GLOBAL COMMISSION

to End the Diagnostic Odyssey for Children with a Rare Disease

Peter Jones
Industry Lead – Healthcare
Microsoft Canada

www.globalrarediseasecommission.com/report

Charting the Path to Shorten the Journey to Diagnosis

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

The Global Commission is a distinct, ground-breaking initiative that is:

- Developing **actionable recommendations** to address barriers to diagnosis of rare diseases
- **Mobilizing diverse entities** to work collaboratively toward a shared ambition
- Focusing on **diagnosing children** with rare diseases
- Working towards achieving **UN Sustainable Development Goal 3**: Ensure healthy lives and promote well-being for all at all ages

Launch of the Global Commission Report-Out

The final digital report-out launched publicly via the updated Global Commission website February 20, 2019 ahead of Rare Disease Day – exactly one year after it was announced.

Current Global Commission Co-Chairs

Dr. Wolfram Nothaft
Chief Medical Officer
Takeda

Dr. Simon Kos
Chief Medical Officer & Senior Director
Microsoft Worldwide Health

Yann Le Cam
Chief Executive Officer
EURORDIS-Rare Diseases Europe

The Global Commission Members are:

A diverse group of patient advocates, physicians, and other experts in the field who provide unique expertise to **solve challenges** affecting the rare disease community.

Cross Section of Leaders to Find Solutions

Simon Kos

Microsoft

Yann Le Cam

EURORDIS RARE DISEASES EUROPE

Wolfram Nothaft

Takeda

Moeen AlSayed

Kym Boycott

CHEO RESEARCH INSTITUTE

Pamela Gavin

NORD National Organization for Rare Disorders

Roberto Giugliani

UFRGS UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL

Kevin Huang

Derralynn Hughes

Royal Free London NHS

NHS Foundation Trust

Maryam Mohd. Fatima Matar

دبي العطاء Dubai Cares

Dau-Ming Niu

Anne O'Donnell-Luria

BROAD INSTITUTE

Boston Children's Hospital

Until every child is well

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

Why Focus on the Diagnostic Journey?

6,000+
rare diseases worldwide

There are more than 6,000 identified rare diseases worldwide. Almost 5 percent of the world's populations – an estimated 300-350 million people – are living with a rare condition.

40%
of patients are misdiagnosed

Up to forty percent of rare disease patients are misdiagnosed more than once or diagnosis is delayed for a variety of reasons.

80%
of rare diseases are genetic in origin

Most rare diseases are present throughout a person's life, even if symptoms do not immediately appear.

50%
of rare diseases typically occur in children

Approximately 50 percent of the people affected by rare diseases are children.

5+ years
to receive a diagnosis

It can take on average 5 years (and sometimes much longer) before a person with a rare disease receives the right diagnosis, with visits to different physicians.

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

Our Challenge

Significantly shorten the multi-year journey patients endure to receive a rare disease diagnosis

New interventions needed to help physicians identify patients.

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

Technology provides new opportunities to accelerate the time to diagnosis.

Streamlining processes in a complicated healthcare system to improve diagnosis.

Sergio’s Story

Global Commission Year One Report

www.globalrarediseasecommission.com/report

Year One Report: *Recommended Solutions*

TRACK 1

Empowering Patients and Families

TRACK 2

Equipping Frontline Providers with Tools for Diagnosis and Referral

TRACK 3

Reimagining the Genetic Consultation

www.globalrarediseasecommission.com/report

Year One Report: *Technology Pilot Programs*

PILOT 1

Multifactorial Machine Learning to Recognize Symptom Patterns

PILOT 2

Enable Collaboration Tools for "Intelligent Triage" and Clinical Geneticist Virtual Panel Consultation

