

**Ibero-American Information and Orientation
Service on Rare, Orphan or Infrequent Diseases
(ALIBER SIO)
Ibero-America United by Rare Diseases**



<http://aliber.org>

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A project managed by Mondo Agit. Translator: Barbara Trein

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TABLE OF CONTENTS

Justification for the Need..... 3

 The Reality of Rare or Infrequent Diseases 3

European Experience 6

 Linha Rara de Raríssimas (Portugal) 6

 FEDER Information and Orientation Service on RDs (Spain)..... 7

Latin American Experiences..... 8

 Graphic 1. Consultations handled per year..... 10

General Project Data 11

 Project Objectives12

 Methodology and Implementation Process12

 Summary and Benefit of the Project15

 Beneficiaries.....17

 Territorial Scope.....17

 Resources.....17

 Personal Data Protection18

 Visibility and Sustainability Perspective.....19

 Follow-Up.....19

 Budget Proposal.....20

 Bibliographic References.....21

Justification for the Need

The **Ibero-American Alliance for Rare, Orphan or Infrequent Diseases (ALIBER)** is a nonprofit organization that coordinates actions to strengthen associative movement, gives visibility to rare diseases and represents people with RDs in Ibero-America in dealings with local, regional, national and international authorities, creating a space for joint and permanent collaboration to share knowledge, experiences and good practices in the social, health, educational and employment sectors.

Founded in Totana in 2013 within the framework of the VI National Congress of Rare Diseases and I Ibero-American Meeting for Rare, Orphan or Infrequent Diseases by 11 patient organizations, today **ALIBER** has established itself as a coalition that integrates and empowers different patient associations with RDs to defend the rights of those affected and their families, **representing a collective of 47 million affected by infrequent diseases across more than 500 organizations that bring together their 59 partners in 16 Ibero-American countries** (see list in Annex 1).



ALIBER's **strategic objectives** are aimed at:

- 1) Promoting the recognition of infrequent diseases as social, health, educational and employment priorities,
- 2) Representing people with infrequent diseases in Ibero-America, and
- 3) Increasing the empowerment of its member entities.

To achieve these objectives, **6 specific lines of action** have been designed: social visibility, social action, advocacy, training and knowledge, solidarity alliances/networks and, lastly, associative management. The present project of an **Ibero-American Information and Orientation Service on Rare, Orphan or Infrequent Diseases** corresponds to the first two themes of social visibility and social action.

The Reality of Rare or Infrequent Diseases

There are around eight thousand rare, orphan or infrequent diseases, which collectively affect between 6 and 8% of the population, equivalent to 30 million people in Europe, 25 million in the United States of America and 47 million in Ibero-America. The figures mentioned constitute what EURORDIS (2005) has called the rarity paradox, pointing out that although the diseases are rare, there are many patients.

Regardless of their heterogeneity, the RDs share common aspects such as complex medical and social problems due to their severe consequences and the considerable burden on individuals and their families (Houřez, Sánchez de Vega, Brignol, Mazzucato and Polizzi, 2014). Studies that describe the reality of people with RDs in Spain, Germany, Slovakia and the United Kingdom render account to very similar experiences (Spanish Federation of Rare Diseases, 2009; Budyč, Helms and Schultz, 2012; Ramljaková, 2013; Muir, 2016), which are not far from those described for Latin America by the Ibero-American Alliance for Rare Diseases (2016) and that can be summarized in the following points:



Diagnosis and Treatment

- ✓ Difficult access to information about the disease: diagnosis and treatment.
- ✓ Delayed diagnostic process and frequent wrong diagnoses.
- ✓ Absence of diagnosis, causing a barrier to the access to coordinated care and appropriate treatment.
- ✓ Late diagnosis, depriving people of timely therapeutic interventions preventing the approach to the disease.
- ✓ Diagnostic delay, which reduces the possibilities of access to social assistance that people are entitled to.

Health Personnel

- ✓ Limited number of trained specialists to attend to different cases and mostly in the metropolitan areas.
- ✓ Great lack of coordination between professionals in the field.
- ✓ Lack of effective treatment, which worsens the patient's health.

Policies and Health Infrastructure

- ✓ Difficult access to experimental treatments, implying a strong struggle against bureaucracy and health system regulations.
- ✓ Shortage of reference centers and little known, if around.
- ✓ Research is scarce, fragmented, insufficient and undisclosed.
- ✓ Limited policies to motivate scientific research.
- ✓ Lack of political and social awareness.
- ✓ Few epidemiological records, which impedes the development of policies and support services for those affected.

Impact on Families

- ✓ Shortage of reference centers, which forces many affected people to move and sometimes even settle in other cities near the point of care.
- ✓ Shortage of social and health aid mechanisms, which leads to impoverishment.
- ✓ Difficulties for family finances due to the patient's employability or, in the case of minors or dependents, due to the resignation of at least one of the parents from the workplace to perform the necessary care.

Against this backdrop, the patient faces **social and psychological challenges**, such as access to necessary resources to develop an independent life in every possible way; lack of socialization and acceptance in their society; assessment processes of disability and dependency that do not consider their pathological conditions and/or symptoms; management of a diagnosis when there is an incurable disease and/or the acceptance of waiting times until a diagnosis is made (which sometimes takes more than 10 years); feeling isolated from doctors and researchers as well as from other patients with the same pathology; difficult access to education, employment and social inclusion; absence of practical assistance in everyday life – all produced by a common problem:

- ✓ Unawareness of the origin of the disease, confusion and disorientation at the time of diagnosis.
- ✓ Lack of disease protocols.
- ✓ Lack of legal coverage to promote research.
- ✓ Lack of specific medications to treat the disease.
- ✓ Social rejection and loss of self-esteem.
- ✓ Sense of loneliness, of being isolated.
- ✓ Misinformation about care and technical assistance to improve their life.
- ✓ Lack of financial aid.
- ✓ Lack of sufficient social awareness of this type of disease, which complicates social acceptance.

