RARE DISEASES INTERNATIONAL

MARCH 2025

RARE DISEASE EXPERTISE, CENTERS & NETWORKS

Results of the Public Survey



REPORT

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Acknowledgement

EXECUTIVE SUMMARY

Rare Diseases International (RDI) conducted a public survey to gather information on rare disease expertise, centers, and networks. A total of 228 valid responses were received from 63 countries across all six WHO regions.

RARE DISEASE EXPERTISE

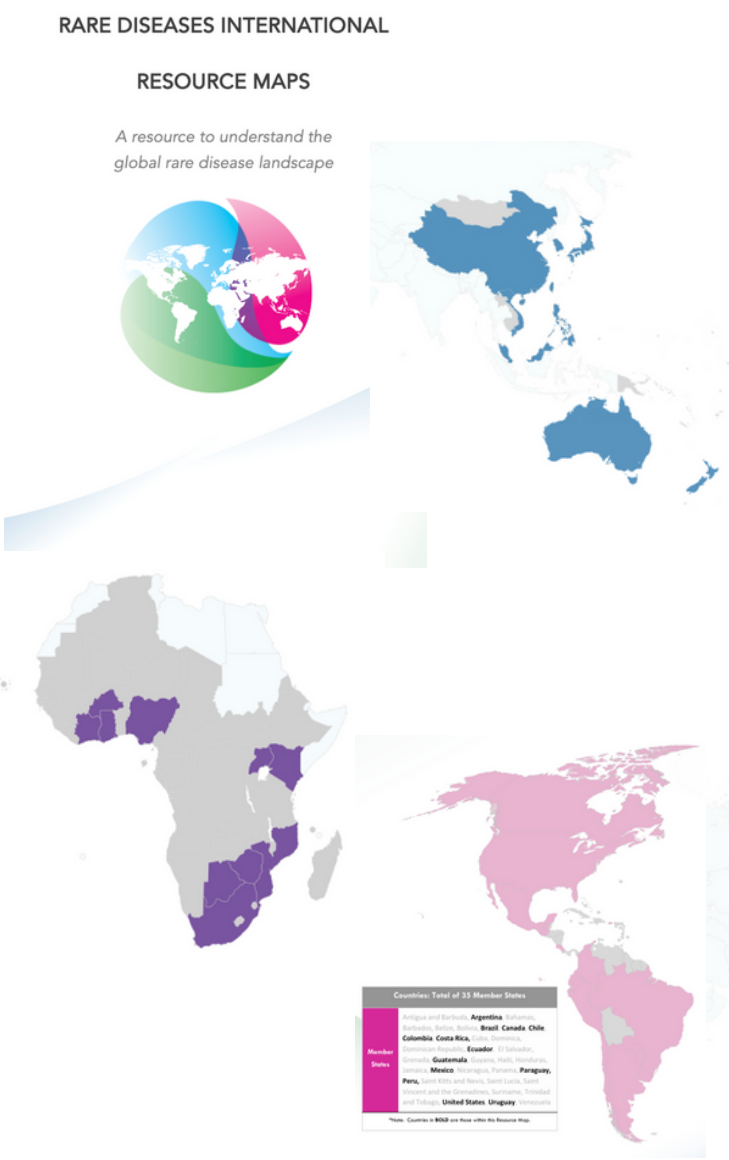
- **84% of respondents confirmed the presence of rare disease expertise in their country.**
- **73% of respondents reported that experts collaborate in the country, either informally (42%) or through formal networks (31%).**
- **59% of experts are engaged in international collaborations.**
- **46% of experts are involved in cross-border clinical services, particularly in diagnostics and treatment.**
- **About 60% of respondents indicated that there is some form of structured organization of expertise (specialized units/departments or RD centers).**

RARE DISEASE CENTERS

- **67% of centers are recognized in some form at the national level.**
- **Main criteria for recognition: expertise in specific rare diseases, availability of diagnostic/treatment facilities, research capacity, patient volume and presence of multidisciplinary teams.**
- **33% of centers provide or receive cross-border clinical services.**
- **These services are managed through: international networks, ad-hoc or informal arrangements or bilateral/multilateral agreements.**

INTRODUCTION

Over the past two years, Rare Diseases International (RDI) gathered data on rare disease landscape through conversations with patient representatives and medical experts spanning the six World Health Organization (WHO) regions. This work culminated in the publication of the first version of the Resource Maps in March 2024. These Resource Maps provide an initial overview of the rare disease landscape across various countries, concentrating on three critical areas: the policy landscape, healthcare systems, and patient organizations. This initial data collection serves as a tool to better understand the existing resources in the regions and identify gaps that need to be addressed.



In autumn 2024, RDI conducted a survey to update the existing Resource Maps and to identify centers and networks dedicated to rare diseases. The data collected is a valuable resource for the rare diseases community and is instrumental in informing the implementation of the Global Network for Rare Diseases (GNRD). Identifying rare disease centers and understanding their characteristics will facilitate and encourage future collaborations between centers in different countries.

METHODOLOGY

Objectives

The primary aim of this survey is to map the landscape of rare disease expertise, identify centers and networks dedicated to rare diseases in different countries and regions. This data will inform the GNRD by providing an overview of available resources and capabilities and understanding how to build on them for future collaborations.

The objectives are:

- To build on and update the information that has already been collected
- To map centers for rare diseases and identify their characteristics
- To understand the criteria used to recognize these centers, if any
- To explore the types of collaboration and referral systems established between these centers

Survey design and dissemination

RDI developed the survey in collaboration with a small group of experts and Regional Members. The survey was designed to capture information about centers and networks for rare diseases, and included a set of multiple-choice questions, as well as sections for collecting references and citations.
The target audience for the survey included RDI members, participants of the GNRD Multistakeholder Forum, and any individuals with knowledge of centers for rare diseases. The survey was available in English and Spanish, and was disseminated through the following channels:

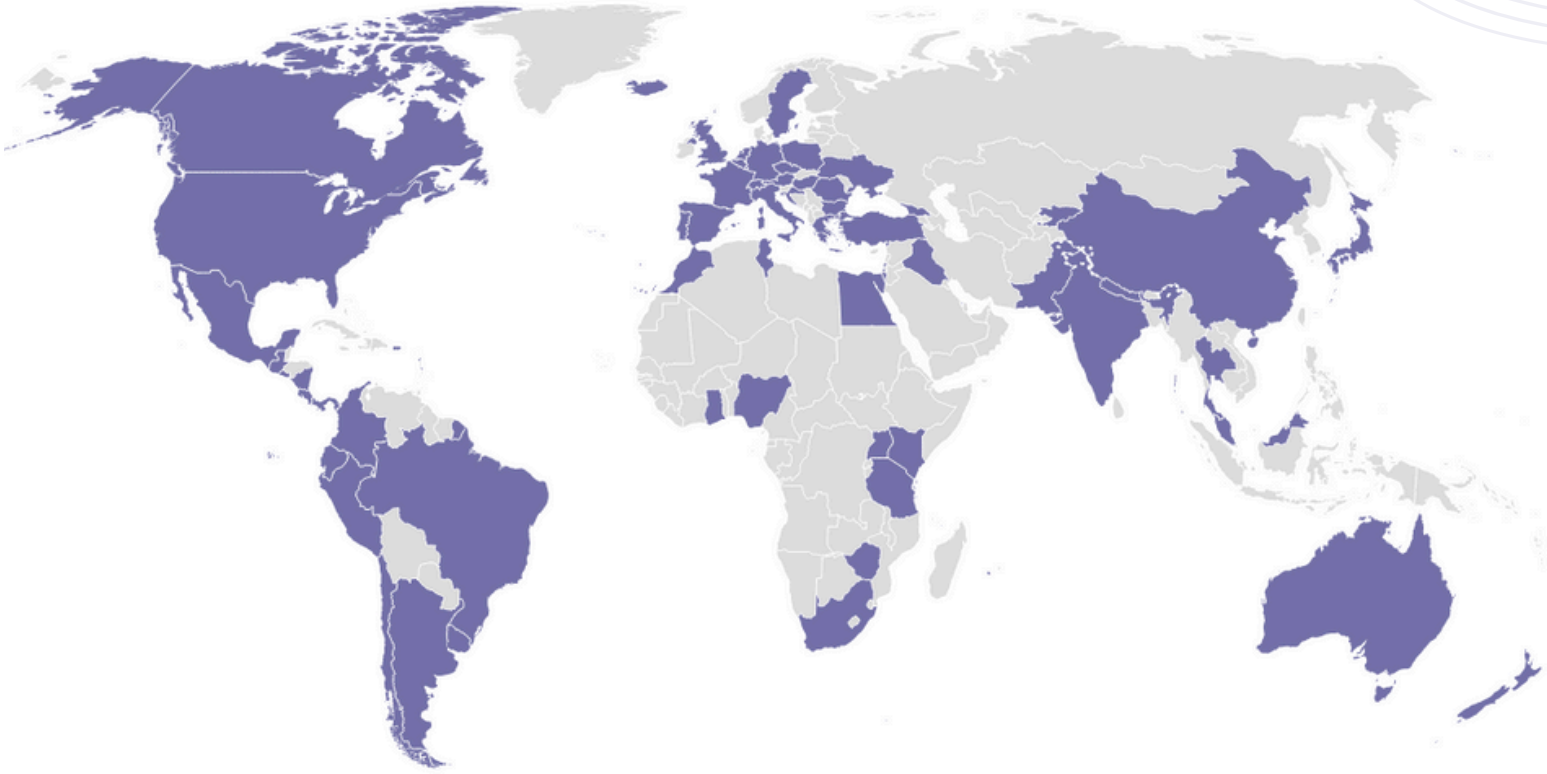
- Emails specifically targeting RDI Regional Members and participants of the GNRD Multistakeholder Forum
- RDI's social media channels and newsletter
- Follow-up reminders to ensure maximum participation

SURVEY SAMPLE

The survey collected a total of 232 responses. Of these surveys, 3 were empty, and 1 lacked country information, leaving 228 valid responses for analysis.