PILOT 3

Explore a Blockchain-based Patient Registry and Rare Disease Passport

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Key Focus Areas

Centers of Excellence

Genetic Screening

Data Sharing

Privacy

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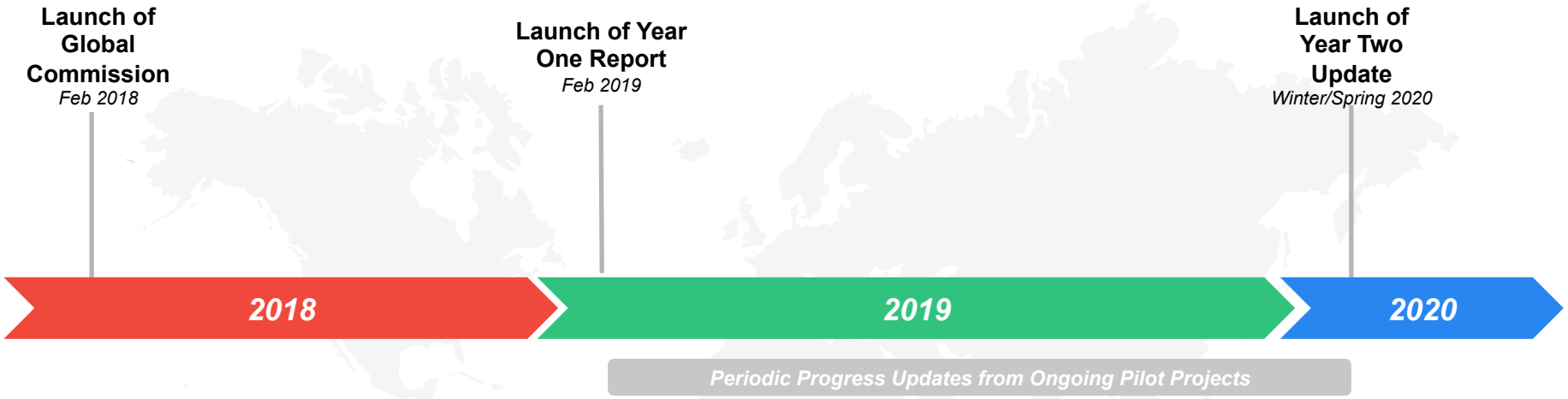
Global Commission: Timeline



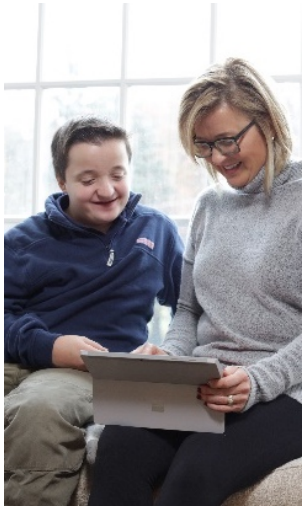
Looking Forward: 2019-2021

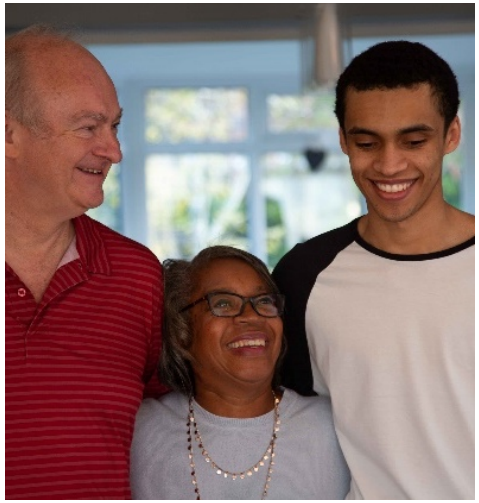
Vision | A year of action: Implementing and amplifying the Global Commission's recommendations

By 2020, the Global Commission aims to achieve the following goals |

- Continue to disseminate the Year One Report and recommendations to inspire global action in reducing time to diagnosis
- Show measurable progress on the three initial pilots launched in 2018
  - *Multifactorial Machine Learning to Recognize Symptom Patterns*
  - *Enable Collaboration Tools for "Intelligent Triage" and Clinical Geneticist Virtual Panel Consultation*
  - *Explore a Blockchain-based Patient Registry and Rare Disease Passport*
- Explore opportunities to enlist additional partners and leverage resources to push forward implementation of recommendations
- Support the effort to achieve recognition of rare disease as a priority within the World Health Organization (WHO) global agenda

Calling on Rare Disease Champions



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