Action Area 4: Research and Innovation



Resarch and Innovation

FOCUS OF THIS SECTION:

Sustained investment in research to advance prevention, diagnosis, treatment and care for rare diseases

Innovation to help address unmet needs, improve access, and strengthen health system capacity, across the continuum of care

The meaningful engagement of PLWRD and their families in research

4.1 RESEARCH CAPACITY BUILDING

Countries need to invest in research infrastructure, develop research skills and encourage collaboration between researchers, health professionals, people living with rare diseases, industry and other stakeholders. This is particularly important in low- and middle-income countries.

A shared global research agenda can help coordinate efforts, avoid duplication and unnecessary costs, and accelerate progress in preventing, diagnosing, treating and caring for rare diseases.

PROPOSED ACTIONS

For Member States:

Encourage collaboration among policymakers, health and research authorities, academic institutions, clinicians, patient organizations and others to build a research agenda that addresses rare diseases

Build the rare disease research workforce through training, fellowships, mentorship, and research collaborations

Invest in research infrastructure and partnerships

Strengthen research governance, ethical standards, oversight, and meaningful participation of PLWRD

For the WHO:

Support Member States in setting priorities for rare diseases.

Engage WHO collaborating centres, academic institutions, research organizations and alliances to strengthen the capacity for research on rare diseases, with particular attention to the needs of low- and middle-income countries

For International & National Partners

Support research in rare diseases and highlight the importance of prioritizing funding for national research organizations

Participate in priority-setting exercises involving PLWRD and contribute to the dissemination of research findings to policymakers and the public

Support, establish and strengthen multidisciplinary research consortia to advance collaborative rare disease research across disciplines and countries.

Promote research in LMICs by strengthening capacity and international collaboration

Action Area 4: Research and Innovation

4.2 INVESTMENT IN RESEARCH

Rare disease research should cover all stages, from understanding the causes and biology of diseases to developing and testing treatments and improving health services and care.

Research should include well-designed clinical trials that assess the safety and effectiveness of treatments and their impact on patients' lives, while taking into account the small and dispersed populations affected by each disease.

It should address the medical, social and psychological needs of PLWRD and involve them in shaping and evaluating research. Research should be inclusive and equitable, with greater participation from LMICs.

PROPOSED ACTIONS

For Member States:

Increase and sustain investment in rare disease research, and leverage complementary funding

Create, renew, or update national incentive programmes that facilitate the development of research and innovations for rare diseases

Establish and support research networks to investigate the genetic causes of undiagnosed diseases, advance the analysis of genomic data, strengthen shared genomic databases and enable innovative designs for clinical trial.

Support research to assess rare disease care, identify gaps, and develop solutions

Conduct research into disparities that occur among the diverse subpopulations of people living with a rare disease and prioritize interventions that are responsive to their needs and address social determinants

For the WHO:

Encourage international coordination mechanisms to facilitate harmonized global research efforts in rare diseases and build a globally connected research community.

Provide guidance and technical support to Member States to ensure equitable, diverse and inclusive participation in research, particularly in low- and middle-income countries.

For International & National Partners

Engage the research community, academia, health professionals, policymakers and the private sector in mobilizing financial support for research in rare diseases to promote innovation while ensuring the equitable and affordable access of these products.

Support and conduct research to generate knowledge about barriers to the diagnosis, treatment, and care of rare disease and effective strategies to overcome such barriers.

AREA 4: RESEARCH AND INNOVATION

63. Sustained investment in biomedical, clinical, public health, implementation and translational research is essential to advance prevention, diagnosis, treatment and care for rare diseases.

64. The complexity surrounding rare disease research requires improved coordination in the research environment, with multistakeholder involvement, research consortia, and public–private partnerships, and allocation of sufficient resources. Cultivating an environment that fosters research collaborations is vital to reduce duplication, identify knowledge gaps, fast-track innovation and build capacity, especially in low-income settings.

65. Innovation is essential to improve outcomes and should extend across the continuum of care—from diagnostics, including genomic technologies, to medicines, advanced therapies, rehabilitation, disease monitoring and assessment tools, digital technologies and models of care. Evidence-based innovation can help address unmet needs, improve access and affordability, strengthen health system capacity and support more effective, person-centred and equitable care.

66. The meaningful engagement of people living with a rare disease, their families and caregivers to better support and guide the research and development of innovative solutions for rare diseases is a principal component of the research agenda.

