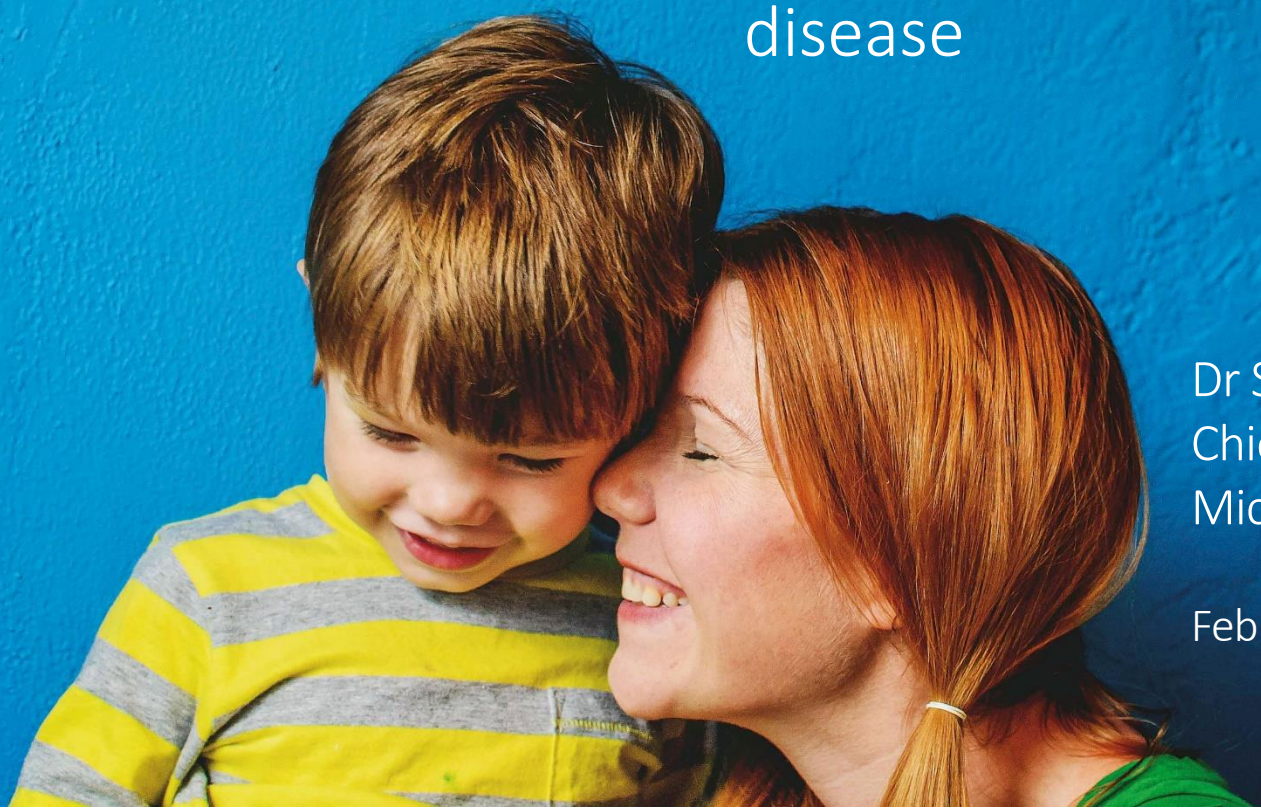
New opportunities to improve the diagnosis of children with a rare disease