The reality described shows that **the RDs pose an important challenge in terms of public health** due to the extensive lack of information, development and tendencies.

The situation described so far reflects the need for information and guidance for people with RDs and their families as well as for professionals who work with them. Such need for information covers different aspects (general information about the disease, treatments, reference centers and/or experience, specific professionals, conducting clinical and/or diagnostic tests, contact with other people with the same pathology, aspects of the educational and employment sectors, emotional support, etc.) and requires a multiple perspectives approach: information, support, guidance, and training.

This information, which patients, family members and professionals need, generally comes from patient organizations, entities that allow the collective action of those affected to have a greater influence and control over their determinants of health (Aymé, Kole and Groft, 2008). For the World Health Organization (1998), patient associations enable people to apply their skills and resources in collective efforts with the aim to address health priorities and satisfy respective needs.

In view of the difficulties already mentioned, such as those deriving from the pandemic we are currently experiencing where the appearance of COVID-19 is taking its toll on health systems and highlights the therapeutic development challenges of patients with rare diseases turning them into people at higher risk due to their already vulnerable situation, our project regains meaning. It concentrates on specific resources that can help people with rare diseases get to know processes and guidelines appropriate to the situation in each of the Latin American countries.

For all this, the specialized information and orientation services are fundamental and must be provided in a professional way, despite the help of volunteers who, even when encouraged by the improvement in the quality of life of patients, do not manage to offer reliable, extensive and informative support.

We already know that **one of the services that contributes to the wellbeing of those affected by RDs and their families in an important way are the helplines.**

According to Sellen (2009), these services have been gradually set up under the impetus of associative movement, facilitating the relationship between health systems and users.

These information systems on RDs at the national or regional level are very important for patients and family members according to EURORDIS (2011), since knowledge and information can help them manage the disease as well as empower patients. In this regard, research on such resources indicates that information is identified as a key action area (Houyez et al., 2014) and that parents appreciate obtaining detailed information about the disease and are grateful to receive support for the multiple factors they encounter (Smith and Daughtrey, 2000; Rahi et al. 2004; Carnevale et al. 2006; Dellve et al. 2006 and Winkler et al. 2006, quoted by Gischler, Mazer, Poley, Tibboel and Van Dijk, 2008).

European Experience

In Europe, there is a European network of helplines that includes 17 lines with support and information services on rare diseases. The countries that provide this service are: Belgium, Bulgaria, Croatia, Denmark, Spain, France, Italy, Portugal, Romania and Switzerland. Among them is the Information and Orientation Service of the Spanish Federation of Rare Diseases (FEDER SIO) and Linha Rara de Raríssimas, both member organizations of ALIBER.

Linha Rara de Raríssimas (Portugal)



Linha Rara is an information and support service provided by Raríssimas – Associação Nacional de Deficiências Mentais e Raras (National Association of Mental and Rare Disabilities), a helpline that has three main lines of action: information, promotion of patient communities and health education. Its mission can be summarized in four pillars: listening, informing, advising and guiding.

The objective of Linha Rara is to identify and respond to the needs of people with RDs and their families, via telephone, email or through on-site assistance at their headquarters in Moita (Portugal) during working days, from 10.00 to 18.00.

The support team is made up of a psychologist and a social worker who have previous experience in carrying out activities with patient associations and other nonprofit institutions. The service relies on the backing of the Raríssimas Legal Counsel who provides support when legal guidance is required as well as backing of the Clinical Director of Raríssimas who provides support in medical/clinical matters.

Through Linha Rara it has become possible to identify the real needs of the population affected by a rare disease. Since its creation in December 2009, and until 2019, Linha Rara has received **15,000 information and support requests**, 23% of which were scientifically validated information requests on a particular rare disease. 10% of requests were related to medical matters about receiving or confirming the diagnosis and 11% required information on therapeutic options, medications and clinical trials. A significant number of requests, 19%, were for legal support, social support and responses, and rights of patients and their caregivers. 11% of requests were regarding patient groups and support associations specific to each disease and, lastly, 5% were seeking psychological support.

Most of the information and support requests received by Linha Rara were made by patients with an RD (28%) followed by the mothers of those affected, being the main caregivers (26%). 20% of the requests were made by other family members and friends of people with RDs. 6%

of the total requests were made by students and 6% by health professionals or other professionals (social workers, psychologists, etc.), and, lastly, 2% by teachers and another 2% by journalists.

The consultations originated from more than 40 countries, Brazil being second with the highest number of requests (8%), after Portugal making up 87%. 0.3% came from Spain and 0.04% from Colombia.



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FEDER Information and Orientation Service on RDs (Spain)

The Information and Orientation Service on Rare Diseases of the Spanish Federation of Rare Diseases is a care and support process which helps define the needs of the people affected by rare diseases, people who have no diagnosis, their family members and professionals.

In the support process, and once emotions and thoughts have been externalized, the communication process continues to try to organize the more immediate needs of the person who consults.

FEDER puts its SIO at anyone's disposal. It is a free service for the people who use it and its objective is to improve the quality of life of the people with a RD and their families by facilitating access to quality information, as well as accommodating, supporting and guiding the people who consult us.

In addition, the SIO encourages the creation and promotion of networks for those affected, family members and professionals and provides information to our administrations in order to improve the level of care of the people affected.

We rely on the collaboration of a Scientific Advisory Committee consisting of 26 specialists and researchers from main health care branches for people with rare diseases (geneticists, neurologists, internists, physiotherapists, pharmacists, among others). Additionally, we have a team of technicians consisting of 11 different types of professionals such as social workers, biologist, lawyer and educational psychologists, who have extensive experience to assist all the people who need guidance in relation to infrequent diseases.

This is currently the **only comprehensive support line for people with infrequent diseases in Spain**, and thanks to its work experience since 2002 it is at the forefront of the European Network of Helplines for Rare Diseases.

The SIO responds to socio-health requests, undiagnosed cases, legal requests and educational requests.

The work is carried out with an action protocol and a SQL SERVER database, registered at the Spanish Data Protection Agency, for the collection of data and information structure.



