# Action Area 2: Diagnosis and Treatment



## Diagnosis and Treatment

## FOCUS OF THIS SECTION:

Equitable access to early detection and diagnosis, including new methods such as genomic testing

Public health measures to decrease the frequency of certain rare diseases, adapted to local contexts

Strengthening of health systems, including access to medicines and assistive technology

## 2.1 DIAGNOSTICS

Early detection and diagnosis of rare diseases can prevent the onset of disease symptoms or delay disease progression and improve quality of life for individuals, their families, and caregivers and society

Identification of rare diseases can be strengthened through expanded newborn screening, as well as equitable access to accurate diagnostic tools and appropriate counselling and post-diagnostic support.

Accurate and timely rare disease diagnosis requires trained personnel and adequate laboratory capacity and equipment

New diagnostic methods like genomic testing offer great benefits but they are complex and expensive. Diagnosis should be equitable and prioritize accurate and timely diagnosis wjere the impact is greatest, and with a balanced approach towards the use of limited health system resources.

## PROPOSED ACTIONS

### For Member States:

Invest in diagnosis by training primary health providers and improving access to diagnostic tools

Expand access to specialized diagnostic expertise

Implement evidence-based strategies like newborn screening

Improve access to genetic counselling and other support for individuals and families after diagnosis

### For the WHO:

Support the exchange of knowledge on public health measures, newborn screening, early diagnosis and post-diagnosis support, including genetic counselling

Update technical guidance to include tools relevant for rare diseases

Provide technical guidance around workforce training and establishing regulatory frameworks for screening and diagnosis

### For International & National Partners

Promote efforts for improving access to affordable, safe, effective, and high-quality diagnostics

Partner with health care professionals, academia, industry and PLWRD and their families to create and enhance coordinated platforms for diagnosis

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## 2.2 MEDICINES AND HEALTH PRODUCTS

The development of medicines, medical devices, assistive products and procedures is limited.

Regulators and health technology accessors face challenges in interpreting limited clinical and quality of life evidence. The models they use do not fully take into account rare conditions, how they progress, or how they are treated.

Even when treatments and technologies are available, high costs mean that access is delayed, inconsistent and inequitable. PLWRD need better availability, affordability and access to medicine and health products.

## PROPOSED ACTIONS

### For Member States:

Promote dialogue to identify data gaps and agree on approaches.

Promote access to medicines and health products through regulatory mechanisms

Encourage HTA evaluation frameworks that consider the circumstances of each rare condition

Align national treatment guidelines and medicine and health product lists to include interventions relevant to rare diseases

Facilitate regional or multi-country procurement mechanisms to improve affordability and availability, and stockpiling, when appropriate.

Invest in resilient, ethical and equitable supply chains for medicinal products

### For the WHO:

Support Member States to increase equitable access to medicines and other health products for PLWRD

Provide guidance and support to help countries speed up the approval of medicines and other health products by relying on assessments already carried out by WHO or other trusted authorities

Provide technical support and guidance in establishing HTA appraisal processes

Provide guidance to help countries update standards and recommendations to improve access to medicines and other health products

Provide technical support for development, implementation and scale up of procurement strategies, such as pooled procurement mechanisms

### For International & National Partners

Promote research that includes diverse populations, measures outcomes and uses real-world data

Improve economic evidence by using reliable, repeatable methods taking into account uncertainty, healthcare costs and wider impact

Work to develop and implement procurement and supply approaches to improve availability, affordability, and quality of medicines and health products

Facilitate processes for patent pooling, voluntary licenses, and evidence for tiered pricing.

Engage relevant stakeholders, including PLWRD and their families, in efforts to improve access to affordable, safe, effective and quality medicines and other health products.

# AREA 2: DIAGNOSTICS, MEDICINES AND HEALTH PRODUCTS

28. Early detection is vital for the prevention, diagnosis, case management, monitoring and appropriate treatment and care of rare diseases. Early detection and diagnosis are useful even in the absence of treatment as it supports people living with a rare disease, their families and caregivers in accessing communities facing similar issues and learning about facilitating care and improving quality of life. Equitable approaches to early detection and diagnosis of rare diseases should ensure timely access to appropriate, evidence-informed screening and diagnostic services, including for people in rural and underserved settings.

29. Rare disease treatments include a range of modalities, from managing symptoms to advanced and potentially curative therapies, including gene and cell therapies. Pharmaceutical treatment options can be designated as orphan medicinal products when they are authorized for the diagnosis, prevention or treatment of rare diseases.

30. Preventive public health measures are important to decrease the frequency of certain rare diseases through the mitigation of risk factors or the reinforcement of protective factors while respecting regional, national, and local context, as well as individual autonomy and informed choice.

