The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease

A bold commitment on behalf of patients

ECRD 2018: Session 0301 The digital patient

Linn Parrish
Head of Responsibility
Shire

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

Why focus on the diagnostic journey?

6,000+
rare diseases worldwide

80%
of rare diseases are genetic in origin

5+ yrs
to receive a diagnosis

~40%
of patients are misdiagnosed

50%
of rare diseases typically occur in children

Source: Statistics and Figures on Prevalence of Genetic and Rare Diseases, Global Genes

Overcoming barriers to diagnosis with solutions

New interventions are needed to help physicians identify patients with a rare disease

Streamlining processes in a complicated healthcare system can improve time to diagnosis

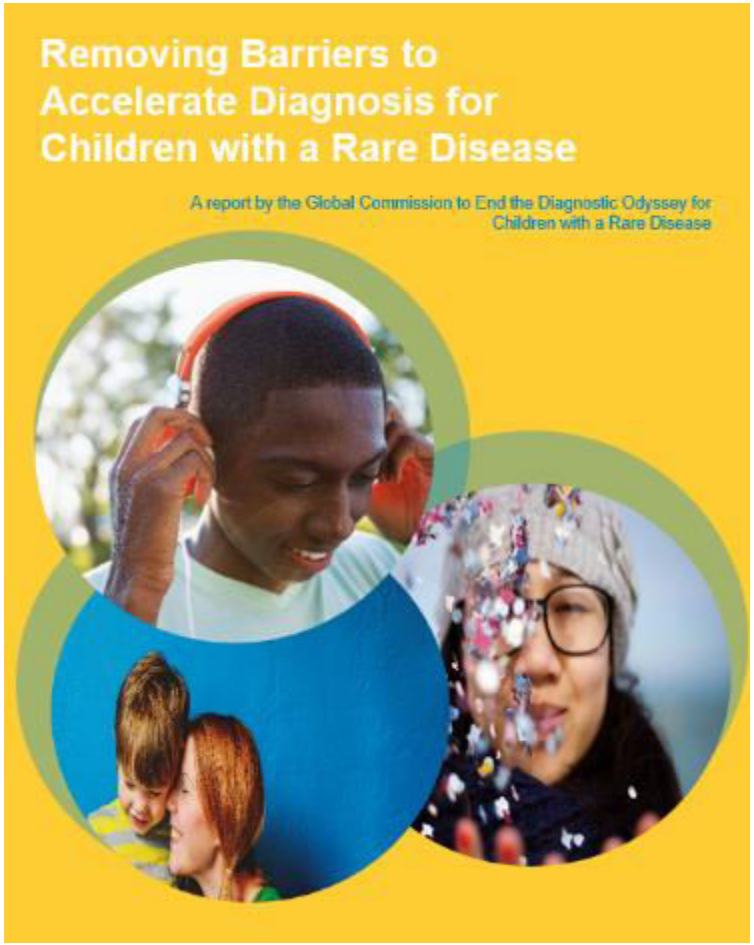
We do not have to wait for more geneticists and other specialists

There is opportunity to apply new technology to the rare disease field

Charting the path to shorten the journey to diagnosis

Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease

- Develop a roadmap to guide the rare disease field, addressing barriers to diagnosis and ways to eliminate them
- Mobilize diverse entities to work collaboratively toward a shared ambition
- Focus on diagnosing children
- Work towards achieving UN Sustainable Development Goal 3: Ensure healthy lives and promote well-being for all at all ages
- Provide actionable roadmap in early 2019



Illustrative report example

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Cross section of leaders to find solutions

Simon Kos

Microsoft

Yann Le Cam

EURORDIS RARE DISEASES EUROPE

Flemming Ornskov

Shire

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

Dubai Cares

Dau-Ming Niu

Mike Porath

The MIGHTY

Arndt Rolfs

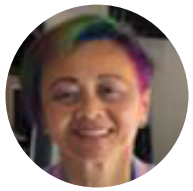
CENTOGENE THE RARE DISEASE COMPANY

Richard Scott

Genomics england

Marshall Summer

Children's National



Durhane Wong-Rieger

CORD Canadian Organization for Rare Disorders

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Solution tracks

Tech innovation

Primary care engagement	**Patient and caregiver empowerment**
Practice redesign	**Global policy recommendations**

Examples of enabling technologies

Artificial intelligence and machine learning everywhere

In combination to provide predictions and personalization around rare disease feature constellations

Patient and provider engagement

Around social computing; Create trust in the ecosystem through blockchain

Cognitive services

To add phenotypical recognition to genomic data to help overcome the difficulty of gene expression

Lowering cost

Of genome screening and analysis with cloud services and advance analytics