
Takeda, Microsoft and EURORDIS release report with action plan to help rare disease community shorten the diagnostic odyssey

Global Commission's recommendations focus on three solution pathways that leverage technology to address key elements of the rare disease diagnostic odyssey

Rare diseases remain undiagnosed as patients struggle to navigate complex health systems to receive accurate diagnoses, which can take an average of five years to receive, even in countries with sophisticated health systems.

New York, NY, and Brussels, Belgium - Feb. 20, 2019 – The Global Commission to End the Diagnostic Odyssey for Children (“the Global Commission”), co-chaired by Takeda, Microsoft and EURORDIS-Rare Diseases Europe, an alliance of more than 800 rare disease patient organisations, today announced its actionable [recommendations](#) in a report to address the barriers to diagnosis for people living with a rare disease.

In 2018, the Global Commission Co-Chairs, Shire (now Takeda), Microsoft, and EURORDIS, joined forces to bring together a multidisciplinary group of patient advocates, physicians and other experts to help solve the complex challenges impacting the rare disease community. Over the past year, the Global Commission gathered input from patients, families and other expert advisors to gain key insights to guide solutions to shorten the rare disease diagnosis timeline. The roadmap's recommendations can be mapped back to three solution pathways:

1. **Empowering patients and families:** Create opportunities to develop tools for caregivers to connect seemingly unrelated symptoms, inquire about additional testing and work together with physicians to achieve a correct diagnosis.
2. **Equipping frontline providers:** Examine ways to equip physicians with the knowledge and tools to quickly and effectively identify patients who may have a rare disease and take appropriate action through solutions such as machine learning technology, expert-level guidance and genetic testing opportunities.
3. **Reimagining the genetic consultation:** Identify innovative ways to enable medical geneticists to see priority patients more quickly, such as standardizing reporting methods and utilizing telemedicine for increased access to more patients.

“There are more than 6,000 identified rare diseases, the vast majority of which begin in childhood, with patients receiving a misdiagnosis more than once in 40 percent of cases,” said Wolfram Nothaft, M.D., Chief Medical Officer of Takeda and Global Commission Co-Chair. “As champions for those living with a rare disease, we’ve outlined real solutions to lessen the time to diagnosis for the more than 300 million people affected worldwide.”^{1,2,3}

“The too often long road to diagnosis presents one of the greatest challenges affecting the health, survival, well-being and indeed the very identity of people affected by a rare disease and their families. This report identifies concrete policy and technical actions, mobilizing diverse actors to build on genetic and digital cutting-edge advances.” said Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe and Global Commission Co-Chair.

The Global Commission is supporting three pilot projects to bring its solution pathways to life, utilizing the expertise of its members and engaging like-minded partners who are pushing the boundaries of innovation. The pilot programs include 1) multifactorial machine learning to recognize symptom patterns, 2) collaboration tools for “intelligent triage” and clinical geneticist virtual panel consultation, and 3) developing a secure a blockchain-based patient registry and rare disease passport, which may use emerging technologies like blockchain.

¹ European Medicines Agency. Orphan Medicines in the EU.

http://www.ema.europa.eu/docs/en_GB/document_library/Leaflet/2017/12/WC500240710.pdf

² Global Genes. Survey says: Misdiagnosis of rare diseases is common. <https://globalgenes.org/raredaily/survey-says-misdiagnosis-of-rare-diseases-is-common/>.

³ Global Genes. RARE Disease: Facts and Statistics. <https://globalgenes.org/rare-diseases-facts-statistics/>

“We believe that technology provides an unheralded opportunity to help overcome the barrier of ‘rare,’ and unfortunately, ‘rare’ often means ‘off the radar,’” said Dr. Simon Kos, Chief Medical Officer and Senior Director, Microsoft Worldwide Health and Global Commission Co-Chair. “Many of our recommendations address distinct challenges within rare disease that technology is uniquely equipped to solve.”

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: Centers of Excellence, Genetic Screening, Data Sharing and Privacy.

The Global Commission will discuss its recommendations live and via global [livestream](#) at the simultaneous roadmap launch events at Microsoft Headquarters in New York, NY and Brussels, Belgium, beginning at 11:00 a.m. ET / 6:00 p.m. CET. On February 28, in Beijing, China, coinciding with Rare Disease Day, the Global Commission will co-host a launch event with the Chinese Organization for Rare Disorders (CORD) to further discuss the implications of the Global Commission’s recommendations for the rare disease community.

