



Fact Sheet

THE DIAGNOSIS BURDEN

It is estimated that rare diseases affect 300-350 million people worldwide. About half of all rare diseases begin in childhood and many patients endure lifelong suffering. For a child living with a rare disease, an accurate and timely diagnosis can be the key to a longer, healthier life. However, on average, it takes five years before a rare disease patient receives the correct diagnosis. Up to 40% of rare disease patients are misdiagnosed more than once or diagnosis is delayed for a variety of reasons.

THE GLOBAL COMMISSION

Shire, Microsoft, and EURORDIS-Rare Diseases Europe have announced a strategic alliance to address the diagnostic challenge for patients living with a rare disease. The long road to diagnosis is one of the most important issues affecting the health, longevity and well-being for rare disease patients and their families.

The Global Commission to End the Diagnostic Odyssey for Children ("the Global Commission") is a multi-disciplinary group of experts with the creativity, technological expertise and commitment required to make a major difference in the lives of millions of children and their families.

THE GLOBAL COMMISSION GOAL

The Global Commission will develop an actionable roadmap to help the rare disease field to shorten the multi-year diagnostic journey, considered a key to a longer, healthier life.

Within its roadmap, the Commission will offer recommendations designed to address core barriers preventing timely diagnosis impacting all rare disease patients, of which approximately half are children, such as:

- Improving physicians' ability to identify and diagnose patients with a rare disease in order to begin care and treatment
- Empowering patients and their families to have a more active role in their health care
- Providing high-level policy guidance to help achieve better health outcomes for rare disease patients

THE GLOBAL COMMISSION MEMBERS

Under the leadership of its co-chairs, Flemming Ornskov, M.D., M.P.H., Chief Executive Officer, Shire, Simon Kos, M.D., Chief Medical Officer and Senior Director, Worldwide Health, Microsoft, and Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe, the Global Commission will bring 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.

