



Innovations in Research, Therapies and Policy for Rare Disorders

Innovations in research leading to effective therapies have been promoted by and also have provoked enabling policy that stimulates R&D initiatives, facilitates patient access, and supports appropriate real-world use and learning. Given the recently announced CIHR funding of research grants for rare diseases and Health Canada’s imminent promulgation of an orphan drug/rare diseases regulatory framework, this two-day conference is designed to enable understanding of the broad spectrum of issues and enhance collaboration among all stakeholders.

As we celebrate the 2nd official Rare Disease Day, we at the Canadian Organization for Rare Disorders marvel at how far we have come since the 1st Rare Disease Day in 2008, and we are wildly optimistic about the future for Canadians with Rare Disorders. Canada is undoubtedly at the Tipping Point for major contributions to rare diseases.

Those of you who followed our 1st Arctic Quest for Rare Diseases in August 2010 will know our slogan: “What can patients with rare disorders do? Anything, with the right care, treatment and support.”

Day 1: February 29, 2012

8:30 a.m. – 9:00 a.m.	Continental Breakfast Registration	
9:00 a.m. – 9:05 a.m.	Welcome and Purpose	Durhane Wong-Rieger
9:05 a.m. – 9:15 a.m.	CIHR Rare Diseases Emerging Teams Announcement (Dr. Colin Carrie, Parliamentary Secretary to Minister of Health)	Dr. Colin Carrie, Parliamentary Secretary to Minister of Health
9:15 a.m. – 10:45 a.m.	Developing Innovative Therapies for Rare Conditions (Part 1)	
	<ul style="list-style-type: none"> Challenges for Orphan Drug Development: How to Meet Mission Small Patient Populations and Clinical Trials: Example of Antitrypsin for Alpha 1 Antitrypsin Deficiency <i>Targeting (Rare) Lung Cancer with Genetic Diagnosis and Highly Specific Therapy</i> 	Dr. Arthur Tzianabos, Shire Therapeutics Dr. Ken Chapman, University of Toronto Dr. Glen Goss, U Ottawa, TBC
10:45 a.m. – 11:00 a.m.	Health Break	
11:00 a.m. – 12:30 p.m.	Developing Innovative Therapies for Rare Conditions (Part 2)	
	<ul style="list-style-type: none"> Therapies for Rare Diseases and Targeted Patients: Policy Challenges Multiple Receptor Tyrosine Kinases Working for Multiple Cancers (Kidney, GIST, Liver, Pancreatic NET) Autoimmune Disorders and Evolution of Treatment 	Dr. Stan Gleezer, sanofi Dr. Marjana Zaremski, Pfizer) Dr. Ron Laxer, Toronto Hospital Sick Kids (TBC)
12:30 p.m. – 1:30 p.m.	Lunch	

Innovating Policy from Research to Real World-Use (1:00 p.m. – 2:45 p.m.)

Since the 1983 US-FDA Orphan Drug Act, regulatory policies to support development and market access of therapies for rare conditions has been, for the most part, proactive and designed to accommodate characteristics of rare diseases: small numbers, poorly understood natural history, life-threatening or severely debilitating effects, reliance on surrogate markers, and lack of alternative treatments. In contrast, policies and review processes related to patient access and (public) funding have been, for the most part, unchanged from those governing access to “common” therapies and reactive to the exigencies of orphan drugs, whereby real-world, long-term effectiveness and safety may be considerably less certain and where per-patient cost may be considerably higher than for common conditions.

The purpose of the next panel is to consider appropriate policy for patient access and reimbursement of drugs for rare diseases. Internationally, there has been considerable discussion around the appropriate application of health technology assessment and/or “cost-effectiveness” models to innovative therapies, including rare diseases, end-of-life therapies, and emergent personalized medicine. In Canada, we have recently implemented specific HTA process for oncology drugs, the pan-Canadian Oncology Drug Review and in Ontario, the Drugs for Rare Diseases Program has been working for the past three years. Internationally and in Canada, there are various market access schemes that are being used for innovative and rare therapies, and we can learn from these. Given that Canada is an international leader in HTA and has a strong tradition in compassionate access to necessary therapies, we are in an ideal position to develop access strategies and funding models that meet needs of patients for life-saving therapies, of health systems for assuring appropriate and financially sustainable use, and of manufacturers for incentives to continued investment in innovative therapies.