On the whole, since 2003, they have accounted for the support of 38,332 people across more than 78 thousand consultations. During 2019, they provided support to 4,518 people who made 8,865 information requests, receiving a total of 1,113 calls at their *Call Center* and achieving 92.63% of user satisfaction. Of these cases, 290 of them were undiagnosed cases.

Requests from Ibero-America 2003-2019

- Argentina: 1,587
- Mexico: 1,034
- Colombia: 1,033
- Peru: 634
- Chile: 606
- Venezuela: 507
- Ecuador: 282
- Uruguay: 200
- Cuba: 262
- Guatemala: 171
- Brazil: 104
- Portugal: 89
- Bolivia: 82
- Costa Rica: 77
- Panama: 69
- El Salvador: 89
- Honduras: 34
- Paraguay: 53
- Nicaragua: 57
- Dominican Republic: 57
- Puerto Rico: 11

So far, the FEDER SIO handles the majority of the **consultations in Latin America** due to the language and, as it happens, **7,038 requests** (SIO database) of 4,390 users have been addressed in the period from **2003-2019**. The main duties of the Information and Orientation Service in Latin American countries, including specific data, have been:

- ✓ Facilitation of reference association data (FEDER): 351
- ✓ Sending more specific information on the disease and on patient management: 3,539
- ✓ Facilitation of reference association data not included in FEDER: 1,423
- ✓ Facilitation of contact with others who are affected by the same pathology: 187
- ✓ Sending information about professionals: 546

Latin American Experiences

At present, **there is not a similar experience known in Latin American countries** since ALIBER, although there are different highly centralized services on specific pathologic conditions or in specific sectors that respond to needs for information and guidance in terms of rare diseases. Let us take a look at what they are:

The **Mexican Federation of Rare Diseases (FEMEXER)** is the only organization in Latin America that seems to formally offer an information, guidance and psychological support program for people with rare diseases, called **Accesalud**.

According to the information on its website, the main objective of Accesalud is to operate a comprehensive psychological care model, which, due to the empowerment through information, guidance and psychological support, improves the quality of life of patients with RDs and their families.

The program incorporates five lines of action: information, guidance and psychological support for those affected by RDs, awareness and promotion of the RDs, and support and cooperation with patient associations. Unfortunately, to our knowledge, there is no published data on its operation.



Since 2014, the **Argentine Federation of Rare Diseases (FADEPOF)** provide support to the consultations that patients and family members have via a form on their website. In addition, they maintain a section on their website with current information on reference centers for pathologic conditions and assist in the search for entities that can advise on particular rare diseases. Volunteers handle the doubts and queries that are received. The requests received are forwarded to patient associations of the specific pathology or health professionals for referral. The data is recorded, but has not yet been systematized.

Furthermore, in Argentina, the Liaison, Research and Support Group for Rare Diseases (GEISER) published a **Resource Guide for Rare Diseases in Argentina** together with IntraMed in 2009, which was updated in 2011. The guide consists of the care description and resources in said country for 64 rare pathologic conditions.

In Ecuador, the **Ecuadorian Foundation for Turner Syndrome (FEPAST)** has provided information and guidance for people with this pathology since 2011. This task is performed by volunteers and above all consists of the guidance for parents of newly diagnosed girls, holding informative talks and different workshops on the disease. There is no record of the service provided.

In Colombia, the **Ibero-American Foundation for Multiple Arthrogryposis Congenita and Other Uncommon Diseases (ASOIBEROAMC)** relies on an online orientation and information service for Arthrogryposis. The service has been in place since 2010 and is run by a psychologist who is also affected by this condition and founder of the organization. The association does not keep statistics on the service.

Furthermore, in Colombia, the **CroniCare Foundation**, despite not having a properly established information and orientation service, receives information requests, to which a response is given in the shortest possible time, according to its network of inter-institutional resources: Cronired, which has been operating since 2015. Through this service, requests are received, which are responded to with the support of the Foundation's Medical Advisor when they are related to the diseases that CroniCare treats, or are forwarded to other umbrella entities when they have no information on the diseases in question. Two people who are hired for this purpose handle this service. In addition, they have a communication channel with scientific management with an expert in different orphan diseases not based at Cronicare, offering orientation in the case of emergency. The organization does keep a record of the requests.

Finally, in Brazil, the **Rare Lives Institute** has recently launched a helpline (**Linha Rara Brasil**) to guide and advise people with a rare disease and their families, health professionals, students, teachers and the public in general, via a specific telephone line and an email (linharara@rarissimas.org.br). This helpline has been made possible thanks to the collaboration between the Rare Lives Institute, Raríssimas Portugal and Raríssimas Brazil and relies on 6 professionals to provide advice, in addition to a health coordinator.