Geographical distribution of the sample

Countries where one or more surveys were collected are shown in purple.



228 RESPONSES

63 COUNTRIES

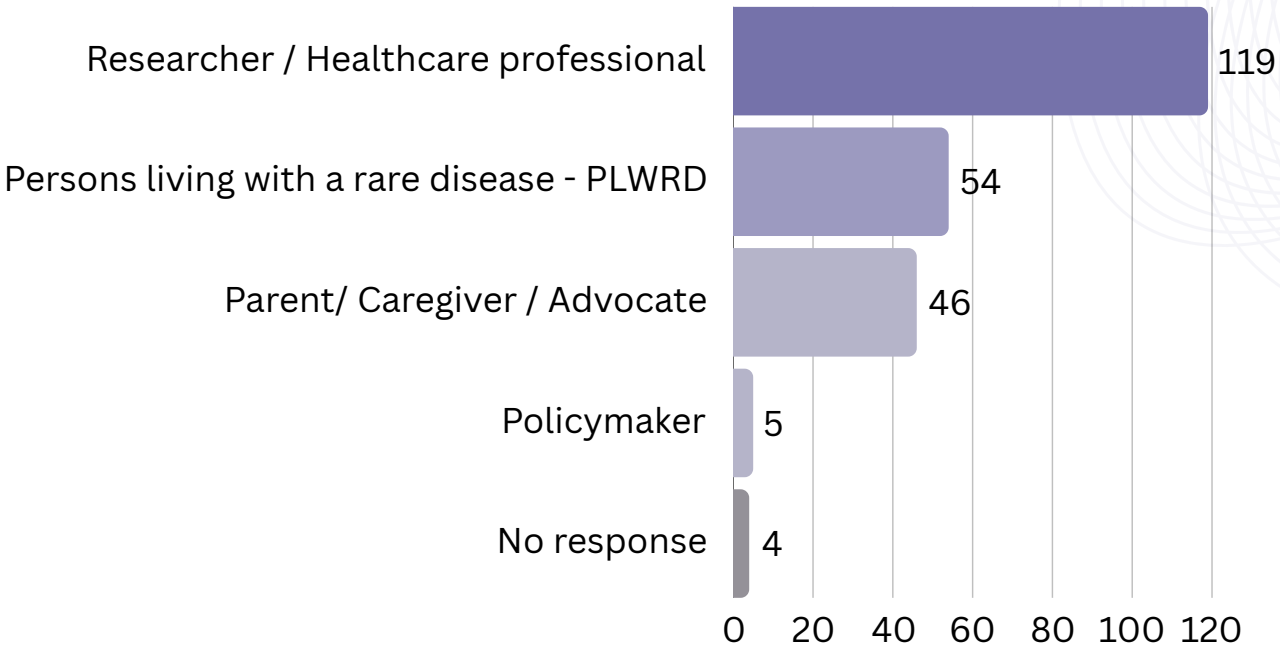
List of Regions and Countries

Region	Survey filled	Countries represented
AFR - African Region	68	7
AMR - Region of the Americas	52	15
EMR - Eastern Mediterranean Region	13	6
EUR - European Region	61	26
SEAR - South East Asian Region	13	3
WPR - Western Pacific Region	21	6

The surveys gathered responses from 63 countries worldwide, including:

Argentina
Australia
Austria
Bahrain
Belgium
Brazil
Bulgaria
Canada
Chile
China
Colombia
Costa Rica
Croatia
Czechia
Ecuador
Egypt
El Salvador
France
Georgia
Germany
Ghana
Greece
Guatemala
Hungary
Iceland
India
Iraq
Israel
Italy
Japan
Kenya
Kyrgyzstan
Luxemburg
Malaysia
Malta
Mexico
Morocco
Nepal
Netherlands
New Zealand
Nicaragua
Nigeria
Pakistan
Panama
Peru
Poland
Portugal
Romania
Singapore
South Africa
Spain
Sweden
Switzerland
Tanzania
Thailand
Tunisia
Turkiye
Uganda
UK
Ukraine
Uruguay
USA
Zimbabwe

Participant Categories



The survey participants represented various categories, with researchers and healthcare professionals making up 52% (119 respondents). Persons living with a rare disease (PLWRD) accounted for 24% (54 respondents), followed by parent/caregiver advocates at 20% (46 respondents). Policymakers represented a small portion of respondents, comprising just 2% (5 respondents).

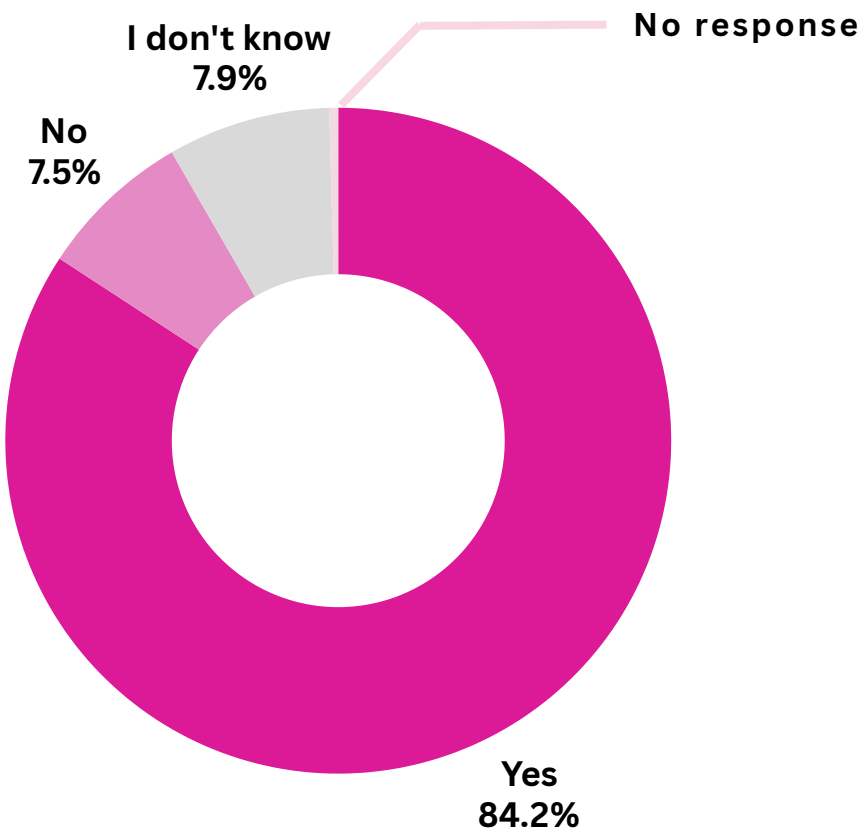
RESULTS - SECTION ONE

The first section of the results focuses on the expertise available for rare diseases within each country. It explores whether rare disease experts are present, their areas of specialization, and how they are organized or collaborate to provide care and support for PLWRD. Participants were invited to share insights into the structure, roles, and working practices of these experts to better understand the current landscape of expertise in rare diseases.

AVAILABILITY OF EXPERTISE IN THE COUNTRY

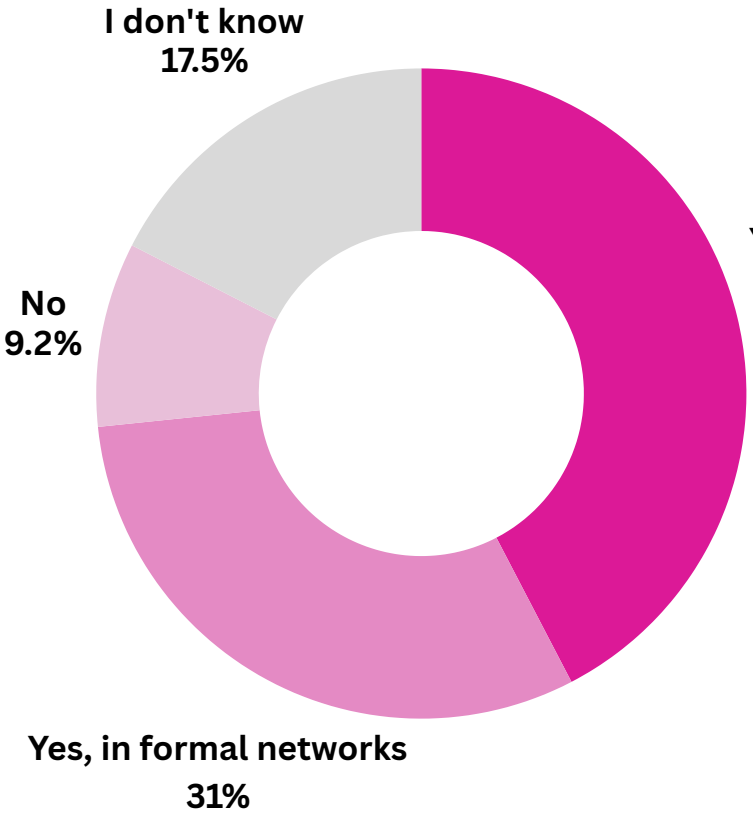
Are there rare disease experts in your country?

