Theme 6 | Global Rare Equity: Are we there yet?

Session 0602 Friday, 11 May (16:30– 18:00)
Global Access to Timely Accurate Diagnosis and Optimised Care

Moeen AlSayed, MD, FACMG, MBA

Professor of Genetics, College of Medicine, Alfaisal University
Director, MSc Genetic Counselling Program, Alfaisal University
Chairman, Department of Medical Genetics
King Faisal Specialist Hospital & Research Center

GLOBAL COMMISSION to End the Diagnostic Odyssey for Children with a Rare Disease

9th European Conference on Rare Diseases & Orphan Products (ECRD 2018)

The Diagnostic Odyssey

Rare diseases affect 300-350 million lives worldwide (7000 Diseases)

About 60% begin in childhood and are chronic.

On average, it takes five years before a rare disease patient receives the correct diagnosis.

Up to 40% of rare disease patients are misdiagnosed more than once.

An accurate and timely diagnosis is a key for effective treatment and prevention for the heritable forms. Also a key for understanding variation, natural history, modifiers and research.

The Global Commission

To address this challenge Shire, Microsoft, and EURORDIS-Rare Diseases Europe have announced a strategic alliance:

GLOBAL COMMISSION | to End the Diagnostic Odyssey for Children with a Rare Disease

A multi-disciplinary group of experts with the creativity, technological expertise and commitment required to make a major difference in the lives of millions of children and their families.

The Global Commission's Goal

1. Address **core barriers** and provide **recommendations** around these themes:
   - **Improving physicians' ability** to identify and diagnose patients with a rare disease.
   - **Empowering patients** and their families.
   - Providing high-level **policy guidance** to help achieve better health outcomes for rare disease patients.