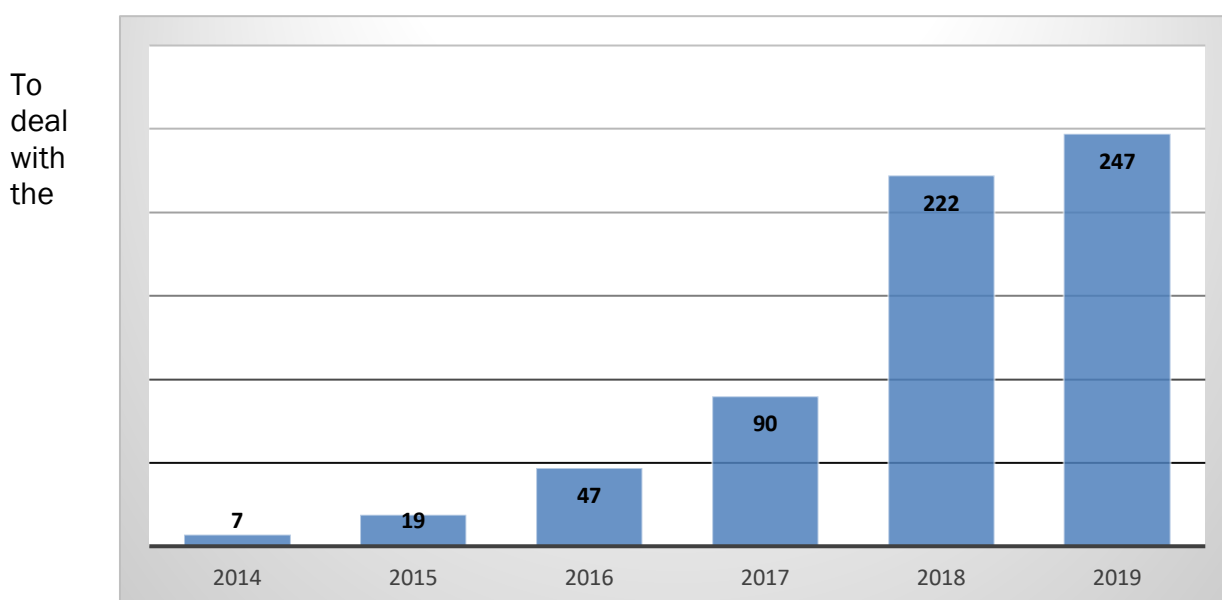
Against this backdrop, we see how **there is no complete care service in Latin America** like there are in Europe, where those affected by any rare disease and in any of the Latin American countries find answers to their questions and can be guided worldwide providing local information, but also from outside their borders so that the patients and their family members can freely make decisions that are most appropriate to their circumstances and needs.



ALIBER established a pilot experience in 2018 to offer its own Information and Orientation Service. The project was launched that year, and research work has been conducted for the collection of data for the database, which we will use to start the provision of the service. In addition, specific training for two technical professionals was carried out in 2019, thus moving towards the development of the Ibero-American Information and Orientation Service on Rare, Orphan or Infrequent Diseases.

From 2014 until 2019, we have recorded and received more than **500 requests** across different channels such as Facebook, mail, WhatsApp, forms, referrals, etc. We can see how **the number of consultations has been increasing exponentially each year**, as can be observed in Graphic 1. **This supports our need and benefit of a free and stable information and care service with reliable information, handled by trained professionals.**

Graphic 1. Consultations handled per year



consultations during these years, we have relied on the collaboration of the FEDER SIO, as well as the member organizations of ALIBER in the different countries, thus offering the information and guidance that they requested from us.

In the six years since the creation of ALIBER, **we have developed a database of Latin American patient organizations, which currently holds information on more than 910 associations** that we can collaborate with whenever required by a pathological condition or a geographic location. At the same time, we have **identified renowned professionals per pathology and categorized the diseases represented by each entity related to the care of people with RDs in Ibero-America.** The project has relied on the collaboration of two technicians from Uruguay and Colombia, from the Association All United by Rare Diseases Uruguay (ATUERU) and the Ibero-American Association for Arthrogyrosis and Other Uncommon Diseases (ASOIBEROANC), respectively.

The experience of our partners in Europe, together with the activities, formal or informal, which are already being carried out from Latin America to provide support to the consultations of those affected by RDs and the collection of information on associations, resources and services that we continue to implement at ALIBER, require us to propose the consolidation of the **Ibero-**

American Information and Orientation Service on Rare, Orphan or Infrequent Diseases for 2020.

General Project Data

Being aware of the enormous complexity of the health reality in the different Latin American countries, due to their diversity as well as for the challenges they face, we believe that rare diseases should also have a specific approach.

In a report carried out as a preface to the creation of ALIBER Guerra and Pino-Ramírez (2013), it is reviewed that the health model in Latin America is mixed, involving public and private care, with public health systems being generally precarious, which practically forces users to get private insurance or depend on social work.

Health resources are also scarce, to which must be added the lack of knowledge that health professionals have about RDs. There are few training programs in the sector for health professionals, which means that the involvement of health officers is low or nonexistent and aggravates the already complex problem concerning those affected.

One of the challenges we are currently facing is the response of health systems to the pandemic produced by COVID-19, and this large issue has resulted in a multitude of consultations around the imbalances that those affected by rare diseases have to face to continue with their therapies, consultations with specialists, access to their medications, etc.

The health challenges regarding the pandemic are not managed from the patients' perspective, which already presents a situation of vulnerability, and which makes them resort to health systems more regularly. Due to the consequences and the measures adopted by the different countries concerning COVID-19, the patients with Rare Diseases are being significantly affected (see EURORDIS reports regarding the Rare Barometer survey COVID-19).

For ALIBER, this backdrop represents a compromise and an opportunity for the consolidation of the Ibero-American Information and Orientation Service on Rare, Orphan or Infrequent Diseases, with three major lines of action:

- a) The visibility of rare diseases,
- b) The organization and centralization of resources for rare diseases in a single database, which is to become a reference like other large databases in Europe or North America (see the databases of Orphanet, Nord or of FEDER, for example)
- c) The provision of orientation and informational support services for people with infrequent diseases as well as for socio-health professionals and those from the research sector, improving patient care with the service and thereby the quality of life of those affected, at the same time as strengthening patient associations.

Project Objectives

General:

To improve the quality of life of the people affected by infrequent diseases in Latin America through the implementation of a unique Information and Orientation Service on Rare, Orphan or Infrequent Diseases in Latin America.

Specifics:

- ✓ To develop a strategic support line between patient associations and members of ALIBER.
- ✓ To raise awareness about the problems around RDs among the Ibero-American society and among political actors.
- ✓ To exchange knowledge and good practices on care from the Information and Orientation Services of RDs.
- ✓ To develop a training and empowerment program on Information and Orientation Services of RDs for the member entities of ALIBER.
- ✓ To prepare a resource guide (associations and services) for the care of RDs in Ibero-America.

Methodology and Implementation Process

The implementation of the Ibero-American Information and Orientation Service on Rare, Orphan or Infrequent Diseases has been planned in various phases.