Dr Simon Kos
Chief Medical Officer
Microsoft

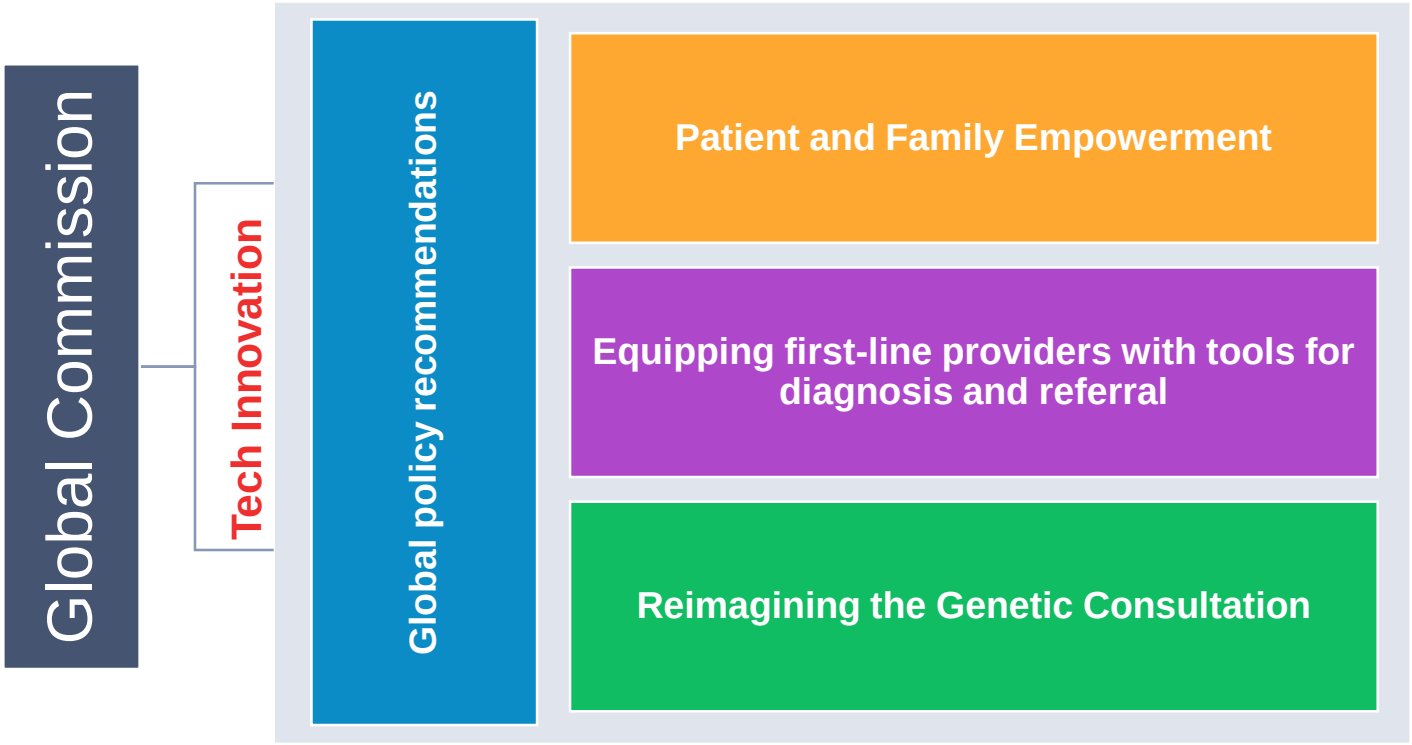
Feb 21st 2019

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

Recommendation tracks

After initial research, the Commission identified numerous barriers to diagnosis of rare diseases and divided those key barriers into three themes or "Tracks."

The Commission then developed technology-enabled recommendations across each Track that aim to address those barriers and accelerate the time to diagnosis.



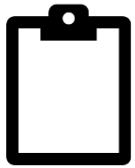
These recommendations are supported by global policy recommendations.

Technology pilots

The Global Commission is supporting pilot projects, which aim to provide a proof-of-concept for distinct initiatives that lend themselves to near-term scale.

Multifactorial machine learning to recognize symptom patterns

"Intelligent Triage" for Genetics Clinics and Virtual Panel Consultation



Explore a patient registry and Rare Disease Passport

MassChallenge: Pitch to accelerate diagnosis of rare disease

Multifactorial machine learning to recognize symptom patterns

- The Global Commission is leveraging the power of machine learning. This collection of digital technologies integrates with EMR systems to support primary care physicians and pediatricians by recognizing symptom patterns that they may not immediately associate with a rare disease, expediting patient diagnosis.
- Three areas of focus: (1) medical records data, (2) patient-reported data, and (3) integration with genomics
    - Current pilot with Hospital La Paz de Madrid in Madrid, Spain