The Journal of Sociology & Social Welfare

Volume 45
Issue 1 *March*

Article 11

2018

The Importance of Social Work in the Latin American Association Movement of People Affected by Low Prevalence Diseases

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This article analyses the experiences of the association movement in Latin America that brings together individuals and families with rare (RD) or low prevalence diseases (LPD). It also looks at their needs from global health, social, research and education perspectives. The nature of social work assessments and interventions in rare diseases helps us better understand the needs of people with RDs or LPDs and facilitates the creation of associations. Social work aims at consolidating the social fabric that will lead to the recognition of RDs as a health and social priority at an international level.

Key Words: Rare Disease (RD), Low Prevalence Disease (LPD), Latin American Alliance of Rare Diseases (ALIBER), Social Fabric, Association Movement—Social Work

Experiences of the Low Prevalence Diseases Association Movement in Latin America

The concept and the definition of 'rare diseases' are fundamentally based on criteria of prevalence and severity. Rare diseases are those in which there is danger of death or chronic disability, and low incidence (less than 5 cases per 10,000 people in Europe), according to the European Commission (Comisión Europea, 1999) and Posada, Martin-Arribas, Ramirez, Villaverde, and Abaitua (2008). We can find other names, such as low frequency disease, minor incidence disease, invisible disease, forgotten disease, orphan disease, uncommon disease, or catastrophic disease. In Latin America, the most frequent name is *low prevalence disease.* The patients and the associations that support them emphasize the importance of realizing that rare diseases can affect anyone at any stage of life. As the president of the Association of Relatives and those affected by Lipodystrophies (AELIP), says, "It's not strange to suffer from a rare disease. The disease is rare, not the people who suffer them" (Pérez de Tudela, 2013). From the medical perspective, rare diseases are characterized by a large number and wide diversity of disorders and symptoms that vary not only from one disease to another, but also within the same pathology. The same conditions can have different clinical manifestations on different persons. Many disorders have a remarkable number of subtypes. It is estimated that there are more than 7,000 rare diseases affecting the patients' physical, mental and sensory capabilities.

The rare diseases (RD) association movement has gained considerable strength and recognition in public forums in many countries (Carrión, Echandi, Banon, & Pastor, 2015). RDs are not circumscribed to races or countries; they can appear at any stage of life and anywhere in the world. It is estimated that 42 million people are affected by RDs in Latin America (ALIBER, 2016). The Latin American Alliance of Rare Diseases (ALIBER) is a nonprofit coalition that brings together several organizations of families and patients of Rare Diseases (RD) or Low Prevalence Diseases (LPD) that exist in Latin America.

In 2013, the Year of Rare Diseases in Spain, the Association for Rare Diseases D'Genes and the Spanish Federation of Rare Diseases (FEDER) created ALIBER, with the aims of sharing

knowledge and best practices, and improving the situation of people and families living with a rare disease in Latin America. D´Genes began this initiative after many requests for information from Latin American countries, which made evident the need for it. The Information and Orientation Service (SIO) of FEDER also received an increased number of requests for information and other assistance from Latin American countries (Arcos, Solves, & Rius, 2016). Since 2002 the SIO responded to 3564 queries, as detailed in figure 1.

Figure 1. Distribution of queries on RD from Latin America received by SIO-FEDER since 2012.

Country	No. of visits
Argentina	961
Mexico	571
Colombia	465
Peru	326
Chile	323
Venezuela	196
Ecuador	104
Uruguay	90
Cuba	86
Brazil	66
Guatemala	64
Bolivia	45
Portugal	45
Costa Rica	37
Panama	32
El Salvador	31
Spain	29
Honduras	23
Paraguay	23
Nicaragua	21
Dominican Republic	19
Andorra	7
TOTAL	3564

Source: SIO FEDER (http://www.enfermedades-raras.org/)

It became clear that there was an urgent need to establish a joint collaboration among Spanish-speaking countries to respond to this demand. The 2012 agreement among the FEDER foundation, the Colombian Federation of rare diseases (FECOER), and the Spanish Foundation of Help for Guatemala provided a strong precedent. This agreement marked the beginning of this international collaboration. During 2012 and 2013, FEDER worked with Latin America to contact other organizations of patients interested in participating in the project. The end of 2012 witnessed the agreement among the Portuguese Federation of Rare Diseases (FEDRA), the Mexican organization of Rare Diseases (OMER), and the Colombian Foundation CRONICARE (Carrión et al., 2015, p. 116).

