Action Area 5: Data and Monitoring



Data & Monitoring

FOCUS OF THIS SECTION:

Strengthen systems for collecting information about rare diseases, their impact, and services available, especially in low-income countries

Make it easier for policymakers to get reliable data, so they can set priorities and respond to the needs of people living with rare diseases

Keep track of data on rare diseases to better understand changes, identify gaps, and support policies and actions

5.1 DATA COLLECTION

Countries need reliable, standardized and accessible data to understand how rare diseases affect populations, plan health services and develop appropriate responses. However, significant gaps in rare disease data exist in countries at all income levels.

Rare disease registries and biobanks can help collect and store information on diseases, symptoms, progression and outcomes, supporting research and the development of new treatments.

Data should also be broken down by factors such as age, sex and access to health services to identify inequalities and gaps in care. Using common disease classification systems, such as the WHO's ICD, can help countries consistently record and report rare diseases.

PROPOSED ACTIONS

For Member States:

Consider using coding systems to record and monitor rare diseases

Establish a national registry or integrate the monitoring of rare diseases into routine health information systems

Report and analyze data on individuals living with a rare disease to assess progress and identify inequities

Develop and strengthen data protection and privacy policies

For the WHO:

Offer technical support to Members States to develop and/or improve national health information systems and registries to strengthen disaggregated data collection for rare diseases

Provide support to Member States to develop a core set of indicators and targets to monitor in relation to rare diseases

For International & National Partners

Support patient registries and monitoring programmes

Encourage analysis and publication of data on the availability, use and coverage of health and social services and treatments for rare diseases

Advocate for and facilitate the involvement of PLWRD, their families and caregivers in the collection, analysis and use of data on rare diseases

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5.2 DATA SHARING

Because rare diseases affect small and widely dispersed populations, no single country is likely to have sufficient data to characterize the full range of diseases, symptoms, progression and treatment outcomes. Pooling and sharing data across countries and stakeholders helps build evidence, support research and clinical decision-making, and improve diagnosis, treatment, and care.

To safely share health data and knowledge between countries, common standards and definitions are needed. Countries should also ensure that their databases and health information systems can work together, with clear rules for consent and data protection.

PROPOSED ACTIONS

For Member States:

Develop and strengthen data-protection frameworks and processes that enable responsible data sharing and pooling, including open access where appropriate, to advance research and development

Evaluate ways to reduce structural barriers to cross-border data sharing, such as reviewing existing legislation, and documentation and approvals needed

Collaborate with existing regional and international registries, as appropriate, to strengthen capacity on data collection and analysis, while respecting data protection and privacy

For the WHO:

Support Member States in creating cross-country data dashboards that highlight, at a minimum, a core set of indicators related to rare diseases

Provide Member States guidance on harmonizing data standards and case definitions and designing interoperable registries, as well as dealing with privacy and intellectual property concerns related to cross-border data sharing

For International & National Partners

Contribute to national and regional initiatives that facilitate the development of interoperable rare disease registries and data systems, and support responsible cross-border data sharing

Support the creation of exchange and dialogue platforms between countries for good practices in collection, management, use and sharing of data

AREA 5: DATA AND MONITORING

80. Information systems for rare diseases are often rudimentary or absent, especially in low-income countries, which complicates data acquisition on the full spectrum and magnitude of the burden of rare diseases and the availability and utilization of services by people living with a rare disease, their families and caregivers.

81. Absence of reliable information means policymakers do not have the data to inform health system priorities and to respond appropriately to the burden of rare diseases in their respective health systems.

82. The systematic integration of health and social care data collection into population-level and routine health information systems, and the regular monitoring of rare diseases based on a core set of measures, can promote better understanding and detection of population-level changes and trends, support the identification of gaps in service delivery, form the basis of evidence-based policies and actions, and enable implemented actions to be monitored.

5.1 DATA COLLECTION

83. Robust, standardized, interoperable and accessible data are essential for understanding the burden of rare diseases, informing health-system planning, and designing targeted interventions. However, significant data gaps persist in both low- and middle-income and high-income countries.

84. Rare disease registries are valuable for systematically collecting data on disease burden and prevalence, symptoms, progression and outcomes, and for supporting research, natural history studies, clinical trials and access pathways. Biobanks can complement registries by enabling the standardized collection, storage and use of biospecimens and linked health data for research and clinical innovation.

85. To identify and address barriers to equitable care and services, data should be appropriately disaggregated by demographic variables and health-related outcomes, including access and coverage of services. The WHO International Classification of Diseases (ICD) [10] includes specific codes that support the recording and reporting of rare diseases globally.

PROPOSED ACTIONS

86. *Member States*
    a. Consider implementation of interoperable codification systems for rare diseases, such as ICD codes, to enable the recording, reporting and monitoring of rare diseases at national and international levels.

[10] International Classification of Diseases 11th Revision [website]. World Health Organization; 2026 (https://icd.who.int/en/)

b. Establish a national registry for rare diseases or integrate the monitoring of rare diseases into routine health information systems, as appropriate, to collect core data, disaggregated by sex, age and other equity measures. Ensure data covers diagnosis, treatment, health-related and service-related outcomes.
c. Routinely report and analyse disaggregated data on individuals living with a rare disease, including by characteristics relevant to national contexts to assess progress towards the improvement of the status of people living with a rare disease and identify inequities.
d. Develop and strengthen data protection and privacy policies to safeguard confidentiality of personal data and appropriate consent to data use.

87. *Secretariat*
    a. Offer technical support to Members States to develop and/or improve national health information systems and registries to strengthen disaggregated data collection for rare diseases.
    b. Provide support to Member States to develop a core set of indicators and targets to monitor in relation to rare diseases.

88. *International and national partners*
    a. Provide support to Member States in establishing patient registries and monitoring programmes to capture core indicators and patient outcome measures on rare diseases.
    b. Encourage analysis and publication of data on the availability, utilization and coverage of health and social services and effective treatments for rare diseases.
    c. Advocate for and facilitate the involvement of people living with a rare disease, their families and caregivers in the collection, analysis and use of data on rare diseases.

5.2 DATA SHARING

89. Because rare diseases affect small and widely dispersed populations, no single country is likely to have sufficient data to characterize the full range of diseases, symptoms, progression and treatment outcomes. Pooling and sharing data across countries and stakeholders are therefore essential to build a more complete evidence base, support research and clinical decision-making, and improve diagnosis, treatment, and care.

90. Harmonized data standards and case definitions, interoperable databases and health information systems, and transparent consent and data-protection frameworks are needed to enable responsible and secure cross-border data sharing and knowledge exchange.

PROPOSED ACTIONS

91. *Member States*
    a. Develop and strengthen data-protection frameworks and processes that enable responsible data sharing and pooling, including open access where appropriate, to advance research and development.

b. Evaluate ways to reduce structural barriers to cross-border data sharing, such as reviewing existing legislation, and documentation and approvals needed.
c. Collaborate with existing regional and international registries, as appropriate, to strengthen capacity on data collection and analysis, while respecting data protection and privacy.

92. *Secretariat*

a. Support Member States in creating cross-country data dashboards that highlight, at a minimum, a core set of indicators related to rare diseases.
b. Provide Member States guidance on harmonizing data standards and case definitions and designing interoperable registries, as well as dealing with privacy and intellectual property concerns related to cross-border data sharing.

93. *International and national partners*

a. Contribute to national and regional initiatives that facilitate the development of interoperable rare disease registries and data systems, and support responsible cross-border data sharing.
b. Support the creation of exchange and dialogue platforms between countries for good practices in collection, management, use and sharing of data.