31. Health systems should be strengthened around primary health care, with clear links to specialized services, to connect early detection and diagnosis with timely access to appropriate treatment and care, including medicines, assistive technologies and medical devices. Where appropriate, precision medicine can use relevant data to tailor prevention, diagnosis and treatment to individual needs, reducing avoidable burdens and improving health and quality of life.

32. Involvement of people with lived experience, their families, caregivers, and representative organizations, should be encouraged throughout the entire process of research and development of new diagnostics, medicines and health products, prioritization through public policies and monitoring of interventions for impact. Their meaningful participation can help identify specific approaches that are needed to address differences in prevalence, disease presentation, diagnosis, treatment, access and outcomes.

## 2.1 DIAGNOSTICS

33. Early detection and diagnosis of rare diseases can prevent the onset of disease symptoms or delay disease progression, thereby reducing mortality and morbidity, improving quality of life, and conferring significant benefits on individuals, their families, and caregivers and society.

34. Rare disease identification can be strengthened through appropriate approaches, such as expanded newborn screening, as well as equitable access to accurate diagnostic tools and

appropriate counselling and post-diagnostic support. Accurate and timely rare disease diagnosis requires trained personnel and adequate laboratory capacity and equipment as relevant.

35. New diagnostic methods such as genomic and molecular analysis, including next-generation sequencing, proteomics, and detailed clinical phenotyping, have accelerated the identification of previously unknown rare conditions, improved understanding of disease mechanisms and contributed to the emergence of precision medicine. These technologies thus offer new opportunities to translate scientific discovery into equitable, needs-based and data-driven healthcare. Many of the newer diagnostic methods are however complex, expensive and demand special expertise. Expansion of diagnostic services for rare diseases should be approached in an equitable fashion, prioritizing accurate and timely detection and diagnosis where the impact is greatest, and with a balanced approach towards the best use of limited health system resources.

## PROPOSED ACTIONS

36. *Member States*
    a. Invest in access to timely, accurate and equitable diagnosis by training primary health and care workforce, enhancing the availability, access and use of relevant basic diagnostics tools, and improving clinical and laboratory infrastructure.
    b. Expand access to specialized diagnostic expertise, such as multidisciplinary specialist workforce, standardized genomic and phenotypic diagnostic services, supported by validated protocols, integration of precision medicine into the health system, interoperable databases, and collaborations with regional diagnostic centres.
    c. Implement evidence-based strategies, such as universal newborn screening programmes, taking into consideration ethical aspects, equity, cost-effectiveness, and ensuring it is done in a context of a comprehensive continuum of care, with clear and sustainable pathways for confirmatory diagnosis, referral, treatment, rehabilitation, family support and short- and long-term follow-up. Adopt a phased, health-system-based approach that starts with a small, feasible set of priority conditions, and expands as capacity grows.
    d. Create strategies to ensure greater availability and access to genetic counselling and other post-diagnostic support to enable informed decision-making, interpretation of results, and appropriate follow-up for individuals and families.

37. *Secretariat*
    a. Support the exchange of knowledge, experiences and good practices on public health measures, newborn screening, early diagnosis and post-diagnostic support including genetic counselling.
    b. Update technical guidance to include diagnostic tools relevant for rare diseases, through mechanisms such as the WHO Model List of Essential In Vitro Diagnostics.[5]

[5] WHO Model List of Essential In Vitro Diagnostics [website]. World Health Organization; 2025 (https://edl.who-healthtechnologies.org/).

c. Provide technical guidance around workforce training and establishing regulatory frameworks to ensure adherence with quality, safety, and ethical standards across screening and diagnostic programmes.

38. *International and national partners*
    a. Work with all relevant stakeholders to promote efforts for improving access to affordable, safe, effective, and high-quality diagnostics.
    b. Partner with health care professionals, academia, industry and people living with a rare disease, their families, and caregivers, to create and enhance coordinated platforms for diagnosis.

## 2.2 MEDICINES AND HEALTH PRODUCTS

39. The development and access of medicines and other health products for rare diseases, including medical devices, assistive products, and procedures, remain limited. Incomplete understanding of rare diseases, due to small, dispersed populations, leads to limited evidence and uncertainties about the effectiveness of potential new interventions. The heterogeneity of conditions and the severely disabling or life-threatening nature of many of these conditions compound these challenges.

40. Regulators and health technology assessors face challenges in interpreting limited clinical and quality of life evidence and use models that do not fully characterize the progression of the disease or the benefit of treatment.

