A Rare International Dialogue

May 10 – 12, 2019
Sheraton Centre Toronto Hotel
123 Queen St. W., Toronto, Ontario, Canada

1st Time in North America! Three days of dialogue and deliberation on all things rare: from research and policy to action and better outcomes

Hosted by Rare Disease International & Canadian Organization for Rare Disorders.

The largest and most significant gathering of rare disease advocates from patients to policy makers, from gene manipulators to AI gurus, from local support groups to international research networks coming together to Create a Vision for Rare Disease in the 21st Century and Develop Action Plans from Local to Global

DRAFT AGENDA

Friday, May 10, 2019: Vision for Rare Disease in 21st Century

Welcome to Rare Disease in the 21st Century

- Honourable Ginette Petitpas Taylor, Canada Minister of Health (Invited)
- Honourable Navdeep Bains, Canada Minister of Innovation, Science and Economic Development (Invited)
- Dr. Michael Strong, President, Canadian Institutes of Health Research (invited)
- Honourable Greg Hunt, Australia Minister of Health
- International Dignitaries (TBC)

Plenary: How Advocates Transformed the Rare Disease Landscape

1. Celebration of Rare Disease Heroes (and their impact)
   - Yann LeCam, Europe
   - Marlene Haffner, USA
2. **How Rare Disease Helped Transform the Healthcare Landscape**
   - Diagnostic Breakthroughs: From Newborn Screening to Next Gen Sequencing
   - Patient-Partnered Care: From Individualized Protocols to Best-Practice Guidelines
   - Drug Development: From Small Populations to Precision Therapies; Replacing Proteins to Replacing Genes
   - Patient Communities: From Supportive Carers to Engaged Experts

3. **Ideal but Realistic Rare Disease Future**
   - Lessons Learned: Proud’s and Sorry’s
   - What Life Could Be Like for A Child Born with a Rare Disease in 2040

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**Patient-Focused Drug Development - Joint afternoon session with RE(ACT)**

1. **Researchers and Regulators**
   - Marlene Haffner (USA)
   - Nick Sireau (UK)
   - Larissa Lapteva (USA)
   - Calvin Ho (USA)
   - Soo-Kyung Lee (USA)

2. **Patients, Parents, and Patient Groups**
   - Cystic Fibrosis (TBC)
   - Muscular Dystrophy (TBC)
   - Friedreich’s Ataxia (TBC)
   - Cystinosis (TBC)
   - Pompé’s Disease (TBC)
Saturday, May 11, 2019: Educational Forum

**Opening Plenary: Embracing New Opportunities and Challenges for Rare Disease in 21st Century**
- Success Stories: Patients, Treaters, Researchers, Policy Makers
- Panel Discussion: Building on Success and Tending to Unintended Consequences
- Hearing from Heroes of the Future

**Three Parallel Dialogue Streams**

1. Translating Research into Diagnosis and Care
   - Ending the Diagnostic Odyssey: Genomics, AI, Databases
   - Tools and Support for Patient and Parent Engagement
   - Frontline Healthcare Professionals and Other Community Workers

2. Innovations in Therapy: Treating More and Curing Some
   - 1st, 2nd, and 3rd Generation Rare Disease Therapies
   - Pipeline for Previously Untreated Disorders
   - Curative Treatments for Rare Diseases (Cell and Gene Therapies)

3. Addressing Issues of Diversity and Equity in Rare Disease
   - Strategies for Addressing Rare Disease in Emerging Healthcare Systems
   - Equitable Access to Clinical Trials and Therapies Across Nations and Incomes
   - Challenges of Rare Disease Specific to Underserved Populations

**Formats (Three 90 minute sessions)**
- Invited panel discussions: multi-stakeholder, multi-regional, multi-perspective
- Interactive dialogues and debates on hot topics (contrasting perspectives with audience engagement)
- Mini-workshop or demonstration of technology, technique, or process

