INTRODUCTION:
A LETTER FROM THE CO-CHAIRS

More than 300 million people worldwide are currently living with a rare disease, and about half of these diseases appear in childhood. Some estimates show that it can take five to seven years to get an accurate diagnosis of a rare disease, even in countries with the most sophisticated health systems. The impact of these delays can be devastating for children and their families.

Since launching in 2018, the Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease – a multidisciplinary group of rare disease advocates, researchers, physicians and technologists – has been working to end this long and arduous diagnostic journey.

Following publication of the Global Commission’s Year One Report (2019) and roadmap to end the diagnostic odyssey, our members have contributed their expertise, creativity and passion to build ambitious pilot projects that address central challenges to diagnosis. A centerpiece of our efforts is harnessing technology to develop new standards of practice that will benefit patients, families and healthcare providers.

This update highlights the strong progress we’ve made toward our key recommendations:

Empower families to play an active role in finding a diagnosis for their child
Equip frontline providers with tools for diagnosis and referral
Reimagine the genetic consultation

We are proud to share our progress, especially as we reflect on the unprecedented challenges brought on by the Covid-19 pandemic. Amid this global crisis, we’ve also seen the increasingly critical role of digital health solutions in supporting patients, providers and health systems around the world. Given the

rapidly evolving global health landscape, we are more inspired than ever to produce and scale technology solutions that expedite rare disease diagnosis for children, wherever they live.

While much exciting work is underway, we know that we cannot achieve our vision alone. We are eager to collaborate with others pursuing innovations to ensure a timely, accurate diagnosis for all. Please contact us if you would like to join our efforts or if you have any suggestions to share.

Thank you to the committed and passionate members of the Global Commission for pushing us towards a brighter future, even in the face of difficult circumstances.

Sincerely,

Wolfram Nothaft, MD
Chief Medical Officer
Takeda

Gregory Moore, MD, PhD
Corporate Vice President
Microsoft Health

Yann Le Cam
Chief Executive Officer
EURORDIS-Rare Diseases Europe
A clear path to a timely, accurate diagnosis for all children.

BY 2022
Leverage our members’ rare disease and technology expertise and networks to design, foster and scale solutions that overcome major challenges to rare disease diagnosis.

BY 2025
Demonstrably shorten the time to rare disease diagnosis for children in countries around the world.
EMPOWERING FAMILIES TO PLAY AN ACTIVE ROLE IN FINDING A DIAGNOSIS FOR THEIR CHILD

Patient Empowerment & Awareness Campaign

CHALLENGE

Though over 300 million people suffer from a rare disease, the term "rare" has led to a misconception that it is unlikely one will encounter someone with a rare disease. Patients and caregivers often do not suspect that a child may be suffering from a rare disease, thus extending the diagnostic journey. Repeated visits to the emergency room, consultations with various specialists and internet searches can lead to "dead-end" paths, resulting in an ongoing "questioning cycle" common to many families.

SOLUTION

This month, the Global Commission launched RareNavigator, a pilot campaign in San Diego, California, U.S. to raise awareness of rare disease as a consideration among parents and caregivers searching for a diagnosis for their child. Through social media, local media, collaboration with local advocates and healthcare facilities, and more, the campaign identifies parents who are early in their child's diagnostic journey. RareNavigator uses Microsoft technology to connect families to a tailored set of tools and resources that help them find appropriate care for their child. While RareNavigator is not a diagnostic tool, it encourages parents and caregivers to consider rare disease as a possibility and empowers them to have informed discussions with their child’s doctor(s), ultimately contributing to a faster diagnosis.

EARLY SUCCESSES:


NEXT STEPS:

Campaign will be rolled out in cities in developed countries throughout 2021. The intent is to then adapt and scale the campaign to a more diverse set of countries, starting in 2022.
Machine learning to accelerate rare disease diagnosis: Dx29

CHALLENGE
Physicians may lack the awareness to consider a rare disease when a patient has abnormal symptoms and may not be equipped with the right knowledge, tools and time to make the best decision about what to do next. As a result, patients may be referred to a variety of specialists – sometimes across different health systems or regions and with little communication between institutions – delaying diagnosis and increasing the risk of misdiagnosis.

SOLUTION
The Global Commission partnered with Foundation 29 – a nonprofit organization based in Spain – to develop a tool that uses machine learning to identify and link rare disease symptoms. The Dx29 algorithm seeks to save time and holds potential to improve the accuracy of diagnosis by generating a list of possible conditions a patient might have. A physician who suspects a patient is suffering from a rare disease is then able to request additional testing and make appropriate referrals more quickly and efficiently.

EARLY SUCCESSES:
• Dx29 correctly predicted diagnoses for 79% of previously studied cases.
• The tool has been tested among 400 healthcare providers.

NEXT STEPS:
Dx29 is expanding beyond Spain to reach providers and patients in additional countries.
REIMAGINING THE GENETIC CONSULTATION

Improving triage and facilitating access to geneticists: Genetics Opinion App

CHALLENGE
About 80% of rare diseases are genetic in origin, which means many patients are dependent on clinical geneticists to find a diagnosis. However, there is a global shortage of these specialists, making it difficult for patients to receive a genetic consultation and appropriate diagnostic tests in a timely manner. Some estimates show it can take between three and nine months for a patient to be seen by a geneticist.5

SOLUTION
The Global Commission is working with Children’s National Hospital in Washington, DC, to develop the Genetics Opinion App, a tool that helps providers more efficiently determine the best next steps to support a patient’s diagnosis, including referrals to the appropriate specialist(s).

Through the app, physicians can share patient data and solicit guidance from specialists about the right course of action. The goal is to reduce unnecessary testing and ensure more effective use of geneticists’ limited availability.


EARLY SUCCESSES:
In early use of the app, physicians received input from clinical geneticists within days compared to the usual several months. With increased uptake of the tool, the average wait time for patients to see a clinical geneticist is estimated to decline by 50%.

NEXT STEPS:
Test the app with Children’s National Hospital and area hospitals that refer to them. Scale to other clinical genetic centers in the U.S. and potentially scale to other countries.
**Our 2021 Priorities:**

1. **Launch and Expand the Patient Empowerment and Awareness Campaign:** Evaluate the U.S. pilot campaign and adapt learnings for implementation in cities in developed countries. Explore expanding to more diverse contexts in 2022 and beyond.

2. **Scale Technology Pilots:** Roll out the technology pilots to new health systems, regions and countries. Gather meaningful data and continue to refine the tools so they meet the needs of patients, families and health providers around the world.

3. **Support New Technology Solutions:** Work with Global Commission members and others in the rare disease community to explore additional technology solutions that could be piloted.

4. **Advocate for Enabling Policies:** Continue to support national and global policies that help end the diagnostic odyssey, including policies that enable the use of technology to inform health decision making.

**Get Involved!** Check our website for news updates or contact us if you want to learn more or participate in any of the Global Commission’s initiatives.