Most respondents (84% - 192 individuals) indicated that expertise in rare diseases is available in their country. A small proportion, 7.5% (17 respondents), reported that no expertise is available, while 7.9% (18 respondents) were unsure.



COLLABORATION BETWEEN EXPERTS IN THE COUNTRY

Are Rare Disease Experts collaborating in some way in your country?

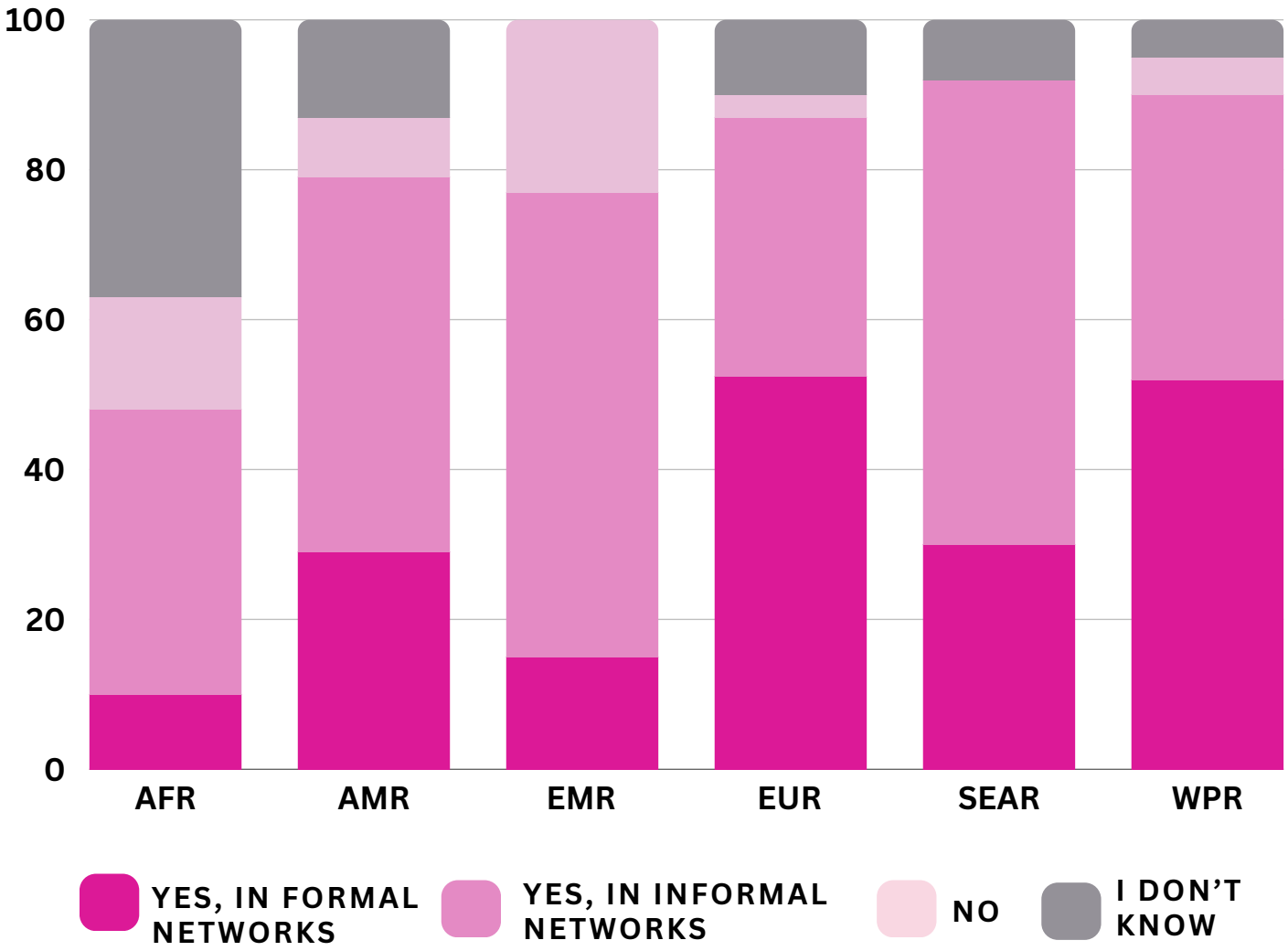


Merging formal and informal collaboration, 73% of respondents affirmed that experts collaborate in some way.

When asked about collaboration between experts in their countries, 42% of respondents (97 individuals) reported informal collaborations, and 31% (71 individuals) noted formal networks. However, 9% of respondents stated that there is no collaboration. Similarly, around 18% of respondents answered "I don't know".

COLLABORATION TYPE BY REGION

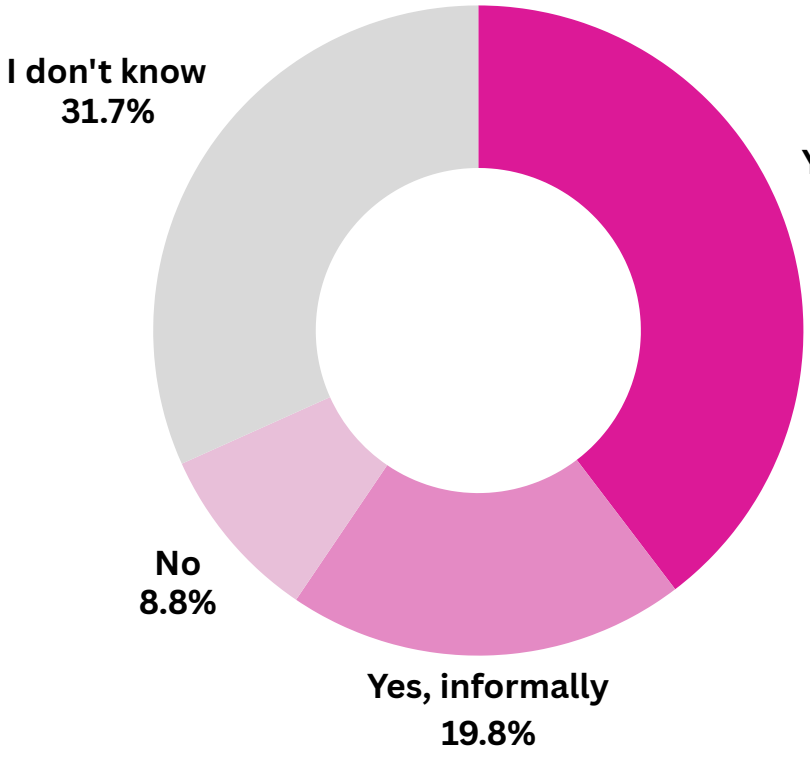
Breaking down collaboration types by region, the responses reveal distinct patterns: formal networks are predominant in EUR and WPR (52% in both regions), while informal networks dominate in AFR, AMR, EMR, and SEAR, with percentages ranging from 50% to 62%.



Formal type of collaboration is predominant in EUR and WPR, informal type of collaboration is predominant in AFR, AMR,EMR and SEAR.

PARTICIPATION IN INTERNATIONAL NETWORKS

Do experts for rare diseases in your country participate in any international collaboration/network related to rare diseases?