1:30 p.m. – 3:15 p.m.	<p>Options for Sustainable Access to Rare Disease Drugs</p> <ul style="list-style-type: none"> • Recommendations from CORD Tipping Point Conference—Sep 2011 • Developing Effective Policies for Managing Technologies for Rare Diseases • Developing a Canadian framework for evaluation and decision-making for expensive drugs for rare diseases through innovation, value, and priority setting • Review of Drugs for Rare Diseases: Challenges and Emerging Solutions • Provincial Programs for Drugs for Rare Diseases: Lessons Learned and New Challenges 	<p>Dev Menon, Moderator</p> <p>D. Wong-Rieger, CORD Dev Menon & Tania Stafinski, U of Alberta</p> <p>Larry Lynd, UBC</p> <p>CADTH (TBC)</p> <p>Diane McArthur, Ontario Ministry of Health</p>
3:15 p.m. – 3:30 p.m.	<p>Day 1 Conclusions and Next Steps</p>	

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Canadian Rare Disease Research

In 2010, the Canadian Institutes for Health Research under the leadership of the Genetics Institute announced the first CIHR research fund competition for Emerging Teams on Rare Diseases. Funding for both basic biomedical research as well as health services has been awarded.

At Genome Canada, genetic research programs that relate to rare diseases have also been funded and supported.

This session provides an overview of some of the current and emerging research projects taking place in Canadian universities and institutions, many of which are comprised of national and international partnerships.

Day 2: March 1, 2012

8:00 a.m. – 8:30 a.m.	Continental Breakfast Registration	
8:45 a.m. – 9:00 a.m.	Welcome and Purpose	
9:30 a.m. – 10:15 a.m.	Overview of Rare Diseases Research Canadian Institute for Health Research	Paul Lasko, CIHR
	Clinical Trials for Small Patient Populations (TBC, Health Canada)	
9:30 a.m. – 10:15 a.m.	Regulatory Support for Rare Disorders Health Canada Regulatory Framework for Rare Disease (David Lee, Health Canada) Clinical Trials for Small Patient Populations (TBC, Health Canada)	David Lee, Health Canada TBC, Health Canada
10:15 a.m. – 10:30 a.m.	Health Break	
10:30 a.m. – 11:45 p.m.	Studying Genes and Finding Therapies <ul style="list-style-type: none"> • FORGE: Finding of Rare Disease Genes • Choroideremia: expanding our understanding, exploring treatments • Orphan Diseases: Identifying Genes and Novel Therapeutics to Enhance Treatment (IGNITE) • Application of next generation gene sequencing technology and analysis strategies: example of Dyskeratosis Congenita 	Kym Boycott, CHEO) Dr. Ian MacDonald, University of Alberta) Dr. Karen Bedard, Dalhousie University) Dr. Mark Hills, Terry Fox)
11:45 a.m. – 12:30 p.m.	Lunch	



12:30 p.m. –
1:15 p.m.

Disease Modeling to Improve Patient Treatment

- Chronic childhood vasculitis: Characterizing the individual rare diseases to improve patient outcomes
- Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS): from models to treatment strategies

Dr. Susanne Benseler,
Toronto HSK

Dr. Bernard Brais, Montreal
Neurological Institute

1:15 p.m. –
2:00 p.m.

Stem Cell Research for Rare Diseases

- Induced pluripotent stem (iPS): Applications for Rare Disease
- Cancer risk related to DNA deficiencies and stem cell failure

William Stanford, U Alberta
Geraldine Aubert, Terry Fox

2:00 p.m. –
2:45 p.m.

Improving Healthcare for Rare Disorders

- Emerging team in rare diseases: achieving the "triple aim" for inborn errors of metabolism
- The Scleroderma Patient-centered Intervention Network (Dr. Brett Thombs,

Beth Potter, U Ottawa

Brett Thombs, Jewish
General Hospital

2:45 p.m. –
3:00 p.m.

Day 2 Conclusions and Next Steps

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