ENDING THE DIAGNOSTIC ODYSSEY

FOR CHILDREN WITH A RARE DISEASE

The Rare Disease Diagnosis Burden

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

It is estimated that rare diseases affect **350 million** people worldwide.[1,2] About half of rare diseases begin in childhood[2,3] and many patients endure considerably reduced quality of life.[4] For a child living with a rare disease, an accurate and timely diagnosis can be the key to a longer, healthier life.[3] However, on average it takes six to eight years before a person with a rare disease receives the correct diagnosis. More than **40%** of rare disease patients are misdiagnosed at initial presentation and a correct diagnosis can be delayed for a variety of reasons.[4,5]

Delays in diagnosis are devastating to a child's – and family's – health and quality of life.

Receipt of inappropriate treatments[4,5]

Reduced quality of life and poor outcomes[3,4]

Delayed access to correct treatment resulting in disease progression and potentially irreversible complications[4]

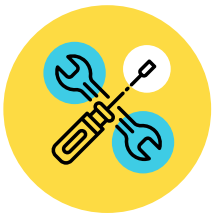
ABOUT THE GLOBAL COMMISSION

In 2018, Takeda (formerly Shire), Microsoft, and EURORDIS-Rare Diseases Europe launched the Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease. This multidisciplinary group of experts brings creativity, technological expertise and a commitment to make a major difference in the lives of millions.

The Global Commission developed an actionable roadmap to help the rare disease field to shorten the multi-year diagnostic journey, considered key to a longer, healthier life. Within its roadmap, the Global Commission offers recommendations designed to address core barriers preventing timely diagnosis impacting all rare disease patients, of which approximately half are children.

Empowering Patients and Families

- Equip families with information and tools to work collaboratively with their doctor to ask the right questions and get the answers they need
- Enable patients and their caregivers to share detailed medical records with multiple physicians and specialists quickly, efficiently and securely



Equipping Frontline Providers with Tools for Diagnosis and Referral

- Use artificial intelligence to identify symptoms that could help frontline providers detect patients who may have a rare disease
- Develop a platform for physicians to consult with genetics experts and specialists who can help guide a patient's next steps on the diagnostic journey

Reimagining the Genetic Consultation

- Facilitate quicker and more appropriate access to genetic consultations by creating a digital triage referral system based on standardized patient data
- Ensure that patients in remote areas have access to high quality diagnostic services by providing tele-consultations and sharing patient information more effectively

The Global Commission's goal is to inspire concerted action and mobilize diverse actors – within and outside the health field – to implement these recommendations and improve the lives of millions of children with a rare disease.

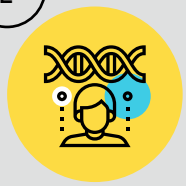
PILOT PROGRAMS + SHAPING POLICY

The Global Commission has launched three **technology pilot programs** that support our recommendations. Successful projects will hopefully be replicated and scaled over the next few years.

1

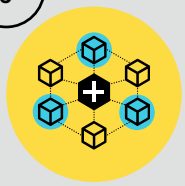
Multifactorial machine learning system to recognize patterns and symptoms common to rare diseases

2



Virtual tools to deliver genetic assessment and counseling remotely to patients and primary care physicians

3



Blockchain-based patient registry to give patients and their families control of their health records

GLOBAL POLICY RECOMMENDATIONS

Centers of Excellence

Data Sharing

In its roadmap, the Global Commission also emphasizes the importance of global policy frameworks for rare diseases to be recognized as an international public health priority.

The policy recommendations, designed to support and enable the specific solution pathways, focus on four key areas.

Genetic Screening

Privacy

THE GLOBAL COMMISSION MEMBERSHIP

Under the leadership of co-chairing organizations, Takeda, Microsoft and EURORDIS-Rare Diseases Europe, the Global Commission brings together a dedicated group of technology innovators, patient advocates, healthcare providers, researchers, family members and other experts from around the world to tackle one of the most serious and heartbreaking challenges within the rare disease space.

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To learn more about the Global Commission's work, please visit: https://www.globalrarediseasecommission.com/report

If you are a healthcare professional and would like to get involved, please contact us at: questions@globalrarediseasecommission.com

References:

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