41. Even when treatments and other technologies are available and approved, high costs lead to delayed, inconsistent and inequitable access within and among countries. Improving availability, affordability and timely access to medicines and health products, including through repurposing and the safe and evidence-informed off-label use of existing medicines, may require innovative regulatory strategies, health technology assessment (HTA) processes, and procurement mechanisms. Clear pathways for accessing home modifications and assistive devices, with funding mechanisms or subsidies to address affordability, should also be established.

## PROPOSED ACTIONS

42. *Member States*
    a. Promote dialogue on evidence and collaboration among developers, regulators, HTA bodies, payers and other stakeholders to identify data gaps and agree on appropriate approaches.
    b. Promote access to medicines and health products through regulatory mechanisms, including measures such as preferential and reliance-based registration and accelerated marketing authorization; cost and availability strategies, including appropriate use of generics, biosimilars and repurposed medicines; and robust quality-assurance measures.
    c. Encourage HTA evaluation frameworks that consider the circumstances of each rare condition, including disease progression, severity, and population affected.

d. Align national treatment guidelines and medicines and health products lists to include interventions relevant to rare diseases, informed by guidance such as WHO standard treatment guidelines and model lists of essential medicines and priority health products.
e. Facilitate regional or multi-country procurement mechanisms, including joint negotiation, pooled or centralized procurement, global financing, donations and managed entry agreements linked to evidence generation and outcomes to improve affordability and availability, and stockpiling, when appropriate.
f. Invest in resilient, ethical and equitable supply chains for medicinal products, including controlled substances, through demand forecasting, reliable distribution, and workforce capacity, based on need and context.

43. *Secretariat*

a. Support Member States to increase equitable access to medicines and other health products for people living with a rare disease, including through facilitating knowledge exchange on evidence-based, context-specific regulatory guidance, cost and availability strategies, and quality-assurance measures.
b. Provide technical guidance and support for the use of facilitated market entrance mechanisms, such as the WHO Collaborative Registration Procedure to enable national regulators to rely on WHO prequalification or assessments by WHO Listed Authorities and other reliance mechanisms.
c. Provide technical support and guidance to Member States in establishing HTA appraisal processes for rare disease treatments that consider the full dimensions of value, fair pricing models, and development of health financing strategies to support access to medicines and health products for rare diseases.
d. Support and provide technical guidance for updating normative guidance that facilitate access to products relevant to rare diseases, such as national essential medicines lists, the WHO Model List of Essential Medicines,[6] WHO Model List of Essential Medicines for Children,[7] WHO disease-specific Standard Treatment Guidelines, WHO Lists of Priority Medical Devices,[8] and the WHO Priority Assistive Products List.[9]
e. Provide technical support to Member states for the development, implementation and scale up of procurement strategies, such as pooled procurement mechanisms.

44. *International and national partners*

a. Strengthen evidence generation for rare diseases through rigorous and innovative study designs, equitable representation of populations, comprehensive outcome measurement, long-term follow-up and appropriate use of real-world evidence.
b. Improve the transparency, relevance and comprehensiveness of economic evidence through robust, reproducible models that reflect uncertainty, health-

[6] The selection and use of essential medicines, 2025: WHO Model List of Essential Medicines, 24th list. Geneva: World Health Organization; 2025 (https://doi.org/10.2471/B09474). Licence: CC BY-NC-SA 3.0 IGO.
[7] The selection and use of essential medicines, 2025: WHO Model List of Essential Medicines for Children, 10th list. Geneva: World Health Organization; 2025 (https://doi.org/10.2471/B09475). Licence: CC BY-NC-SA 3.0 IGO.
[8] WHO lists of priority medical devices [website]. World Health Organization; 2026 (https://www.who.int/activities/prioritizing-medical-devices).
[9] World Health Organization, USAID International Disability Alliance. Priority assistive products list: improving access to assistive technology for everyone, everywhere (WHO/EMP/PHI/2016.01). Geneva: World Health Organization; 2016 (https://iris.who.int/handle/10665/207694). Licence: CC BY-NC-SA 3.0 IGO.

system resource use and the wider impacts on people living with a rare disease, their families and caregivers.

c. Collaborate with health authorities, manufacturers and other regional and international partners to develop and implement procurement and supply approaches to improve the availability, affordability, quality and appropriate use of medicines and other health products for rare diseases.
d. Facilitate processes for patent pooling, voluntary licenses, and evidence for tiered pricing.
e. Engage relevant stakeholders, including individuals living with a rare disease, their families, and caregivers, and the organizations that represent them, in activities to promote efforts for improving access to affordable, safe, effective and quality medicines and other health products.