Since then, D´Genes, together with Mrs. Elizabeth Zabalza, Director of the Spanish Foundation for Help to Guatemala; Mr. Javier Guerra, Delegate of Mundo Marfan Latino; and Mrs. Claudia Delgado, Delegate of FEDER, planned the first Latin American meeting of rare diseases, which was held in October 2013 in Totana, Murcia, Spain. At this event, the Latin American Alliance of Rare Diseases (ALIBER) was created and its first Board of Directors was elected. Its mission included the creation of a network of organizations for patients with rare diseases prevalent in Latin America, the coordination of actions to strengthen the alliance, creating public awareness of RDs, and representing RD patients in local, regional, national and international agencies. This created a forum of permanent collaboration for sharing knowledge, experiences, and best practices in the areas of health, education, and work (ALIBER, 2016).

As for its vision, ALIBER seeks to be a coalition that brings together and empowers the different associations of RD patients in Latin America that defend the rights of the patients and their families. It disseminates information about legal rights and inequalities affecting people with rare diseases in different parts of the world. Towards this end, ALIBER works with other agencies and governments to improve the rights and the quality of life for those families. For the first time, in 2014, the International Day of Rare Diseases was held, and a Decalogue for the defense of the rights of people with rare diseases in Latin America was created. The Second Latin American Congress on Rare Diseases was held in Lisbon, Portugal, in 2014; the Third Latin American Congress on Rare Diseases was held in Guadalajara, Mexico,

in July 2015; and the Fourth Latin American Congress on Rare Diseases was held in Montevideo, Uruguay in September 2016 (ALIBER, 2016).

It is worth noting that the first congress established the guidelines for the subsequent ones (Bañón & Fornieles, 2013). The First Latin American Congress on Rare Diseases was held on October 14-18, 2013, in Totana, Murcia, Spain. It was organized by D'Genes and the Spanish Federation of Rare Diseases and convened professionals as well as delegates from associations. Participating entities included the Colombian Federation of Rare Diseases (FECOER), the Argentinian Federation of Diseases of Low Prevalence (FADEPOF), the All Together Rare Diseases Association Uruguay (ATUERU), the Mundo Marfan Latino, CRONICARE Foundation of Colombia, the Ecuadorian Foundation of Patients with Lysosomal Storage (FEPEL-DASHA), the Mexican Federation of Rare Diseases (FEMEXER), the Argentinian Support Group for Patients with Rare Diseases (GADAPER), the Mexican Association of Patients with Lysosomal Storage (ACOPEL), the Mexican Organization of Rare Diseases (OMER), the Paolist Association of Mucopolysaccharidosis and Rare Diseases (AMPS e Rarissimas) of Brazil, the Federation of Rare Diseases of Portugal (FEDRA), the Orientation and Information Service of Guatemala, and the Augusto Turenne Foundation of Uruguay. As can be seen, this international congress brought together a wide range of organizations.

Without a doubt, the development of an international movement is crucial to promoting solid and enduring improvements in diagnosis, treatment, and equitable access to social and psychological educational resources. For this reason, the common goal is to make this network as wide as possible. The first congress linked the above-mentioned agencies with EURORDIS, the European rare diseases organization. We must ensure, however, that the conclusions and agreements reached during these congresses lead to significant action. One of the most important outcomes of these congresses has been the buy-in of all members of ALIBER and the election of a Board of Directors. Most significantly, the congress generated a climate of cooperation that will permeate all future work. It is vital to develop joint projects between Spain and Latin America, and to disseminate positive work models and best practices to other countries

(ALIBER, 2016). For this reason, it is also important to share information about negative experiences and failed initiatives.

ALIBER is currently composed of 26 organizations that represent 497 associations in 14 countries. Spain is the country with the highest number of associations (Spanish Federation of Rare Diseases, FEDER: 320 associations), followed by Argentina (Argentinian Federation of Diseases of Low Prevalence, FADEPOF: 62), Mexico (Mexican Organization of Rare Diseases, OMER: 19), Uruguay (All Together Rare Diseases Uruguay, ATUERU: 17), and Portugal (Federation of Rare Diseases of Portugal, FEDRA: 16) (ALIBER, 2016).

