A common reference developed by RDI in collaboration with a multi-stakeholder, global panel of rare disease experts to inform decision-makers.

WHAT diseases are considered rare.

HOW MANY persons are affected.

WHY the rare disease population demands specific attention

Share your feedback on the Operational Description with the RDI team: mary.wang@rarediseasesint.org
PLWRD face distinct and significant challenges that arise from the infrequency of their medical conditions, such as a long diagnostic journey, inadequate clinical management, and limited access to effective treatments. The burden of rare diseases on patients, their carers and families, healthcare systems, and society overall, merits greater visibility and recognition.

A rare disease is a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons living in any WHO-defined region of the world.

Rare diseases include, but are not limited to, rare genetic diseases. They can also be rare cancers, rare infectious diseases, rare poisonings, rare immune-related diseases, rare idiopathic diseases, and rare undetermined conditions.

While the frequency of most rare diseases can be described by prevalence, some rare diseases, such as rare cancers and rare infectious diseases, can be more precisely described by incidence.