In the **first phase** (during 2018), the project was presented and project visibility was studied by establishing contact with member entities of ALIBER and receiving data from the information services (support lines) that were being developed in Latin America, their execution, operation, the statistics of the different services, etc.

In the **second phase** (during 2019), we have advanced in the **search for resources, entities, reference and/or experience centers, professionals, entities for rare, orphan or infrequent diseases, etc., of Latin American countries**, and necessary data to feed the service database.

During this phase, **the SIO authorization forms for consultations have also been prepared.**

1. Authorization for the ALIBER SIO consultation
2. Authorization for data collection and processing
3. Authorization for image rights transfer
4. Personal data

Finally, in this phase, two technicians were trained by the coordinator of the Information and Orientation Service of FEDER, Estrella Mayoral, who has extensive experience in this area.

In the **third phase** (during 2020), we have started with the preparation of all the documentation that we will upload to the Server, the tool that will give way to direct care. This documentation

serves the resources and services of the communities where the people live that make requests.

In addition, we are offering responses to requests related to COVID-19, due to having already carried out work to collect specific resources and specific documents regarding the situation for people with rare diseases; collaborations to collect information have also been established, for example with EURORDIS, through the Rare Barometer Voice survey.

Our next steps to move forward in this third phase of the project will be:

- ✓ Knowledge of the services provided by ALIBER partners and other associates related to infrequent diseases (in process).
- ✓ Development of a map of patient associations and services related to RDs in each country (in the process of validation and expansion).
- ✓ Identification of specialized professionals by pathology and country (in the process of validation and expansion).
- ✓ Selection of professionals and invitation to form part of the expert and support committee for answering requests (in process).
- ✓ Preparation of protocols for the referral of consultations to the representatives of the expert committee and/or the corresponding services (pending).
- ✓ Creation of database where all the information will be uploaded as the main tool for the launch of the support service (pending).
- ✓ Information upload to the database (pending).
- ✓ Familiarization with the tool by the technician responsible for receiving and answering requests (pending).
- ✓ Establishment of collaboration agreements with institutions and administrations that support RDs in the different Latin American countries (pending).
- ✓ Promotion of services across the web, social networks, mailing, awareness campaigns, etc. (pending).
- ✓ Start with the reception of requests and direct care through the data of our server.

It should be noted that the last action outlined on the reception of requests and direct care will offer users of the service information related to their pathology in relation to: treatments, professionals, reference centers in their country, organizations that can guide them, specific programs, the possibility to get in contact with other families in their country with the same disease, access to articles and documents with information on their pathology worldwide, etc. **This action will be continued over time without a deadline for completion** since it is about offering continuity to the service and continuous improvement of the information that is offered through the advances that will be made in each country in terms of rare diseases.

On the other hand, a **collaboration and interrelation with the project on the study of socio-health needs of people with rare diseases and their families has been established, ENSERio Latam**, also promoted and executed by ALIBER, through which specific information about professionals and reference centers for rare diseases and per country is being obtained, among other data. From this, it will be possible to relate the needs detected via this study with the requests that reach us at the ALIBER SIO, establishing recommendations to patient entity members of ALIBER like those that do not belong to the Alliance, based on programs and services to launch for people with infrequent diseases. At the same time, linking this data will enable us to develop specific reports to encourage and promote possible public policies in each of the Latin American Countries.

Finally, also in this third phase, **training activities for technicians** will take place, which will be carried out by the coordinator of the Information and Orientation Service of FEDER.

In the **fourth phase** (from 2021), the service will be expanded through the gradual implementation of **local SIOs in different Latin American countries**, offering specific training to the technicians who settle locally so that the project starts to diversify geographically, something which will allow us to have a greater knowledge of local resources, receive the increase of requests that we predict will take place and establish closer collaboration networks with the member entities of ALIBER.

From this year, we will already be able to access specific data on the efficiency of the service, the number of requests and the mechanism of the computer tool. This makes it possible to carry out **improvements of the tool in line with this analysis**, fill the gaps and improve information by backing and empowering the non-governmental organizations that work in each country in favor of people with rare diseases, as well as to add new countries and extend the service for more people.

Thus, in this phase, specific actions related to the following will be developed:

- ✓ Empowerment of entities, from which the provision of the Information and Orientation Service will be expanded with local SIOs.
- ✓ Enabling of the computer equipment of the professionals who provide the service.
- ✓ Evaluation of the service following the first semester from its launch.
- ✓ Presentation of results in congresses, workshops, magazines or any other medium.
- ✓ Use of data that has accumulated in the database to detect and get to know the real needs of patients and family members and be able to propose the development of specific programs that could be implemented by the entities of ALIBER.
- ✓ Strengthening of the skills and abilities of ALIBER SIO technicians through specific training programs based on the difficulties they encounter in order to respond to the requests they receive.
- ✓ Producing a map of the situation of RDs in Ibero-America, which we will achieve by sharing a single database.
- ✓ Establishment of information exchange processes that allow:
 - Sharing information about pathological conditions that have no representation, of ultra-rare or undiagnosed cases.
 - Promote the exchange of good practices between the different local SIOs.
 - Encourage networking and improve the socio-health institutional coordination through the facilitation of mechanisms, action protocols and exchange of information between the different administrations and entities involved.
- ✓ Establishment of an online coordination network between associations – entities that work with RDs to exchange information about them.

Summary and Benefit of the Project

The project is based on two actions that essentially complement each other.

First, to contribute to the improvement of the quality of life of those affected through the informational support of their disease, offering responses to the collective of the RDs by adapting to the resources and services of the communities where they live or, if necessary, guiding them towards the resources and services closest to their place of origin, establishing contact with other people with the same pathology, informing about news regarding their disease, etc. Similarly, the consolidation of this project will promote the empowerment of associations, favoring the development of local information and orientation services on RDs.

Second, to develop a communication strategy of Latin American patient organizations via ALIBER, through its web pages and social networks for it to have a greater capacity for impact on the main actors of RDs: professionals, students, parents, and governments informing on the serious health problem posed by RDs in Ibero-America and their scarce or lack of attention.