The work of ALIBER in partnership with nonprofit organizations of Latin America has increased public awareness of rare diseases and has turned them into more of a political, health and social priority by the national health systems of ALIBER-member countries (ALIBER, 2016). Victims of rare diseases and their relatives are protagonists in the consolidation of this international movement. This movement and related associations have come to answer their questions and respond to their needs.

Features and Purpose of Organizations Related to Rare Diseases

The following are some of the features that characterize the organizations within the rare diseases association movement in Latin America: (a) they were founded by patients or their relatives; (b) they have arisen from the need to create a meeting point and common work arena; (c) they enable patients to contact each other and meet specialists in their particular health area; (d) they offer patients access to treatments and information about their pathology and rights; and (e) they aim to influence public policies and generate changes in the public health systems that may improve the quality of life for affected people.

Patient organizations play a key role in the new biopsychosocial model of health. These patient organizations are being recognized for their knowledge of patients' needs, their ability to collaborate, and their experience in putting RDs on the agenda of every country's health systems (Borrell-Carrió, Suchman, & Epstein, 2004). The primary purpose of RD organizations is to collaborate with health professionals in the care of patients.

The strategic objectives of ALIBER are: to increase public awareness of rare diseases as social, educational, work and health priorities; to represent people with rare diseases in Latin America; and to empower its member agencies.

In view of the above objectives and in response to its organizational mission and vision, ALIBER seeks to:

- Increase social visibility through:
  —Strengthening its strategic communication through its web page and other social media
  —Sensitizing key players in RD such as professionals, students, patients and governments
  —Bringing together key players in this movement through international congresses on Rare Diseases.

- Defend Human Rights through:
  —Establishing working relations and negotiations with the pharmaceutical industry for access to orphan drugs
  —Promoting the creation of alliances with organizations related to health and disability in Latin America
  —Increasing political influence and social mobilization in an effort to incorporate RD in health plans in Latin America
  —Promoting international cooperation
  —Creating a document identifying each country's policy in terms of RD health, education, work and research

- Provide Training and Disseminate Knowledge through:
  —Training a critical mass of professionals, doctors and researchers through the Latin American RD School
  —Promoting a model of care in RDs in each country through consensus
  —Establishing Alliances and Solidarity Networks to stimulate the multilateral cooperation of all sectors and actors related to RD in Latin America

- Effectively Manage Associations through:
  —Knowing and managing the challenges of the associations' movement in every country. This includes identifying the needs and problems related to RDs and their demands in terms of social participation
  —Promoting the creation of RD organizations and associations in Latin America
  —Promoting the participation of associations in international RD congresses.

- Engage in Social Action through:
  —The empowerment of associations and the development of RD Information and Orientation Services (SIO). SIO identifies the needs of affected people and thus develops a social action through improvement proposals based on knowledge of RD reality (ALIBER, 2016).

Challenges to Safeguarding Equality of Health Rights

According to Palau (2010), and Saltonstall and Scott (2013), the main health problems of people suffering from RDs include misdiagnosis, lack of information, lack of scientific knowledge, lack of the appropriate quality care, high cost of the few existing drugs and treatment, and inequality of access to treatment and care, all of which will be explored below.

Misdiagnosis

Delays between the onset of the first symptoms and proper diagnosis involve high risks to patients' health. Misdiagnoses often lead to inadequate treatment in what could be described as the pre-diagnostic labyrinth. The average time between the onset of the first symptoms and diagnosis is 5 years, and for 20% of the sufferers, it can be 10 years or more (FEDER, 2009, pp. 43-44). Many more patients are still waiting for a diagnosis. FEDER (2009) received more than 3,160 queries from people without diagnosis seeking help. According to the ENSERIO

study (Study of Health and Social needs of people with a RD and their families), more than 40% of people with a diagnostic delay receive inadequate treatment or no support whatsoever (FEDER, 2009, p. 47).

Consequences of the delay can be detrimental to patients and their families. This delay deprives patients of the needed therapeutic intervention and often results in physical, intellectual and psychological deterioration. All of these could have been avoided with a more rapid diagnosis. Moreover, a late diagnosis leads to greater difficulty in accessing social help (Avellaneda et al., 2007).

Lack of information, scientific knowledge, and appropriate quality care

Useful and timely information is needed about the diseases and about places where proper help can be obtained. It is important to highlight that the lack of qualified professionals in the RD area represents a serious problem (Esteban, Ruano, Guerra, & Motero, 2015). Lack of scientific knowledge makes it difficult to develop therapeutic tools, defining intervention strategies and identifying appropriate medicines and medical approaches. Without combining the different knowledge fields associated to RDs, such as physiotherapy, nutrition, psychology, social work, etc., people affected can live for years in unstable conditions, without competent medical attention or rehabilitation. They often remain excluded from the health care system, even after diagnosis.