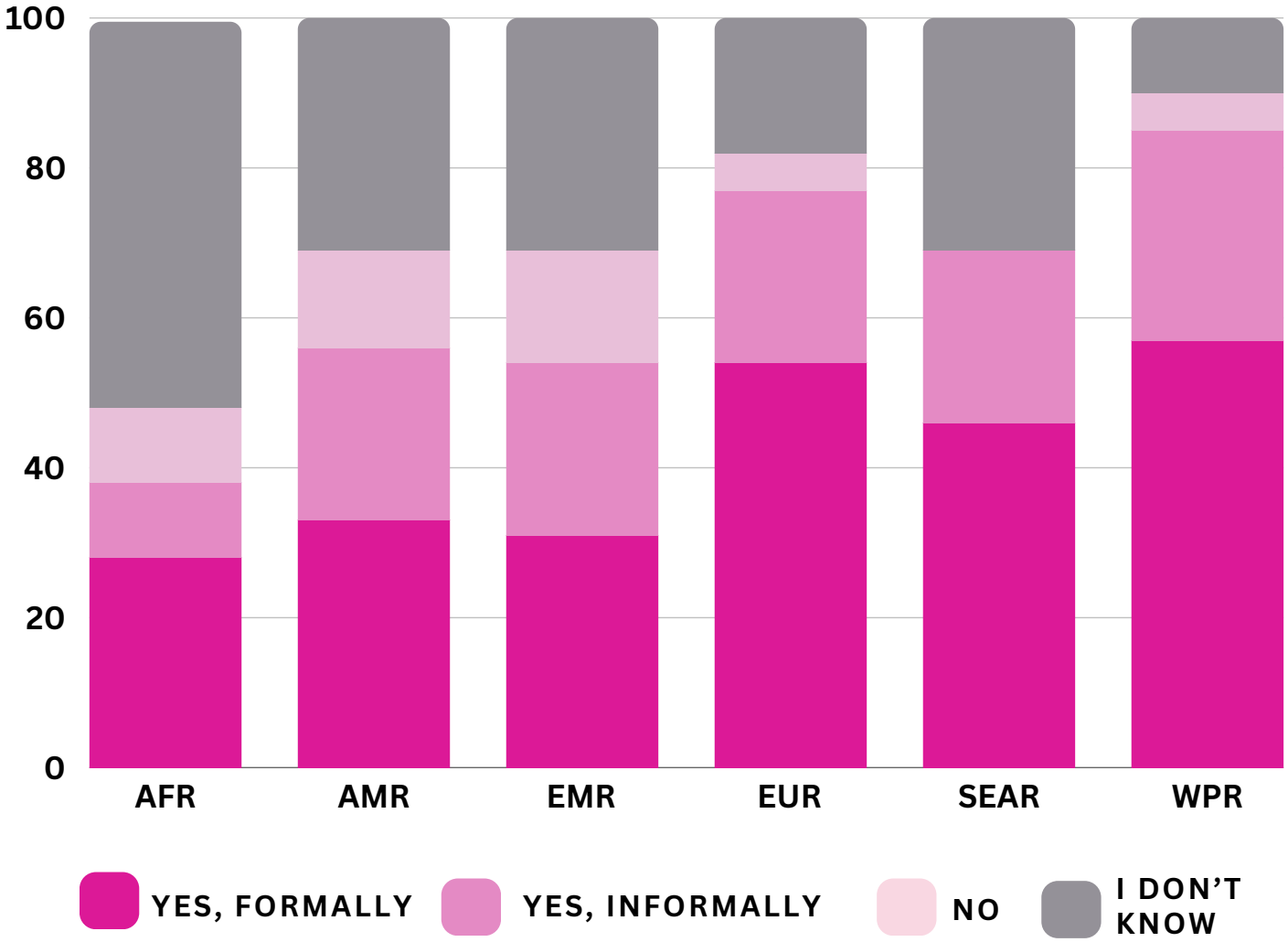


Around 59% of experts engage in international collaboration or networks, either formally or informally

The survey investigated whether experts are involved in international collaborations or networks focused on rare diseases. 39.6% of respondents (90 individuals) reported participating in formal international collaborations, while 19.8% (45 respondents) noted involvement in informal networks. However, 31.7% (72 respondents) were unsure about such collaborations, and 8.8% (20 respondents) indicated no participation in any international networks.

INTERNATIONAL COLLABORATION BY REGION

Breaking down the results by region, formal collaborations were reported with 28% in AFR, 33% in AMR, 31% in EMR, 53% in EUR, 46% in SEAR, and 57% in WPR, while informal collaborations were indicated for the 10% in AFR, 23% in AMR, 23% in EMR, 23% in EUR, 23% in SEAR, 29% in WPR.



Formal collaborations were the most commonly reported. However, a substantial portion of collaboration happens informally

MOST COMMONLY REPORTED NETWORKS & COLLABORATIONS

Participants were asked to specify which international networks or collaborations the experts in their countries may be involved in. The most commonly reported ones are listed here.

IRDiRC	International Rare Diseases Research Consortium
UDNI	Undiagnosed Diseases Network International
ERNs	European Reference Networks
ERDERA	European Rare Diseases Research Alliance
JARDIN	Joint Action of integration of ERNs into National Health Systems
GNNRD	Global Nursing Network for Rare Diseases
CEPCAL	Colaborativa para Enfermedades Poco Frecuentes en el Caribe y América Latina
APARDO	Asia Pacific Alliance of rare diseases organizations
CENCAM	Consortium for the Epidemic of Nephropathy in Central America and Mexico
IRCI	International Rare Cancer Initiative
ICORD	International Collaboration on Rare Diseases and Orphan Drugs
RIBERSER	Ibero-American Network of Health Experts on Rare Diseases

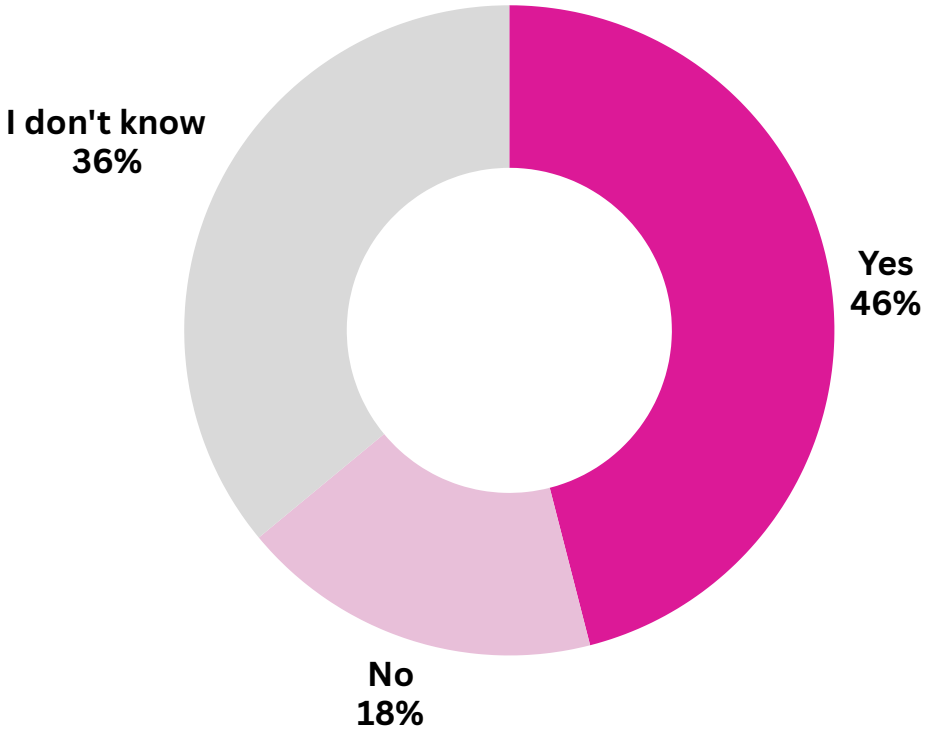


CROSS-BORDER CLINICAL SERVICES FOR RARE DISEASES

“Clinical service" refers to any procedure involved in diagnosing or managing rare diseases. This includes, but is not limited to, consultations, medical examinations, diagnostic tests, treatment plans, and follow-up care.

The survey asked respondents whether rare disease experts in their country are involved in providing or receiving cross-border clinical services for rare diseases. 46% of respondents indicated "Yes," demonstrating active international collaboration in clinical services for rare diseases. A smaller proportion (18%) responded "No," while 36% were uncertain about such involvement.

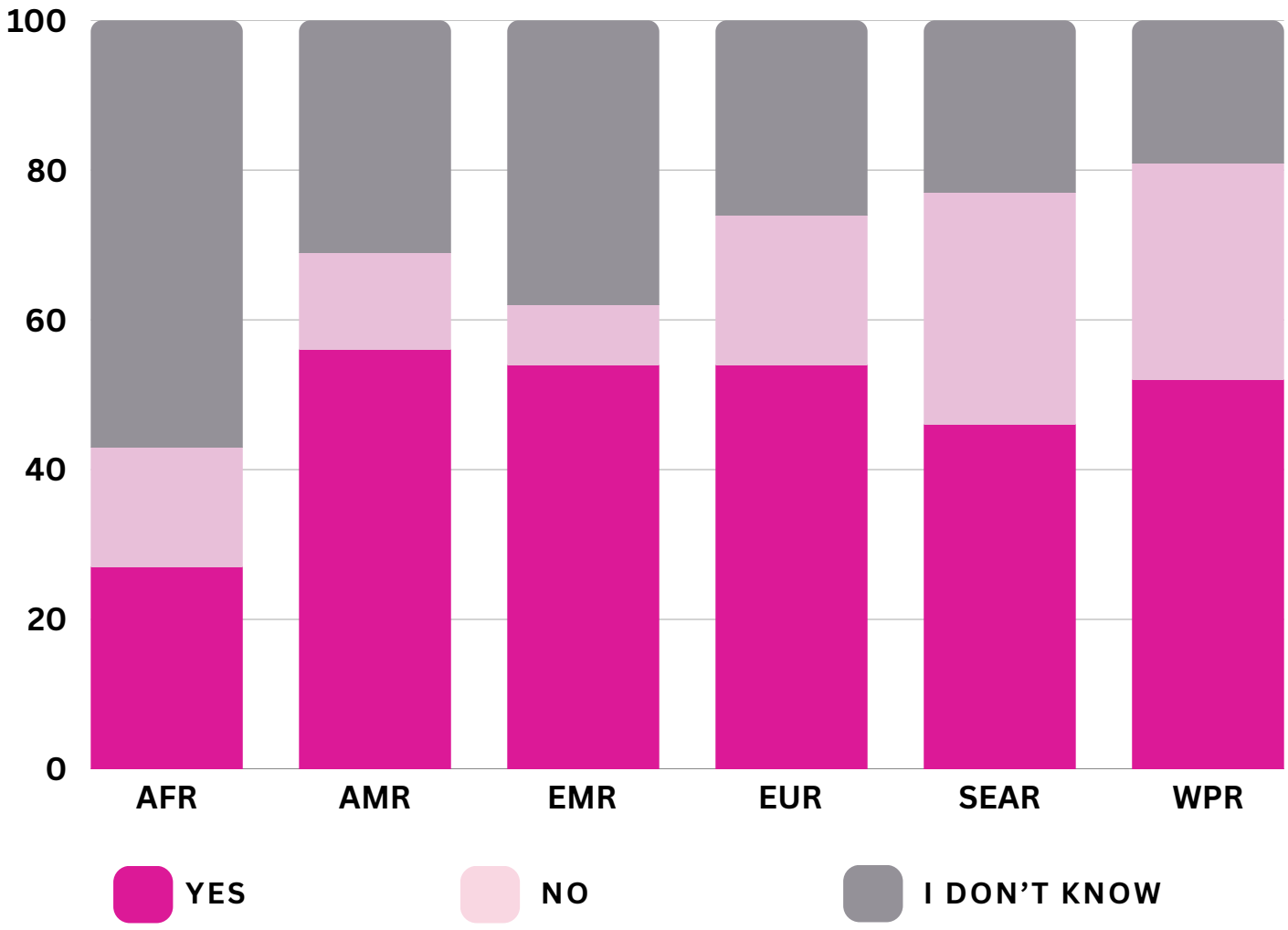
Are rare disease experts in your country involved in providing or receiving cross-border services for RD?