Thus, this project has three main themes: (research, diversification of care and expansion of social visibility):

- ✓ In relation to the research theme, we will continue with **research activities and recognition of resources specific to RDs: patient associations, reference centers, experience centers, skilled professionals in RDs, research centers, diagnostic centers, clinical trials, treatments available for pathologic conditions, rehabilitation services, etc.** All these resources will be uploaded to a database that will host the necessary information for the launch of the service with guarantees of success, and it will be fed constantly by updating its data and introducing new data that we will be accessing regularly or in situations like the one we are dealing with now with the pandemic.

The correct and efficient content structure in a single database will allow the technicians responsible for receiving requests to give precise information swiftly, as well as to keep a record of the incoming requests and the responses provided. By classifying these into different categories, they will offer us very relevant data based on specific needs, concerns, interests and future action lines from the alliance, of supportive as well as political value.

- ✓ In relation to the diversification of care, the project will incite the geographic expansion where responses to incoming requests will be offered from, by promoting **training activities and the exchange of knowledge with technicians from entities for rare diseases in Latin America for the creation of local SIOs.** The intention is to create a network of professionals who follow the same protocols and use the same database, and who receive requests from different parts of Latin America offering responses to the users of the service, as well as to continue with the ongoing research and feeding of the database with new resources.



To perform this training, we will rely on the experience of the Information and Orientation Service of FEDER. The coordinator of this service, Estrella Mayoral, who has been in charge of it for more than 15 years, will be able to instruct the technicians to settle in various parts of Latin America so that they form part of the technical team of the service and receiving training on information and orientation activities focused on:

- a) Providing the necessary information to people who need to know a diagnosis of their disease and who suffer from symptoms associated to RDs, as well as supporting the process for socio-health resources and technical assistance,
 - b) Accompanying and supporting people with RDs, by referring users to more specific services,
 - c) Informing and advising entities,
 - d) Participating in the management of projects to detect the needs of patients and family members with RDs, in addition to promoting the creation and consolidation of intervention programs to alleviate these needs,
 - e) Contributing to the empowerment of member associations of ALIBER through the development of support and information services for RDs for ALIBER and its partners, etc.
- ✓ Finally, in relation to the **social visibility** theme, the project will maintain a clear purpose of making the reality of people with RDs known, highlighting their general situation at the level of each country and of the region, for which **the design of a joint strategy of all the countries of Ibero-America is proposed**, as well as the promotion of the adherence to the Ibero-American Pact by the RDs of governmental entities at a local, regional and national level. Through the campaign, we have informed the institutions and organizations of Ibero-America related to health about the importance to promote health policies to improve the quality of life of the community. **Specific data based on consultations on the needs of people with rare diseases in each country and the existing resources to alleviate them must be obtained to prepare exact reports. This will be essential to point out to governments what their countries or their localities lack and what may be necessary to maintain the wellbeing of people with RDs.**

Therefore, the development and strengthening of the ALIBER SIO will facilitate:

- **The promotion of public health policies and actions.**
- **Similarly, the service, especially once local SIOs have been implemented, will act as an observatory for the difficulties of citizens affected by these diseases, constituting a valuable tool that will provide feedback on the performance of programs for the care of affected populations and the impact of the national and regional public policies related to RDs.**
- **Last but not least, these services will encourage and help the creation of patient registers both at a local and regional level.**

Beneficiaries

- ✓ Number of direct beneficiaries: 8,181
- ✓ Number of indirect beneficiaries: 24,543
- ✓ Number of potential beneficiaries: 29 million
- ✓ Direct beneficiaries will be all those people with a rare disease who live in a Latin American country and require information related to the needs that arise according to their disease at a clinical and social level. Currently and in line with the registered data, we can consider the population of users directly from the number of users from Latin America that the FEDER SIO has served from 2003 until the end of 2019 (7,011) and those served by Linha Rara de Raríssimas Portugal (1,170).

However, it is expected that, with the appropriate infrastructure and promotion, the consultations will increase exponentially.

- ✓ For their part, indirect beneficiaries will be family members of the people living with an infrequent disease, the number reflected being the estimated number of family members who live with the affected person (normally three), according to data from the World Family Map (2015).
- ✓ Finally, the potential beneficiaries have been established based on the estimated number of those affected by the RDs in Latin America (except for Brazil, which, as we have seen above, will continue to rely on the support line that Raríssimas has in Portugal due to its good performance, language and consolidating its own helpline).

Territorial Scope

Our territorial scope of action covers all the Latin American countries.

Resources

IT and Telecommunications Infrastructure

The support to the users of the SIO will be online in the first instance, preferably via email, however, different situations will be addressed based on their complexity and need requested and/or encountered, and visual face-to-face processes have also been established through secure platforms like Cisco Webex Meeting.

Contact with the Ibero-American Service for Rare, Orphan or Infrequent Diseases may be made by the affected themselves, family members and/or professionals, via email, WhatsApp, Facebook or the form on the ALIBER website.

For access to support via email, there are already specific forms available to receive the request, and this is registered on a platform, from which statistical data can be obtained. These

are the two ALIBER domain emails: sio@aliber.org and coordinación.sio@aliber.org. The users will receive the answers to the questions they send us from these emails, relying on a high-speed internet service for this.

The tool that will centralize all the work of collecting information and consultations will be a tailored database, developed by ILUNIUN, a company that will work with the Spanish Federation of Rare Diseases (FEDER) in the creation of its new database for the FEDER SIO. At ALIBER we will have our own database due to the mutual collaboration that these two entities maintain.

The database will allow:

- ✓ To monitor the consultations and actions that take place.
- ✓ To maintain a list of resources for RDs available in Latin America, general and permanent as well as specific and transitory like the ones generated by COVID-19 and other future situations we may face in socio-health matters.
- ✓ To carry out reports and research.
- ✓ To facilitate contact between all the actors involved (people affected, family members, professionals, associations, health and governmental institutions).
- ✓ To facilitate the detection of coordination needs between services, such as the lack in care, which the countries present.