For more information on the solutions and related research, the full report can be accessed [here](#).

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

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease was created in collaboration by Takeda, Microsoft and EURORDIS-Rare Diseases Europe. The purpose of the Global Commission is to establish a roadmap for the rare disease field that focuses on solutions to core barriers preventing timely diagnosis for all rare diseases – with an emphasis on those affecting children. The Global Commission brings together representatives from multiple sectors to provide diverse perspectives on rare disease diagnostics.

[The Global Commission Members:](#)

Simon Kos, MBBS, BSc (Med), MBA, Chief Medical Officer and Senior Director, Microsoft Worldwide Health (Co-Chair)

Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe (Co-Chair); Rare Diseases International Council Member

Wolfram Nothhaft, M.D., Chief Medical Officer, Takeda (Co-Chair)

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Kym Boycott, M.D., Ph.D., FRCPC, FCCMG, Clinical Geneticist, Senior Scientist, CHEO Research Institute; Professor, Department of Pediatrics, University of Ottawa

Pamela K. Gavin, MBA, Chief Strategy Officer, National Organization for Rare Disorders

Roberto Giugliani, M.D., Ph.D., Professor, Department of Genetics, Federal University of Rio Grande do Sul; Medical Genetics Service, Hospital de Clínicas de Porto Alegre, Brazil

Kevin Huang, Founder and President, Chinese Organization for Rare Disorders; Rare Diseases International Member

Derralynn Hughes, Ph.D., Clinical Director Haematology Oncology and Palliative care, Senior Lecturer and Investigator Lysosomal Storage Disorders Unit, Royal Free & University College Medical School

Anne O’Donnell-Luria, M.D., Ph.D., Associate Director, Center for Mendelian Genomics, Broad Institute; Physician, Division of Genetics and Genomics, Boston Children’s Hospital

Maryam Matar, M.D., Founder and Executive Director, UAE Genetic Diseases Association

Dau-Ming Niu, M.D., Ph.D, Chairman, Department of Pediatrics, Director, Medical Genetics Center, Taipei Veterans General Hospital; Professor, Institute of Clinical Medicine, National Yang Ming University

Mike Porath, Founder and CEO, The Mighty; Board Member, Dup15q Alliance

Arndt Rolfs, M.D., CEO, Centogene

Richard Scott, Ph.D. Clinical Lead for Rare Disease, 100,000 Genomes Project at Genomics England and Consultant and Honorary Senior Lecturer in Clinical Genetics, Great Ormond Street Hospital for Children and the UCL Institute of Child Health

Marshall Summar, M.D., Director: Rare Disease Institute, Chief, Genetics and Metabolism Margaret O’Malley Chair of Genetic Medicine Children’s National Medical Center, Washington, D.C. Professor of Pediatrics, George Washington School of Medicine

Durhane Wong-Rieger, Ph.D., President & CEO, the Canadian Organization for Rare Disorders; Council Chair, Rare Diseases International, Director, Asia Pacific Alliance of Rare Disease Organizations

About Takeda Pharmaceutical Company Limited

Takeda Pharmaceutical Company Limited (TSE:4502/NYSE:TAK) is a global, values-based, R&D-driven biopharmaceutical leader headquartered in Japan, committed to bringing Better Health and a Brighter Future to patients by translating science into highly-innovative medicines. Takeda focuses its R&D efforts on four therapeutic areas: Oncology, Gastroenterology (GI), Neuroscience, and Rare Diseases. We also make targeted R&D investments in Plasma-Derived Therapies and Vaccines. We are focusing on developing highly innovative medicines that contribute to making a difference in people's lives by advancing the frontier of new treatment options and leveraging our enhanced collaborative R&D engine and capabilities to create a robust, modality-diverse pipeline. Our employees are committed to improving quality of life for patients and to working with our partners in health care in approximately 80 countries and regions.

For more information, visit <https://www.takeda.com>

About Microsoft

Microsoft (Nasdaq "MSFT" @microsoft) is the leading platform and productivity company for the mobile-first, cloud-first world, and its mission is to empower every person and every organization on the planet to achieve more.

About EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 800 rare disease patient organisations from 70 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services. Follow [@eurordis](#) or see the [EURORDIS Facebook page](#). For more information, visit www.eurordis.org.

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