Almost half of respondents confirmed that experts provide or receive cross border clinical services for RD

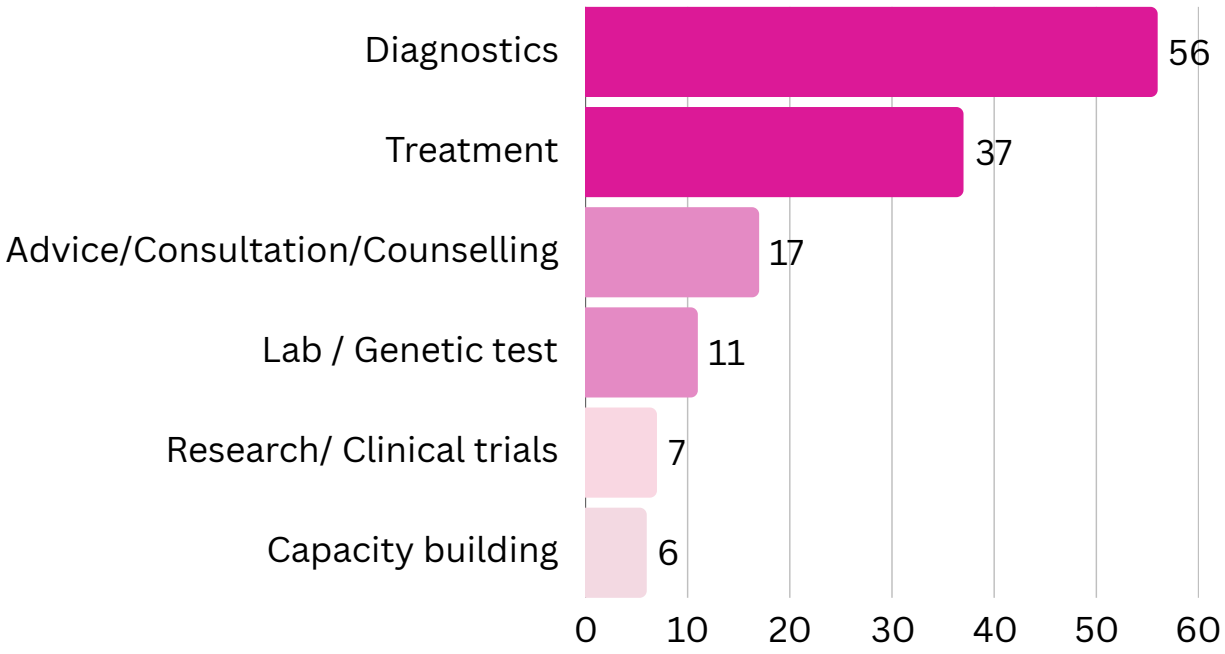
CROSS-BORDER CLINICAL SERVICES BY REGION

Analyzing the responses by region, the participation in cross-border clinical services was reported in AMR for the 56% of respondents, in EUR and EMR 54%, in WPR 52% and SEAR 46%. However in Africa, the majority (57%) were unsure about cross-border clinical services collaborations.



CROSS-BORDER CLINICAL SERVICES FOR RARE DISEASES

In terms of the types of services provided or received across borders, diagnostics was identified as the most commonly involved service, followed by treatment services. Consultation and counseling were also significant contributors. Other services, including laboratory testing/genetic tests, research/clinical trials, and capacity building were less frequently mentioned, though they remain important components of cross-border efforts.



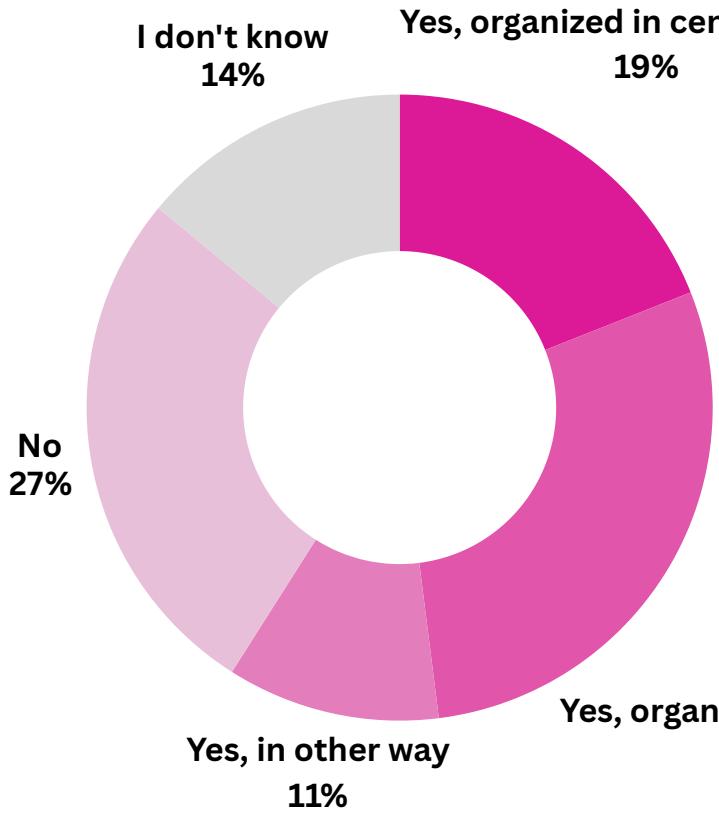
Diagnostics was identified as the most commonly involved service in cross-border clinical activities

EXPERTISE ORGANIZATION AT COUNTRY LEVEL

The survey asked participants about how rare disease expertise is organized within their countries. 19% of respondents reported that expertise is organized in centers dedicated to RD, while 29% indicated that expertise is provided through specialized departments or units within larger hospitals or structures. Additionally, 28% noted no organization of expertise for RD, and 14% were unsure. A smaller portion, 11% mentioned other organizational methods. These respondents specified that expertise is organized through private clinics, NGOs, or trusts/foundations.

The term "center" can have different meanings depending on the country, and the organization of expertise varies across regions. For this reason, the question was designed to be inclusive of all structures and forms of expertise organization.

Is rare disease expertise in your country organized in any specific way at the country level?



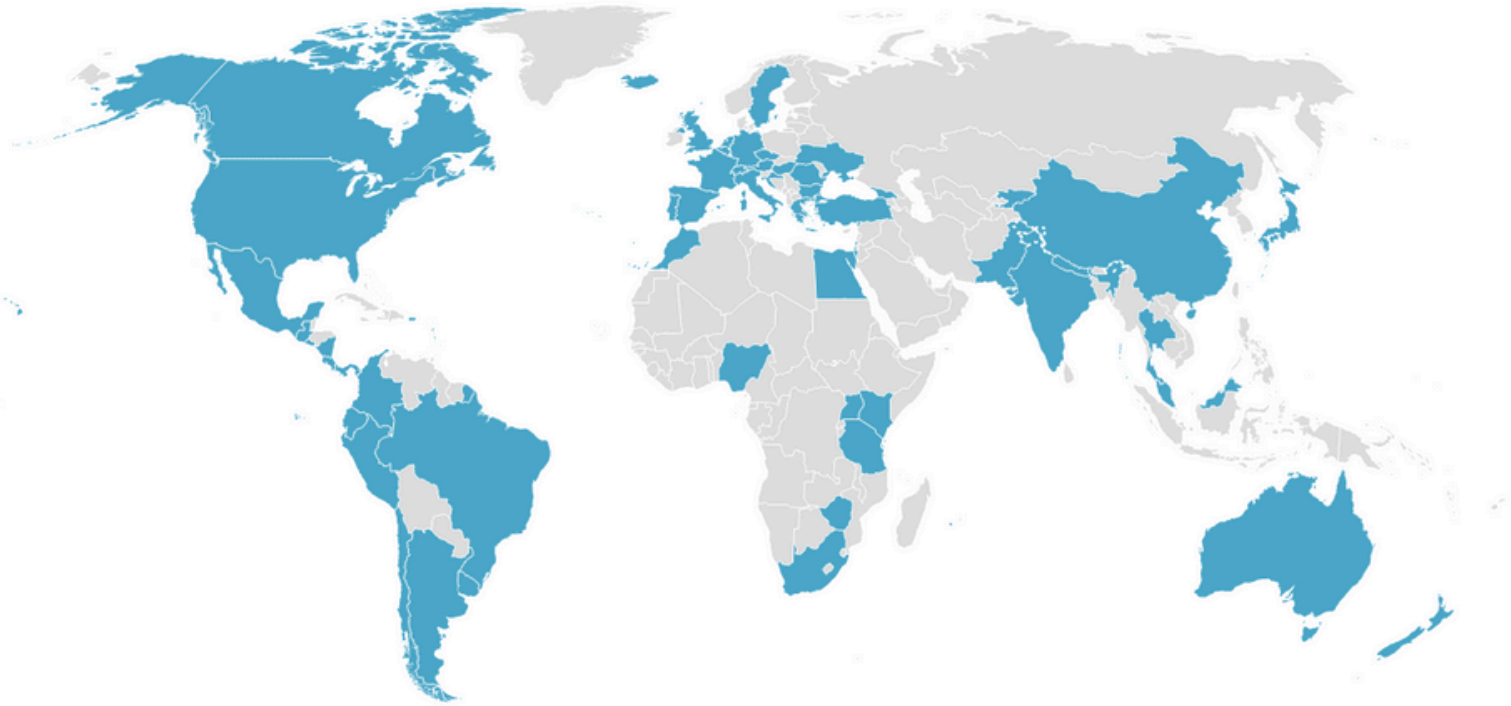
When merging all structured organization types, 59% of respondents reported some level of structured organization of rare disease expertise in their country.