Personnel

To give continuity to this project and to be able to consolidate the ALIBER SIO, a full-time director/coordinator will be hired as well as a technician for the service who will carry out all the tasks described for 2020.

Regarding the development of local SIOs, new technicians who will be able to look after the service will be hired, who will be trained by the personnel of the FEDER SIO as well as by the director/coordinator who will have been performing direct support actions and database management of the ALIBER SIO. The training will particularly refer to the management of the database and the specific actions for the performance of their tasks. It is intended that there be capacity to hire at least two technicians located in two different countries in Latin America during 2021, which are likely to be Colombia and Uruguay since a first training of technicians of two member entities of ALIBER already took place in these countries.

Little by little and depending on the demand of the service, technicians from different countries will be incorporated.

Personal Data Protection

Given that the ALIBER SIO will operate all over Latin America, the current legislations on the protection of personal data in the area have been reviewed, finding that this regulation identifies the databases as an organized set of personal data and indicating that appropriate treatment of such data will have to conform to the principles of legality, formality, quality, proportionality, information and conviction (Guzmán, 2016). In any case, applying the legislation to best protect it will safeguard personal data.



Visibility and Sustainability Perspective

The ALIBER Directorate and those responsible for the FEDER SIO and Linha Rara de Raríssimas have held work meetings. The aim has been to establish a joint working path to allow taking the necessary steps for the creation of the Information and Orientation Services at ALIBER and its partner organizations in Latin America in the mid-term. This advisory relationship is managed by collaboration agreements, already signed with FEDER and in place with Raríssimas. Additionally, we have the support and guidance of the departments of ICT and Communications at FEDER.

Likewise, ALIBER has developed the study of associative movement in Latin America, which allows knowing the already existing patient organizations in this region and the services they provide, a matter of crucial importance for the implementation of the ALIBER SIO. At the same time, a database is being developed with the contact details (name, pathological conditions, telephone numbers, postal address, email, social networks, among others) of the RD patient organizations in Latin America.

On the other hand, a first training session has also been developed for the personnel required to handle requests both in Uruguay and in Colombia through an online training intended to train the personnel in the provision of the service (handling requests, relationship management and operational management of the database). This training will continue with new online sessions and some of them on-site, and will be extended to new professions from different countries as the ALIBER SIO gets implemented in Latin America.

In the same way, we will rely on professionals from the communications department at FEDER for advice on the promotion strategy of the service and the visibility of RDs.

Finally, it is important to mention internet access and fixed and mobile telephony in Latin America, since the success of the service will depend on it to a large extent. According to data from ECLAC (2018), the percentage of inhabitants who use the network grows each year, with 56% of inhabitants using the network in 2016. Although the service is mediated by income, the percentage of households with a fixed internet connection has increased in all countries and in all income quintiles. Similarly, the use of mobile broadband has increased, which has surpassed the use of fixed broadband.

Follow-Up

Of the visibility actions, the follow-up of the project will be via the online communication platform and with a global reach, which enables the dissemination of information on the activities of the Ibero-American countries that are part of ALIBER.

In coordination with the partners, the news and their follow-up will be directed to the Ibero-American institutions and organizations related to health and advocacy regarding the existence of the organizations of RDs in Ibero-America.

In terms of the information service, the database will be able to offer reports of all kinds: types of consultation, types of performance activity, number of those affected by diseases and per

country, available resources in each country, experts, consultations by age, gender, type of user, among others. It is about making the most of said information.

Budget Proposal

PLANNED EXPENDITURE			
HUMAN RESOURCES			
Position	Gross Salary	Social Security	Total
Service Coordinator (full working day: 6 months)	9,310.00 €	2,886.10 €	12,196 €
1 Support Technician (half working day: 6 months)	4655.00 €	1443.05 €	6,098 €
Subtotal Human Resources			18,294 €
MATERIAL AND TECHNICAL RESOURCES			
Computer assets and equipment			1,133.33 €
Promotional material			2,000.00 €
Software, office automation and antivirus			618.00 €
Database			3,000.00 €
2 Computer systems, 2 telephones and 2 printers			2,500.00 €
Subtotal Material and Technical Resources			9,251.33 €
MANAGEMENT			
Telephone and ADSL			1,600.00 €
Mail and courier service			700.00 €
Insurances			200.00 €
Subtotal Management			2,500 €
EXTERNAL SERVICES			
Design and maintenance of website			800.00 €
Tax, Accounting and Labor Consultant			1,500.00 €
Maintenance of domains and servers			500.00 €
Subtotal External Services			2,800 €
ALLOWANCES AND TRAVEL			
Allowances and travel			1,500.00 €
Subtotal Allowances and Travel			1,500 €
OTHER			
Contingencies and other			2,000.00 €
Subtotal Other			2,000 €
TOTAL BUDGET			36,345.48 €