High cost of the few existing drugs and treatment and inequality of access to treatment and care

The high cost of health care and drugs, combined with the lack of social benefits, causes an impoverishment of the family and drastically increases inequality of access to care for RD patients (Graf & Frank, 2015). There is unequal access to innovative treatments due to delays in price setting and refund decisions. Additionally, doctors lack experience, and few are involved in RD clinical trials. There is also lack of consensus about best treatments.

Human Rights Related to Education and Communication: Problems Integrating School, Work and Society

Rare or low prevalence diseases have significant adverse impacts on the physical health of the sufferers to the point of endangering their lives. This danger in combination with social and relevant psychological processes negatively impacts the well-being of the patients. RDs may lead to stigmatization, exclusion, low self-esteem, loneliness, and discrimination in affective relations, employment, finances, and/or the way they are represented in the media. In fact, when asked, patients often make reference to their social, educational and communication needs (FEDER, 2009; Jaeger, Rojvik, & Berglund, 2015; Pavol, 2015).

RDs have implications for all areas of life including education, job choice, leisure activities, and relationships with friends or one's partner. They can lead to stigmatization, isolation, exclusion from the community, difficulty in obtaining insurance (travel, life, mortgage, etc.), and reduction of job opportunities.

RDs affect education in particular. Fornieles et al. (2014), comment on the following 14 major topics that are relevant to RDs in Latin America:

- It is necessary to create interdisciplinary groups that consider rare diseases, gather experiences and offer solutions. All key players must be present in the group [pupils, teachers (also with rare diseases), parents (also with rare diseases), etc.].

- The lack of understanding of low prevalence diseases is common in schools. This ignorance spawns prejudices related to what students with these conditions can or cannot achieve.

- It is necessary to identify educational models that will effectively mainstream children with RDs. These models should lead to innovative projects and curricular adaptations.

- RDs must become part of primary and secondary education curricula. Because of their interdisciplinary nature, they can be incorporated into subjects as diverse as science, art, humanities, or new technologies.

- We must continue the efforts to share information and increase sensitization regarding RDs. This sensitization should focus on the search for equality and social justice in education, health and employment.

- Students with RDs represent a special challenge to their families. Educational institutions should make every effort to support families, and particularly siblings attending the same school. Families must be encouraged not to avoid the problem and not to overprotect these children.

- Students with RDs may become our future doctors, teachers, journalists or managers. Their education is important for our society in general, even though it represents a challenge. Challenges help people grow.

- We must pay special attention to the transitions between the different stages of education. Ignorance of RDs may result in the need to repeat educational content at the beginning of every semester or academic year, and may lead to the loss of educational support. It is particularly necessary to support students with RDs as they transition from secondary education to college and from college to higher university degree programs.

- Research on education and RDs must increase. It is important for researchers to contact persons with RDs who have been university students in order to identify the hurdles they encountered. Researchers are encouraged to visit educational institutions to interview students and teachers.

- Self-image during childhood and adolescence has not been taken sufficiently into account. This is

significant, given that victims of physically visible and invisible RDs may experience discrimination and bullying that may damage their self-image.

- On occasion, students with RDs are excluded from extracurricular activities, assuming that they cannot or do not want to participate. This may be due to teachers' reluctance. We should examine whether this problem can be solved with professional help or with the use of a mediator.

- There is a need to improve the collaboration between schools and hospitals, given that some pathologies require frequent hospitalizations for treatment or due to complications.

- RDs have become a new field in biomedical research. The same level of interest is necessary as we look at education. There is a need for original alternatives that may facilitate on-campus and off-campus education.

- Experts in teaching and pedagogy must collaborate with the Institutes of Education Sciences and Teachers Centers to generate proposals for new methodologies.

Problems in communication

During the last 3 years, the FEDER agency has conducted an exhaustive analysis of RDs in Spain (Solves, Bañón, & Rius, 2015). Without a doubt, the same should be done in Latin America, as it would help to gain a better understanding of what citizens know about RDs, as well as assess the level of exposure of RDs through the media in each country.