Multiple selections were allowed

RESULTS - SECTION TWO

The second section of the results is dedicated to exploring the organization and characteristics of rare disease care-providing centers, hospitals, and specialized units or departments. Respondents were asked to share insights about the rare disease centers or units they are familiar with, including their recognition, structure, and role in delivering specialized care to PLWRD.
Out of 228 survey respondents, 181 participants named a center or department dedicated to rare diseases. From this point onward, the analysis focuses exclusively on these 181 responses. It is important to note that for certain questions within this section, fewer respondents were able to provide a response. These variations in response rates will be highlighted as needed throughout the analysis.

Geographical distribution of the centers

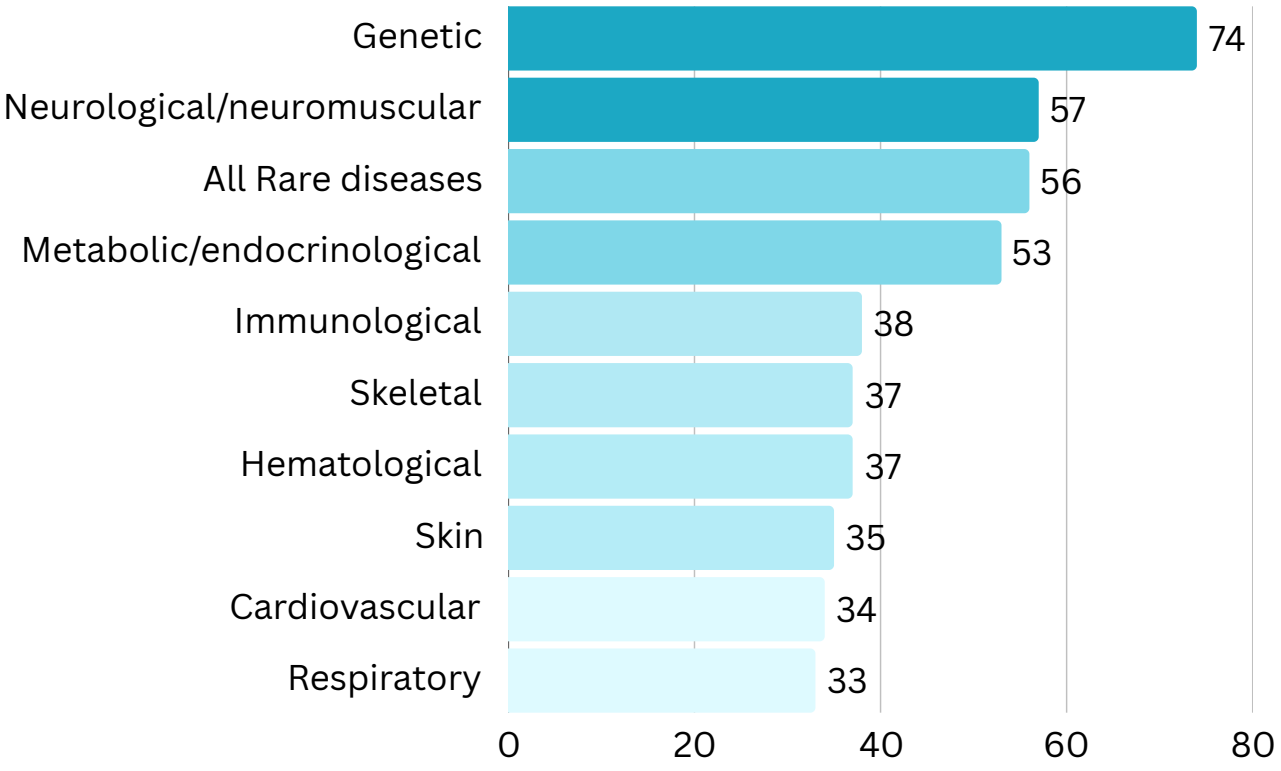


Countries where one or more centers, departments, or units for rare diseases were identified are shown in blue.

THERAPEUTIC AREAS ADDRESSED BY THE NOMINATED CENTERS

Among the 181 centers nominated by the participants, the diseases covered are distributed across various therapeutic areas.Multiple selections were allowed. Respiratory diseases are addressed by 33 centers, while cardiovascular diseases are covered by 34. Skin diseases are supported by 35 centers, and hematological and skeletal diseases are both addressed by 37 centers each. Immunological diseases are slightly more represented, with 38 centers covering this area. Metabolic and endocrinological diseases are supported by 53 centers, and neurological and neuromuscular diseases by 57 centers. Genetic diseases represent the most widely covered area, addressed by 74 centers. Additionally, 56 centers indicated their focus on all rare diseases.

What types of rare diseases are covered by the center/specialized unit?

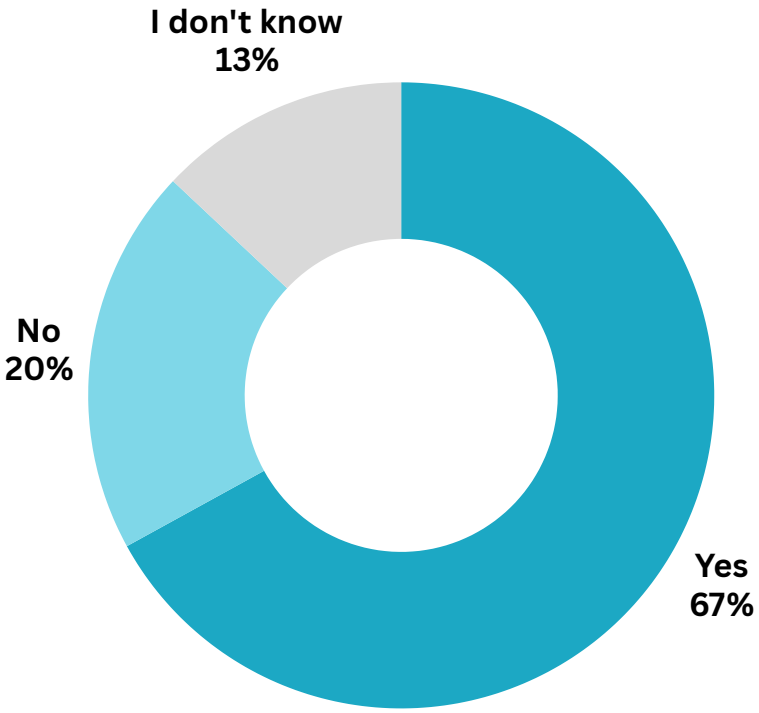


Multiple selections were allowed

RARE DISEASE CENTERS RECOGNITION

Respondents were asked whether the rare disease centers or specialized units they identified are recognized in some way. Out of 181 participants who named a center or department, the majority (67%) indicated that the centers are recognized. Meanwhile, a smaller portion (20%) stated that the centers are not recognized, and 13% of the respondents reported that they did not know whether the centers were formally recognized.

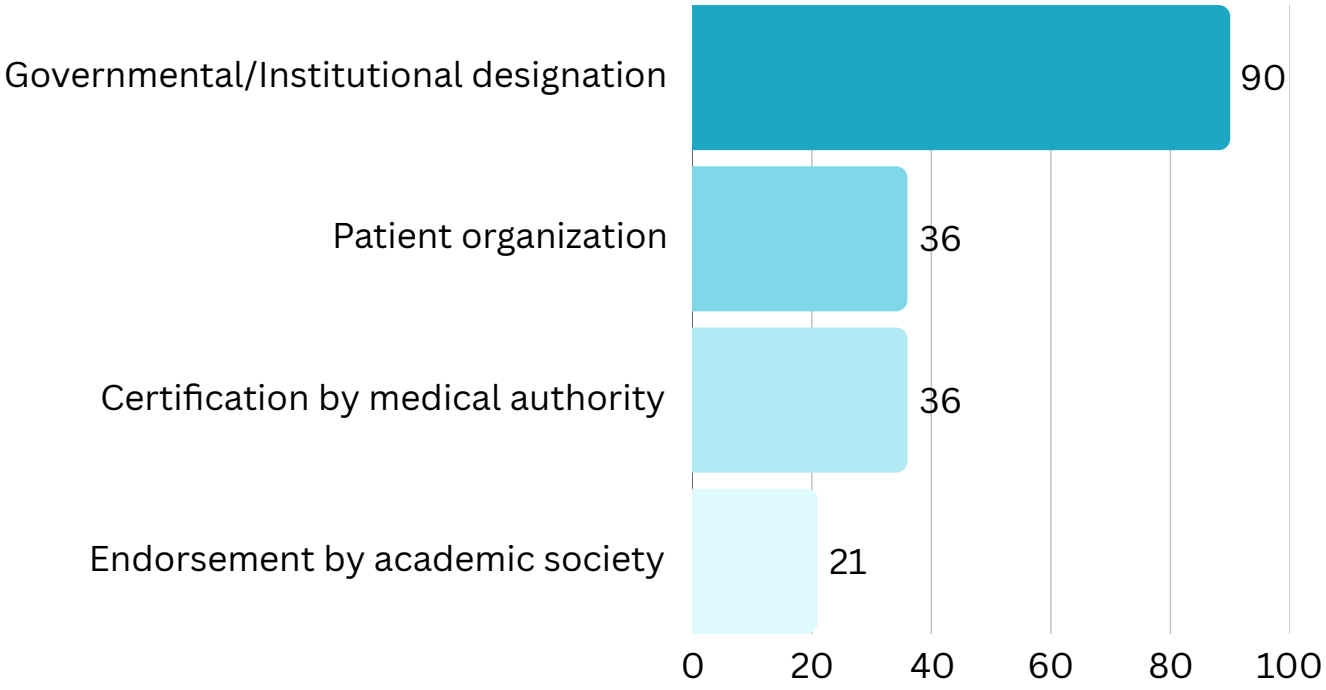
Is the center/specialized unit recognized in some way at the country level?