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ANNEXES

Annex 1. List of ALIBER partners per country

ARGENTINA	Federación Argentina de Enfermedades Poco Frecuentes (Argentine Federation of Infrequent Diseases) (FADEPOF)
BRAZIL	<p>Associação Dos Familiares, Amigos E Portadores De Doencas Graves (Association Of Family, Friends and People With Serious Illnesses)</p> <p>Casa Hunter (Hunter House)</p> <p>Associação Brasileira De Paramiloidose (Brazilian Association Of Paramiloidosis)</p> <p>Instituto Unidos Por La Vida (United For Life Institute)</p> <p>Associação Brasileira De Pacientes De Esclerose Sistêmica- Abrapes (Brazilian Association Of Patients With Systemic Sclerosis - Abrapes)</p> <p>Associação Brasileira de familiares e amigos de portadores de Hipertensão Arterial Pulmonar (Brazilian Association of Family and Friends of People with Arterial Pulmonary Hypertension)</p> <p>Associação Alagoana dos Familiares e Amigos do Mucopolissacaridoses (Alagoas Association of Family and Friends of Mucopolysaccharidoses)</p> <p>Associação Brasileira do Síndrome do Cornélio Lange (Brazilian Association of Cornelia Lange Syndrome)</p> <p>Instituto Vidas Raras / Antes APMPS DC (Rare Lives Institute / Previously APMPS DC)</p> <p>Instituto Contemplo (Contemplo Institute)</p>
CHILE	<p>Fundación Chilena De Pacientes De Enfermedades Lisosomales (Chilean Foundation For Patients With Lysosomal Diseases)</p> <p>Corporación De Apoyo A Pacientes Con Enfermedades Hipofisiarias (Corporation To Support Patients with Pituitary Diseases)</p> <p>Linfoangioma Chile (Lymphangioma Chile)</p> <p>Federación Chilena de Enfermedades Raras (Chilean Federation of Rare Diseases)</p>
COLOMBIA	<p>Fundación Iberoamericana De Artrogriposis Múltiple Congénita Y Otras Enfermedades No Comunes (Ibero-American Foundation For Congenital Multiple Arthrogyriposis And Other Uncommon Diseases)</p> <p>Observatorio Interinstitucional De Enfermedades Huerfanas (Inter-Institutional Observatory For Orphan Diseases)</p> <p>Fundación Apoyo Solidario A Pacientes Con Enfermedades Raras (Solidarity Support Foundation For Patients With Rare Diseases)</p> <p>Así Colombia (Colombia Like This)</p> <p>Asociación Colombiana De Pacientes Con Enfermedades De Depósito Lisosomal (Colombian Association Of Patients With Lysosomal Storage Diseases)</p> <p>Fundación Cronicare (Cronicare Foundation)</p> <p>Fundación CDLS Colombia (CDLS Colombia Foundation)</p> <p>Fundación Colombiana para Enfermedades Huérfanas o Poco Frecuentes (Colombian Foundation for Orphan or Rare Diseases)</p> <p>11qLatinoamérica (11q Latin America)</p> <p>Fundación Colombiana para la Esclerosis Múltiple y Otras Enfermedades (Colombian Foundation for Multiple Sclerosis and Other Diseases)</p>
ECUADOR	<p>Fundación Ecuatoriana Para El Síndrome De Turner (Ecuadorian Foundation for Turner Syndrome)</p> <p>Fundación Ecuatoriana De Pacientes Con Enfermedades De Depósito Lisosomal (Ecuadorian Foundation For Patients With Lysosomal Storage Diseases)</p> <p>Fundación de Apoyo a Enfermos con Trastornos Hipofisiarios (Foundation to Support Patients with Pituitary Disorders)</p>
SPAIN	<p>Federación Española de Enfermedades Raras (Spanish Federation of Rare Diseases) (FEDER)</p> <p>D'Genes Asociación de Enfermedades Raras (D'Genes Association of Rare Diseases)</p>



	<p>Asociación de Familiares y Afectados de Lipodistrofias (Association of Family Members and People Affected by Lipodystrophies) (AELIP) Asociación Española de Raquitismos y Osteomalacia Heredados (Spanish Association of Inherited Rickets and Osteomalacia) Fundación Poco Frecuentes (Infrequent Foundation) Asociación Española de Porfiria (Spanish Association of Porphyria)</p>
GUATEMALA	<p>Fundación En Pro De Niños Con Trastornos Del Crecimiento Y Enfermedades Raras (Foundation For Children With Growth Disorders And Rare Diseases) (PROCRECE) Fundación Metamorfosis En Pro De Niños Y Jóvenes Con Hemofilia (Metamorphosis Foundation For Children And Young People With Hemophilia) (FUNDAMET) Rett Guatemala</p>
LATAM	<p>Sociedad Latina de Hipertensión Pulmonar (Latin Society of Pulmonary Hypertension) (SLHP)</p>
MEXICO	<p>Organización Mexicana De Enfermedades Raras (Mexican Organization of Rare Diseases) (OMER) Asociación De Corazón Por K Síndrome De Turner (Heart Association for K Turner Syndrome) Alianza Mexicana de Familias de Von Hippel Lindau (Mexican Alliance of Von Hippel Lindau Families) Fundación GENES Latinoamérica (Latin American GENES Foundation)</p>
NICARAGUA	<p>Asociación Nicaragüense de Distrofia Muscular Alan Prado (Nicaraguan Association of Muscular Dystrophy Alan Prado)</p>
PANAMA	<p>aYOUdas Panamá</p>
PARAGUAY	<p>Asociación Paraguaya de Esclerodermia y Enfermedades Autoinmunes (Paraguayan Association of Scleroderma and Autoimmune Diseases)</p>
PERU	<p>Federación Peruana de Enfermedades Raras (Peruvian Federation of Rare Diseases) (FEPER) Esperantra Asociación de pacientes con Angioedema Hereditario del Perú (Association of Patients with Hereditary Angioedema of Peru) Asociación de Raquitismos y Osteomalacia Heredados Peru (Association of Inherited Rickets and Osteomalacia Peru)</p>
PORTUGAL	<p>Federação das Doenças Raras de Portugal (Federation of Rare Diseases of Portugal) (FEDRA) Associação Nacional de Deficiências Mentais e Raras (National Association of Mental and Rare Disabilities) (Raríssimas)</p>
URUGUAY	<p>Asociación Todos Unidos por las Enfermedades Raras Uruguay (Association All United by Rare Diseases Uruguay) (ATUERU) Asociación Civil Síndrome Prader Willi del País (Civil Association of Prader Willi Syndrome of the Country) Federación Uruguaya de Instituciones de Discapacidad (Uruguayan Federation of Disability Institutions) (FUDI)</p>
VENEZUELA	<p>AC Humberto Da Silva Fundación Venezolana de Hipertensión Pulmonar (Venezuelan Foundation for Pulmonary Hypertension) Mundo Marfan Latino (Marfan Latino World) Fundación de Familias con Enfermedad de Wilson “Ninoska Hernández” (Foundation for Families with Wilson’s Disease “Ninoska Hernández”)</p>