The role of communication media in the process of inclusion or stigmatization is obvious. We know from various studies on gender, race and disabilities that the media is capable of facilitating these processes. An analysis of the media suggests that despite the availability of online information on RDs, they remain

vastly unknown to the public at large. In a similar manner, despite progress in recent years, RDs remain an unfamiliar topic for journalists and society as a whole (Bañón & Requena, 2014).

As a result, information professionals face multiple barriers, such as ignorance, unknown terminology, technological difficulties, and health and social complexities (Vicente, 2011, p. 395). These barriers could lead to information errors or avoidance, given that these themes may be considered too complex for their audiences. On the other hand, the perception of RD patient associations and patient families is that not enough progress has been made related to media visibility. Although media exposure has increased, it seems to be mostly limited to the International Day of RD. As Bañón and Fornieles say, "As these actions only take place at a particular moment, they only leave a temporary footprint that fails to give a clear picture of the existence and nature of those diseases amongst the public" (2011, p. 12).

From this, it becomes clear that we must carefully examine the accuracy, ethics and adequacy of the RD content disseminated by media such as newspapers, radio, TV and Internet. At the same time, it is necessary to influence the priorities of journalists to make sure RDs receive the coverage they deserve. It is also important to generate guidelines to help us interact with the media. Bañón and Fornieles (2011, pp. 203-206) propose that:

- The growing interest in RDs shown by the Spanish media must be consolidated.
- We must enter a new phase of communication about RDs in that the quality of information must improve.
- Authoritative sources, including researchers and RD patient associations, must be properly used.
- There is a need to better integrate RDs into the affairs of communication media.
- Communication professionals need additional training in the area of RDs.

- We should not use sensationalism to portray patients with RDs nor describe them as people in need of compassion. They should not be perceived as strange people.

- The media should investigate what health authorities say and what they actually do in relation to RDs.

- The attention given to leaders that support RDs should not overshadow the defense of the rights of patients and their families, and working towards better diagnosis, medical research or therapies.

- Media professionals must remember that they are also agents for reporting incorrect actions in health, social or educational fields.

- Media representation of conflicts or collaboration between professionals or public administration and patients should be used to reflect upon good and bad practice.

Recommendations for Improving Patient Wellbeing

The following priorities have been adopted by ALIBER in Latin America

- Promote the inclusion of RDs in public health plans

- Promote RD research

- Disseminate RD-related information to patients, health workers and the general public

- Train health and social workers in RDs

- Recognize the unique social and health rights of RD patients

- Promote the creation of referral centers to improve access to and quality of care for RD patients
- Facilitate access to needed resources for a rapid diagnosis
- Encourage the development of and access to drugs and therapies for RDs
- Support the RD association movement
- Establish national and international collaborations related to RDs (ALIBER, 2016)

Consistent with the recommendations of Fernández, Lozano, and Riano (2015), we propose considering four proposals in the political arena:

- Survey Spanish public opinion on a regular basis and assess the needs of people with RDs in order to formulate social policies that respond to those needs.
- Secure the support of national, regional and local governments in the development of comprehensive plans related to their areas of competence.
- Provide training, increase sensitization, and coordinate specialized health and social services that respond to the diverse needs of people with RDs.
- Work towards the necessary coordination and reciprocal support that should exist between the different government agencies and representatives of civil society.

Conclusion

It is clear that without scientific research, there is no future or hope for the over 7,000 rare diseases currently known. The real challenge for research in rare diseases is to advance

the discovery of treatments and detection of new diseases. It is true that progress has been made (Ministerio de Sanidad y Consumo, 2003, 2006; Ministerio de Sanidad y de Política Social, 2009). However, it is also necessary to make progress in communication and in social and educational issues. It is estimated that between 6 and 8% of the world population may be affected by one of these diseases; that is more than 42 million people in Latin America and more than 350 million people worldwide (ALIBER, 2016).

To approach these problems, we need global exposure and coordinated efforts and resources at the local, regional and national levels. These efforts must involve government authorities, professionals, the pharmaceutical industry, the mass media, patient organizations and society in general (Organización Mundial de la Salud [World Health Organization], 2012). Likewise, a multi-faceted strategy is required to respond to the most urgent health problems through prevention, planning, and primary care (Garcia-Ribes, 2006). Social workers are key players in the association movement in Latin America. The increase in the number of associations and foundations in Latin America in recent years has resulted in the need to meet and share experiences, consolidating the Latin American Alliance of Rare Diseases (ALIBER) as a worldwide model.

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