The wording of this question was carefully chosen to ensure inclusivity in capturing various forms of recognition. Recognition can take different forms, such as designation, accreditation, certification, or other official acknowledgments. To avoid limiting responses to a specific type, the question was deliberately kept broad rather than using a single term.

TYPE OF RECOGNITION

In what way is the center/specialized unit recognized at the country level?



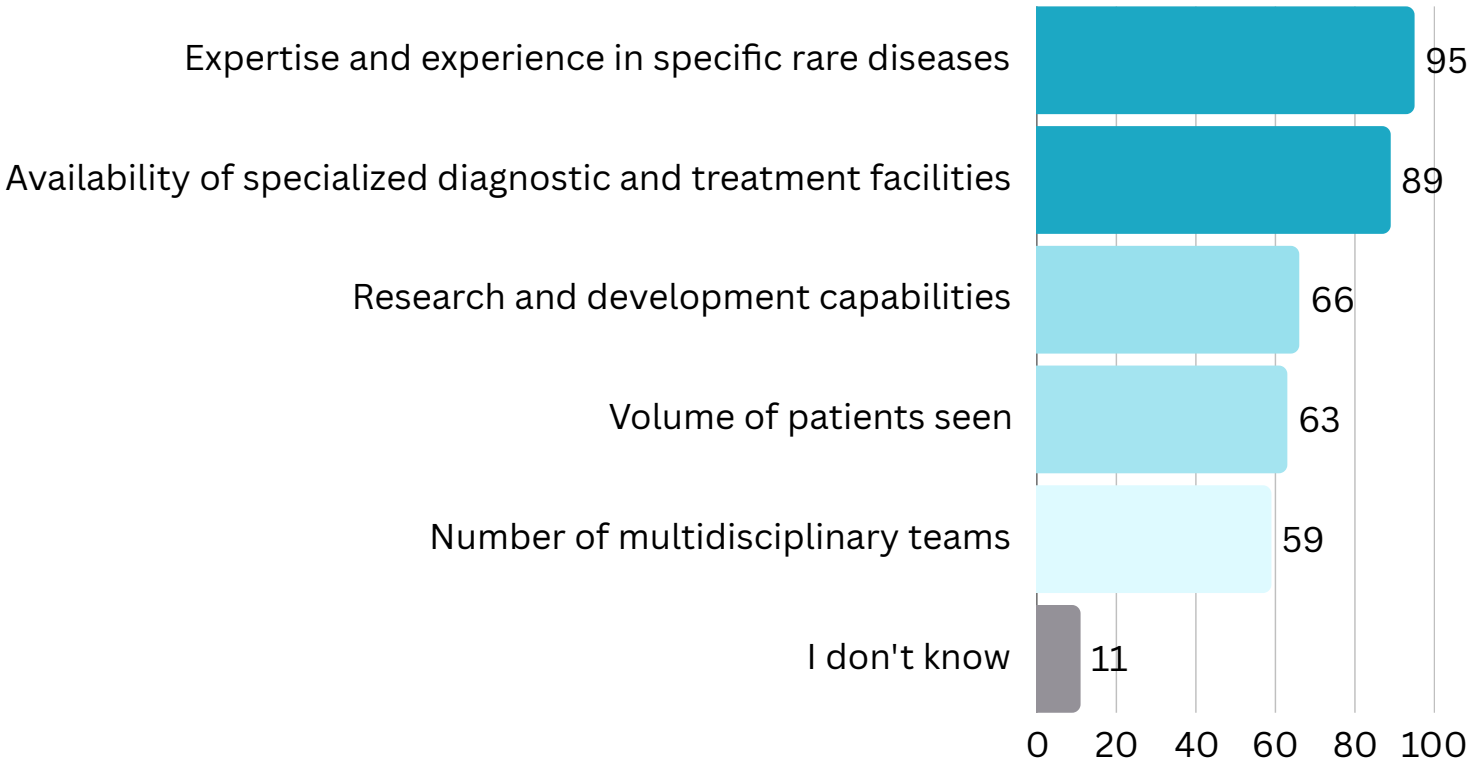
Multiple selections were allowed

When looking at the type of recognition received by the centers, most respondents (90) highlighted governmental or institutional designation as the primary form of recognition. Certification by a medical authority was the second most common type, together with recognition by patient organizations with 36 mentions each. Endorsement by professional societies was the least frequently mentioned, with 21 responses.

CRITERIA FOR RECOGNITION OF RD CENTERS

Among the respondents who confirmed that the centers or units were recognized in some way, several key criteria for recognition of rare disease centers were highlighted.
Expertise and experience in specific rare diseases emerged as the most cited criterion. Availability of specialized diagnostic and treatment facilities followed closely (89 respondents).
Research and development capabilities were identified as another significant criterion, highlighted by 66 respondents. The volume of patients seen and the number of multidisciplinary teams were each mentioned by 63 and 59 respondents. A small number of respondents (11) indicated “I don’t know”, highlighting some uncertainty regarding the recognition criteria.

What criteria are used to recognize the center/unit?

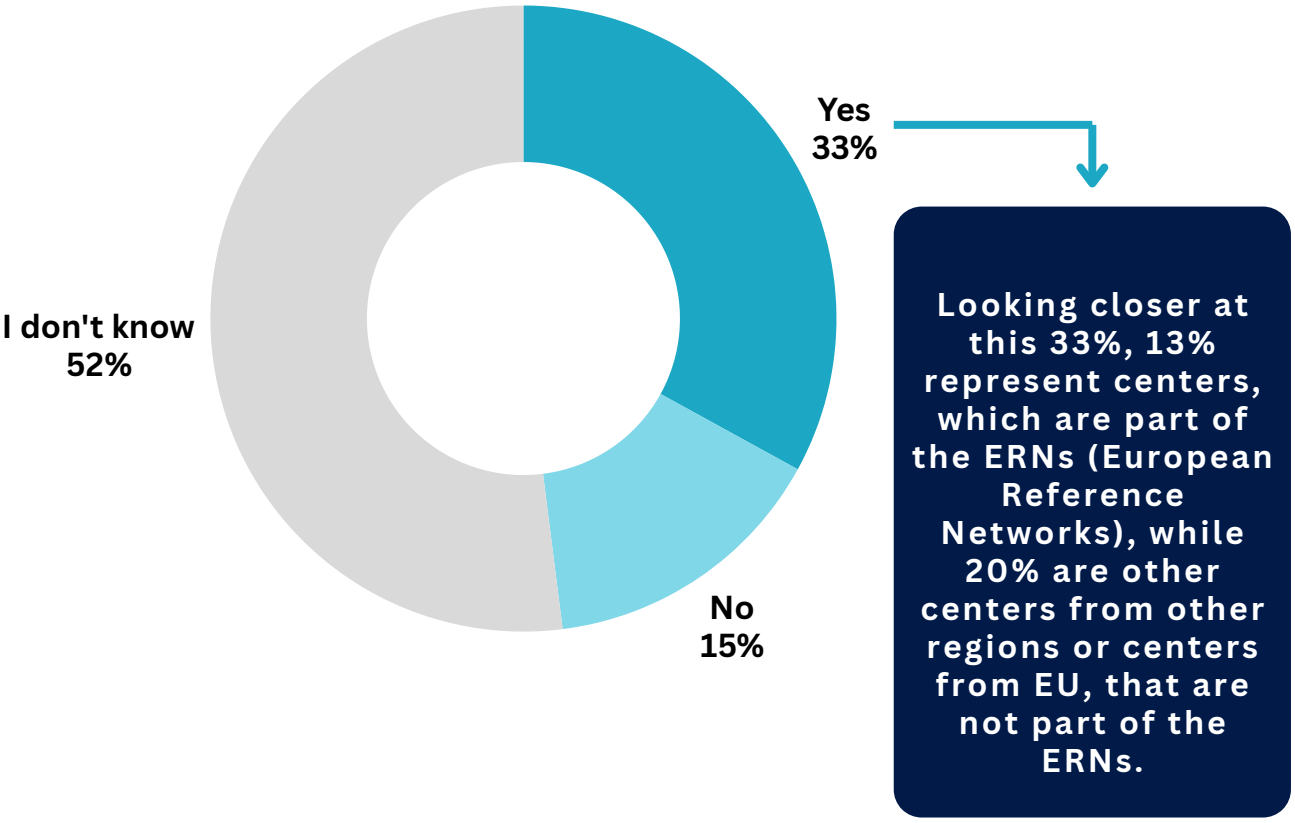


Multiple selections were allowed

CENTERS' PARTICIPATION IN CROSS-BORDER SERVICES

33% of respondents reported that the centers or specialized units they identified are involved in providing or receiving clinical services for rare diseases beyond their country's borders. In comparison, 15% of respondents stated that the centers are not involved in cross-border services, while 52% either did not provide a response or indicated they were unsure.