2. Develop an **actionable roadmap** to help the rare disease field to shorten the multi-year diagnostic journey.

The Global Commission Members

CO-CHAIRS

Simon Kos

Microsoft

Yann Le Cam

EURORDIS RARE DISEASES EUROPE

Flemming Ornskov

Shire

MEMBERS

Moeen Al-Sayed

Kym Boycott

CHEO RESEARCH INSTITUTE

Roberto Giugliani

UFRGS UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL

Kevin Huang

Derralynn Hughes

Royal Free London NHS

NHS Foundation Trust

Daniel MacArthur

BROAD INSTITUTE

Maryam Mohd. Fatima Matar

دبي العطاء Dubai Cares

Dau-Ming Niu

Mike Porath

The MIGHTY

Arndt Rolfs

CENTOGENE THE RARE DISEASE COMPANY

Richard Scott

Genomics england

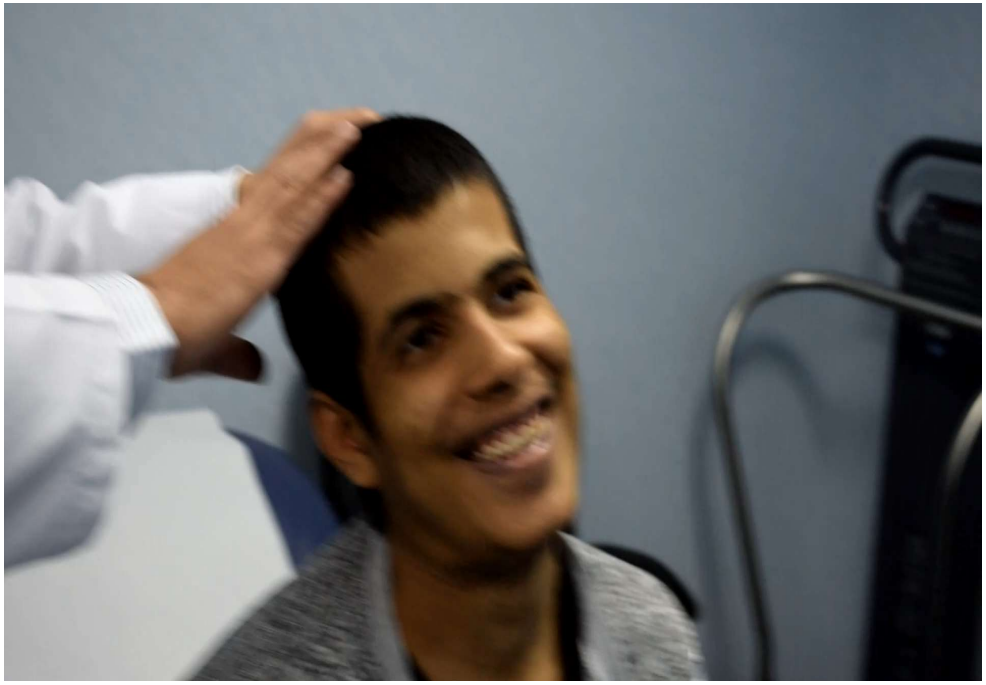
Marshall Summer

Children's National

Durhane Wong-Rieger

CORD Canadian Organization for Rare Disorders

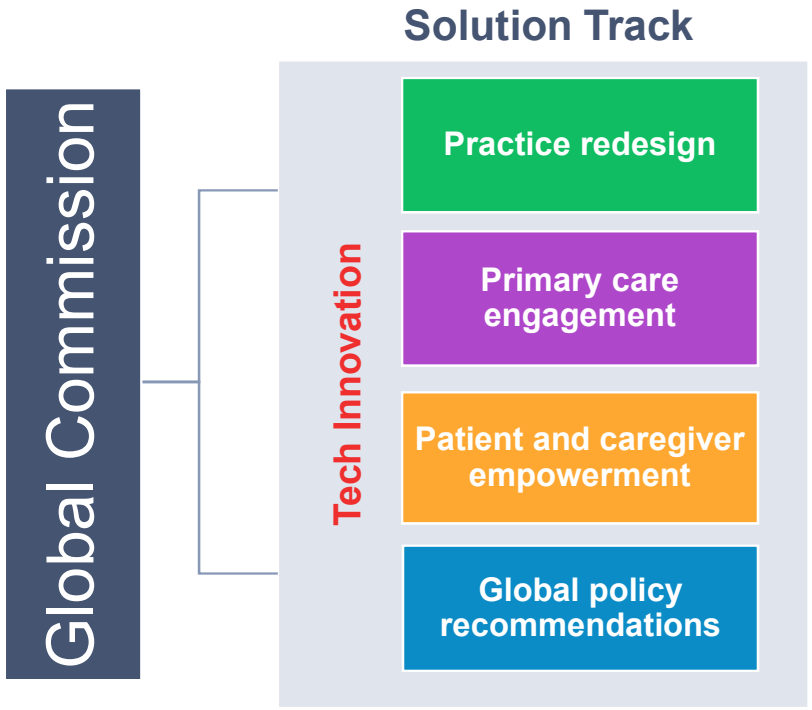
Example from the real world



Video obtained with parental permission

Commission tracks

The Commission will focus on developing solutions in four areas that will lead to accelerating the time to diagnosis – and will organize its work accordingly



Objective

Develop innovative ways to enable geneticists and specialists to operate more efficiently so they can see more rare disease patients quicker – especially given the growing shortage of geneticists

Apply innovation and creative thinking to improve primary care physicians' ability to identify patients with a rare disease and refer to appropriate follow-up care

Create or identify new tools and approaches to empower patients and caregivers so they can navigate the health system more effectively

Determine policy guidance at a global level that can be adapted to meet differentiated regional needs and work with national and local governments

Meeting 1 (April 1, 2018) : Recap

- 15 Commission members in attendance
- Presentation on Microsoft's work in health technology
- Two breakout groups for in depth discussion of solutions to barriers affecting Primary Care & Pediatrician Engagement and Limited Access to Geneticists
- Feedback and input on other group's recommended priorities
- 6 solutions identified for immediate action







Track 1 and Track 2 Barriers

Barriers for Primary Care and Pediatrician Engagement:	• Minimal / non-existent training or exposure to rare disease • Lack of standard criteria to help diagnose many rare diseases • Poor communication between PCPs and specialists • Lack of awareness around which specialty to refer • Access to diagnostic tools
Barriers for Practice Redesign:	• Shortage of geneticists • High workload = inability to see more patients • Fewer specialists in rural areas • Inefficient practices that limit productivity (e.g., diagnosed patients making follow up visits, tests not ordered in advance of patient visit)

Other Key Themes for Discussion

1. **What is the evolution of Next Generation Sequencing?**

As accessibility to NGS increases, how will this affect timely diagnosis?

2. **What is the role of genetics in the future of medicine? And what are the implications for geneticists?**

3. **Given how technology is constantly evolving, how do we plan for the future?**

The world may look different in 5 years, 10 years, etc. and we should be planning for 3 years ahead

Moving Forward

Next steps

- **Initiate pilot projects in areas where we can move quickly and generate evidence**
  - Each member send to Rabin Martin and Linn which Solutions they would like to directly contribute to
  - Members to identify opportunities:
    1. Determine who has data sets that are readily available to share
    2. Define technology that already exists that can be leveraged
- **Look for information on Virtual Meeting 2**
  - Engagement with online platform will begin week of April 30th
  - Track 3: Patient Empowerment
  - Track 4: Global Policy

Thank you

Linn Parrish (Head, Corporate Responsibility)
linn.parrish@shire.com

GLOBAL COMMISSION to End the Diagnostic Odyssey for Children with a Rare Disease

9th European Conference on Rare Diseases & Orphan Products (ECRD 2018)