4.1 RESEARCH CAPACITY BUILDING

67. Concerted action to build research infrastructure, strengthen human resources in research and development and increase collaboration among the research community, health professionals, people living with a rare disease and other relevant stakeholders, including industry, is needed to catalyse rare disease research and development, particularly in low- and middle-income countries.

68. Facilitating a global research agenda for rare diseases will increase the likelihood of effective progress towards better prevention, diagnosis, treatment and care while reducing redundancies and the duplication of research and costs.

PROPOSED ACTIONS

69. *Member States*
    a. Encourage collaboration between policymakers, health and research authorities, academic institutions, clinicians, organizations that represent people with lived experience, the private sector and civil society to build a research agenda that proactively addresses rare diseases.
    b. Build the rare disease research workforce through training, fellowships, scholarships, mentorship, and research collaborations in biomedical, clinical, implementation and translational research.

c. Strengthen institutional capacity for rare disease research by investing in infrastructure, equipment, supplies, data systems and partnerships.
d. Strengthen research governance including establishing ethical standards for responsible conduct, transparent oversight, and meaningful participation of people living with a rare disease, their families, and caregivers.

70. *Secretariat*
    a. Support Member States through global research prioritization and agenda-setting for rare diseases.
    b. Engage WHO collaborating centres, academic institutions, research organizations and alliances to strengthen the capacity for research on rare diseases, with particular attention to the needs of low- and middle-income countries.

71. *International and national partners*
    a. Support and provide evidence on the need for research in rare diseases and highlight the importance of prioritizing funding for national research organizations.
    b. Participate in priority-setting exercises, involving people living with a rare disease, their families, caregivers, and contribute to the dissemination of research findings in user-friendly language to policymakers and the public.
    c. Support, establish and strengthen multidisciplinary research consortia to coordinate research, share expertise and resources, and advance collaborative rare disease research across disciplines and countries.
    d. Address the current concentration of research in high-income countries through context-specific strategies in low- and middle-income countries, strengthened research capacity and international collaboration.

4.2 INVESTMENT IN RESEARCH

72. Rare disease research should span the continuum from basic and preclinical studies to clinical trials, translational research, and health services and implementation research to advance understanding and improve diagnosis, treatment, care and policy.

73. Basic research into rare diseases is important to understand biological processes and their pathomechanisms given the wide range of variability among rare disease conditions.

74. Clinical trials are essential to generate robust evidence on safety, efficacy, patient-relevant outcomes and longer-term impacts of interventions for rare diseases, while requiring innovative, collaborative and patient-centred designs suited to small and dispersed populations.

75. Health services research is critical to understand the medical, social and psychosocial needs of people living with rare diseases and to evaluate and improve care, services and interventions in partnership with people with lived experience and their organizations.

76. Despite the inherent challenges of working with small, dispersed populations, research and innovation in rare diseases should be equity-oriented, inclusive and diverse, with meaningful engagement of people living with a rare disease, their families and caregivers, and improved representation of low- and middle-income countries in all research types.

PROPOSED ACTIONS

77. *Member States*
    a. Increase and sustain investment in rare disease research programmes across the research continuum while leveraging complementary funding from philanthropic, private-sector and international partners.
    b. Create, renew, or update national incentive programmes that facilitate the development of research and innovations for rare diseases. Incentive mechanisms can include but are not limited to orphan designation, tax credits and fee reductions, market exclusivity, advance markets commitments, and cost-sharing.
    c. Establish and support national and international research networks to investigate the genetic causes of undiagnosed rare diseases, advance the analysis of genomic data, strengthen shared genomic databases and enable collaborative and innovative designs for clinical trials.
    d. Support multidisciplinary health services research, including within national centres of expertise, to assess rare disease care, identify service gaps, and develop context-specific solutions.
    e. Conduct research into disparities that occur among the diverse subpopulations of people living with a rare disease and prioritize interventions that are responsive to their needs and address social determinants.

78. *Secretariat*
    a. Encourage international coordination mechanisms to facilitate harmonized global research efforts in rare diseases and build a globally connected research community.
    b. Provide guidance and technical support to Member States to ensure equitable, diverse and inclusive participation in research, particularly in low- and middle-income countries.

79. *International and national partners*
    a. Engage the research community, academia, health professionals, policymakers and the private sector in mobilizing financial support for research in rare diseases to promote innovation while ensuring the equitable and affordable access of these products.
    b. Support and conduct research to generate knowledge about barriers to the diagnosis, treatment, and care of rare disease and effective strategies to overcome such barriers.