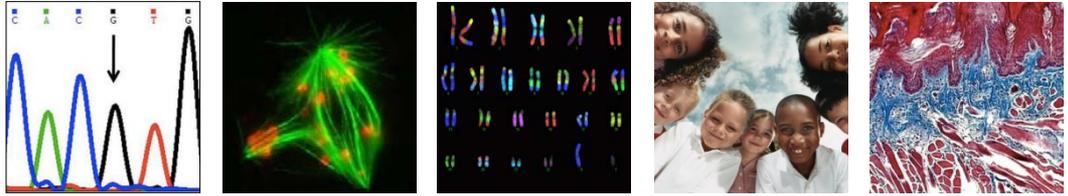
# Opportunities for Canadian collaboration

Paul Lasko

Scientific Director

CIHR Institute of Genetics

CIHR-IG outreach and education  
efforts in the area of rare disease



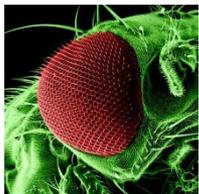
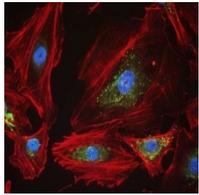
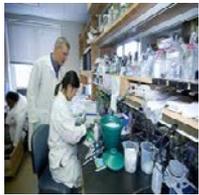
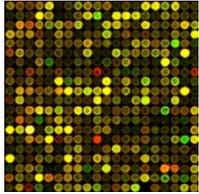
# orphanet

The Orphanet-Canada Project



Paul Lasko, Scientific Director, CIHR Institute of Genetics

# orpha.net – What it is



- Established in 1997, Orphanet is the official European reference portal for information on rare diseases and orphan drugs, for all audiences. Orphanet now has 37 participating countries.
- Orphanet offers a range of freely accessible services:
  - An inventory of rare diseases and a classification of diseases
  - An encyclopaedia of rare diseases in 6 languages (including English and French)
  - An inventory of orphan drugs
  - A directory of specialised services in each of the countries in Orphanet’s network (specialised clinics, medical laboratories, ongoing research projects, clinical trials, registries, networks, technological platforms and patient organisations)
  - An assistance-to-diagnosis tool allowing users to search by signs and symptoms.
  - An encyclopaedia of recommendations and guidelines for emergency medical care and anesthesia.



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### NEWS

Experts communicate  
Canada's current standing  
with rare diseases in the  
National Post 

New Biobank Certification  
Program! Learn how to  
certify a Canadian biobank.  


CIHR Cafés scientifiques on  
Canal Savoir 

The MNMWiki project is  
online! GRIMN is engaged  
with a knowledge-sharing  
project concerning  
neuromuscular diseases 

Subscribe to CIHR's SPOR  
Update 

### INTERNATIONAL NEWS

2nd International Congress  
on Research of Rare  
Diseases - RE(ACT)2014, 5-  
8 March 2014, Bâle,  
Switzerland 

ECRD 2014, 8-10 May 2014,  
Berlin, Germany 

## Orphanet International services

- o An inventory and classification of rare diseases
- o An encyclopaedia
- o A directory of services: clinics, laboratories, research projects, registries, clinical trials, patient organisations
- o An inventory of orphan drugs
- o Guidelines, reports
- o A newsletter

Access these services in:   

## Welcome to the Orphanet-Canada National Website!

This page presents the news, events and documents of national significance.

- To register your activity, please visit <http://orphanet-canada-data.ca/>.
- To access all information on rare diseases, orphan drugs and expert services, please visit the main Orphanet portal ([www.orpha.net](http://www.orpha.net)).

**November 19 2013, Canadian Institutes of Health Research  
Roundtable, Chicoutimi, Quebec**  
***Theme: Toward a coordinated approach in support of care  
for genetic orphan diseases***

Roundtable Co-Chairs:

Étienne Richer, CIHR IG

Catherine Laprise, Canada Research Chair on the Genetics of  
Asthma

Bernard Brais, Montreal Neurological Institute



The Saguenay–Lac-Saint-Jean (SLSJ) region is a pioneer in the development of organizations and specialized services that provide regional support for the most prevalent genetic diseases. In addition to the efforts of these individuals and organizations, CIHR Institute of Genetics recently declared the promotion of orphan disease research in Canada a priority. In keeping with this priority - and to build on the pioneering efforts of the SLSJ region - the Institute of Genetics has invited key players from the SLSJ region, as well as other provincial experts, to share their experiences and ideas about the development of coordinated strategies in support of the care of orphan diseases.

\*See the minutes from this roundtable in the Documents section.\*

Care for Rare: a national consortium aimed  
at diagnosing patients with unknown rare  
diseases and developing new rare disease  
therapies



# CARE for RARE

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## About CARE for RARE

Information about the objectives for CARE for RARE

## WELCOME TO THE CARE FOR RARE WEBSITE.

CARE for RARE is a pan-Canadian collaborative team of clinicians, bioinformaticians, scientists and researchers building upon the infrastructure and discoveries of the FORGE Canada (Finding of Rare Disease Genes) project. The goal of CARE for RARE is to improve clinical care for patients and families affected by rare diseases. This will be done by expanding and improving the diagnosis and treatment of rare diseases.

# Planned Care for Rare Education Activities

- Develop and deliver a “Genomics Technologies for Patient Care” program for clinical trainees (physicians and laboratory diagnostic specialists).
  - Collaboration with NowGen (Dr. Di Donnai, Executive Director, University of Manchester)

# C4R Genomics Technologies for Patient Care Program (under development)

- Series of telehealth lectures and a one-week laboratory-based session.
- Involves the Royal College of Physicians and Surgeons of Canada (RCPSC) Specialty Committee Chair and Examination Chair.
- Involves the Canadian College of Medical Genetics (CCMG) committees on training, clinical practice, laboratory practice, and ethics, education and policy.

# RCPSC Specialty Committee in Medical Genetics

- Administers TELEGRAF (Telelearning for Genetics Residents and Fellows), a monthly curriculum of lectures attended by all MD and PhD trainees in genetics across Canada.
- Administers bi-annual mock examinations in medical genetics.
- Plan is to share expertise with other branches of specialty medicine (internal medicine, neurology, pediatrics, etc.)

# Other linked people and activities

- June Carroll (Toronto) Canadian College of Family Physicians, approached about ISCC, interested in getting involved.
- Bob Roberts (Ottawa Heart) has been involved for some time with ISCC.