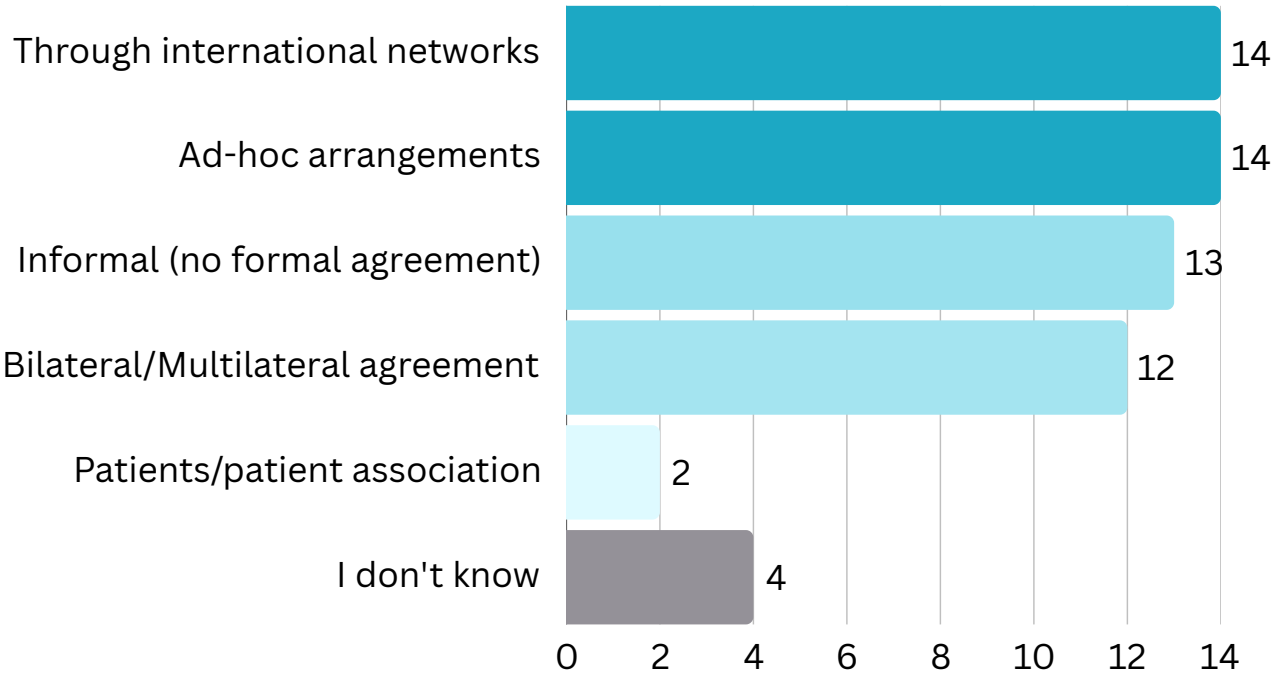
Is the center/specialized unit participating in cross-border clinical services?



MANAGEMENT OF CROSS-BORDER SERVICES

For the respondents who confirmed that the centers provide cross-border services, the management of these services shows a range of approaches. Coordination through international networks and ad-hoc arrangements were the most reported methods, with 14 respondents selecting each. These were followed closely by informal arrangements, mentioned by 13 respondents and bilateral or multilateral agreements, cited by 12 respondents. Other approaches, such as services managed by patients or patient associations, were less frequently mentioned, with only a few responses, while some respondents expressed uncertainty about how these services are managed.

How are cross-border services managed?

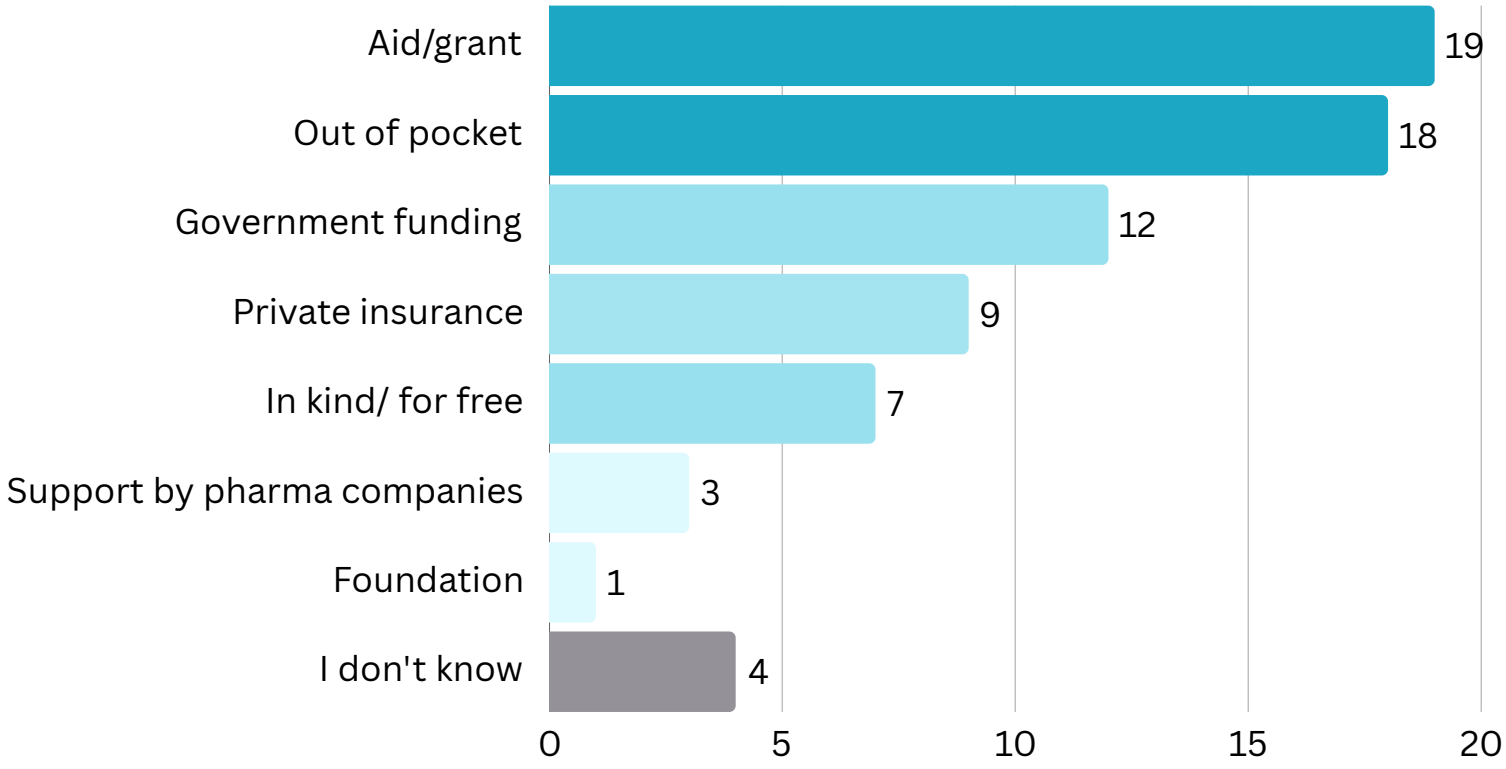


Multiple selections were allowed

FUNDING OF CROSS-BORDER SERVICES

The funding of cross-border clinical services for rare diseases varied across the respondents who confirmed their involvement in such activities. Aid or grants and out-of-pocket expenses were the most reported funding sources, with 19 and 18 mentions respectively. Government funding was the next most frequently cited source, with 12 mentions, reflecting some level of public health system involvement in supporting these services. Private insurance was mentioned less frequently, with 9 respondents. In-kind services were highlighted by 7 respondents.
Other sources of funding were less common. Support by pharmaceutical companies was mentioned by 3 respondents. Foundation was cited by only 1 respondent. A small number of respondents, 4 in total, expressed uncertainty about the sources of funding for cross-border clinical services.

How are cross-border services funded?



Multiple selections were allowed

ACKNOWLEDGMENTS

This study was conducted by Rare Diseases International. The survey was developed in collaboration with a small group of RDI members and experts.

We sincerely thank everyone who took the time to participate in this survey.

About Rare Diseases International

Rare Diseases International (RDI) is the global alliance of People Living with a Rare Disease (PLWRD) of all nationalities across all rare diseases. Representing over 120 organizations on 6 continents, RDI's mission is to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent and empower its members.

RDI is a registered non-profit organization incorporated under French law on 9 October 2018. RDI has been operating since 28 May 2015 as an informal network of rare disease patient organisations to form the global alliance representing patients and families. In 2024, RDI became a Non-State Actor in official relations with the WHO.

https://www.rarediseasesinternational.org/

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