



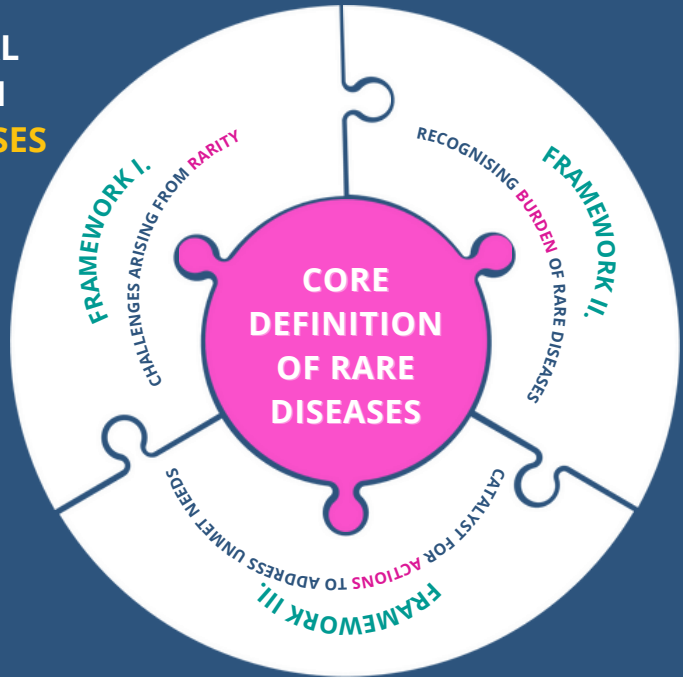
OPERATIONAL DESCRIPTION OF RARE DISEASES

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

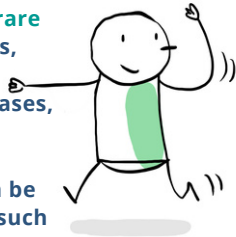
CORE DEFINITION OF RARE DISEASES

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.**