**Panel Topics (Preliminary) (Three 90 minute sessions)**

1. Translating Research into Care and Treatment
   a. Topic 1: Winning the Race to Diagnosis
      i. What If... Routine Genome Sequencing Were Available to All?
ii. Reducing the Diagnostic Odyssey for Children: How A Global Commission is Helping Shape the Path Forward

iii. Are We Ready for AI-Enabled Phenotyping and Genotyping?

iv. How To Build a Program of Newborn Screening for Developed and Emerging Healthcare Systems

b. Topic 2: Designing Pathways to Patient-Centered Care
   i. EU Reference Networks
   ii. Case Examples: Hemophilia
   iii. Developing Best Practice Guidelines

c. Topic 3: Empowering the Whole Person
   i. Designing Care For and By Families
   ii. Designing Universal Health Coverage to Cover the Whole Person
   iii. Underserved Patients: How to Identify, Engage and Empower

2. Innovations in Therapy: Treating More and Curing Some
   a. Topic 1: Why is Orphan Drug Development Still Important?
      i. How Patient Groups Drive and Support Drug Development
      ii. IRDIRC Goal of 1000 New Therapies by 2025: What Will It Take?
      iii. Is Repurposing Drugs A Strategy for Rare Diseases?
   b. Topic 2: Curative Therapies: Next Step for Rare Diseases?
      i. Cellular Therapies
      ii. Gene Therapies
      iii. Targeted Precision Therapies
      iv. Preventive Therapies
   c. Topic 3: Programs and Strategies for Access to Innovative Therapies
      i. Tailoring Access to Income Levels (GDP)
      ii. Public, Private and Private/Public Pharmacare
      iii. Designated Orphan/Rare Drug Access Programs
Models: UK, France, Germany, Australia, Colombia, Taiwan, Japan, Canada, USA.

3. Addressing Issues of Diversity and Equity in Rare Disease
   a. Topic 1: Rare Advances at National, Regional, and Global Levels
      i. China
      ii. Colombia
      iii. EU Countries
      iv. Australia
      v. TBD
   b. Topic 2: Grassroots Strategies Across the Globe
      i. Taiwan Foundation for Rare Disorders
      ii. Agrenska: A Rare Service Model
      iii. Community-based RD Networks
   c. Topic 3: Ethnic and Indigenous Issues
      i. Rarest Communities
      ii. Rural Access
      iii. Indigenous Health
      iv. TBD

Closing Plenary:
- Dialogue Session Feedback: What we learned from each other and how we can use it
- Feedback from All: How to Move Forward Collaboratively
Sunday, May 12, 2019: Skills Training and Capacity Building (Not Just for Patients)

**Workshops (90 or 180 minutes): Basic and Advanced**

1. **Theme One: Cell and Gene Therapy**
   - Basic: Introduction to Gene Therapy
   - Advanced: Specific Applications and What Next

2. **Theme Two: Diagnosis**
   - Basic: Essential Elements of New Born Screening and Genetic Counselling
   - Advancing: How AI (Artificial Intelligence) Will Transform Diagnosing Rare Diseases

3. **Theme Three: Empowering the Patient Community**
   - Basic: Building and Supporting Your Rare Disease Community Using Social Media (Rare Connect)
   - Advanced: Expert Patient Skills and Knowledge Development
   - Building Patient Registry/Natural

4. **Theme Four: National Rare Disease Plans and Strategies**
   - Basic: Defining and Designing Core Areas of National RD Plans
   - Advanced: Implementing, Assessing, and Monitoring Progress in Rare Disease Plans

5. **Theme Five: Access to Rare Disease Medicines**
   - Basic: Principles, Policies and Practices in the Regulation and Reimbursement of Rare Disease Drugs (across nations)
   - Advanced: Alternative Access Schemes (Managed Access (Entry) Programs with Risk Sharing)

6. **Theme Six: Rare Disease in Low-and-Middle-Income Countries**
   - Basic: State-of-the-Art Across Key Countries and Pathways to Progress
   - Advanced: Policies and Practices to Insure “no patient is left behind” regardless of social, economic, and political circumstances

7. **OTHER WORKSHOP THEMES (TBD)**