

Project Proposal

Title: Quality of life and self-perceived needs in informal caregivers of patients with rare diseases: generating evidence for public policies in Chile.

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Introduction:

Most rare diseases have a genetic etiology and manifest themselves in childhood, generating a high burden of morbidity, disability and premature death (1-3). Their rarity and complexity make diagnosis and treatment difficult (3), so patients and families suffer a "diagnostic odyssey" (4), which leads to deterioration in health, economy and family interactions. Most of the time, a caregiver is required, a role that is generally assumed by a family member, who is devoted to caregiving activities and abandons his personal projects (5), with a high overload that impacts on his or her quality of life (6-9). In the case of caregivers of people with rare diseases, this is exacerbated by the high cost or non-existence of treatments, the difficulties in accessing diagnosis and specialized care, the prolongation of care for years and the fear of disease progression (10,11), even more so with the uncertainty experienced when the disease has not yet been diagnosed (12-14).

Although there is evidence on the importance of the well-being of caregivers in the process of accompanying patients (15), there are few studies and public policies that focus on them. In Chile, advances have focused on three strategies: financing of high-cost diseases (16); guaranteeing care for some RD and neonatal screening for phenylketonuria (3,17), but there is no specific regulation for patients with rare diseases, let alone for their caregivers. However, the recently elaborated "Proposal for a National Plan for Rare, Orphan or Infrequent Diseases" (18) represents an opportunity to establish a national commitment to address the problem in a comprehensive and continuous manner, and it is essential to generate evidence to facilitate this process.

Vision:

The support of informal caregivers is essential for people with rare diseases, not only for the development of activities of daily living, but also for access to diagnosis, treatment and rehabilitation. However, their support needs and the deterioration that the role of caregiver generates in their quality of life are not prioritized by public policies. In this context, this research seeks to contribute to the generation of evidence on this kind of "hidden patient" that is the caregiver, even more so in the case of rare diseases, where there is a lack of studies and comprehensive policies with transformative capacity, which would contribute to the achievement of the Sustainable Development Goal of "leaving no one behind".

Aims:

1. To investigate the quality of life, psychosocial support and health care access needs of informal caregivers of people with rare diseases, as well as recommendations for their improvement in Chile, from the perspective of different key actors: caregivers of people with and without a diagnosis, health professionals and decision-makers.
2. To analyze the public strategies for informal caregivers of people with rare diseases in Chile, in relation to the existing international recommendations on the subject.

Impact:

The project will generate evidence on the quality of life and needs of informal caregivers of people with rare diseases, relevant to consider in the processes of diagnosis, treatment and rehabilitation of patients, as well as the existing gaps between these needs and current public policies in Chile, within the framework of international recommendations on the subject. In this way, the results are expected to contribute to public policies in Chile.

Executive Plan:

Aim	Activities	Q1	Q2	Q3	Q4
To investigate the quality of life, psychosocial support and health care access needs of informal caregivers of people with rare diseases, as well as recommendations for their improvement in Chile, from the perspective of different key actors	Elaboration of interview guidelines and consents	X			
	Contacting key informants	X			
	Conducting interviews		X	X	
	Transcription		X	X	
	Qualitative analysis			X	X
To analyze the public strategies for informal caregivers of people with rare diseases in Chile, in relation to the existing international recommendations on the subject	Definition of search strategy	X			
	Search	X	X		
	Design of extraction matrix		X		
	Systematization of selected texts		X	X	
	Information and gap analysis				X

After developing this plan, the process of integrating the results will be carried out, also including quantitative information, the collection of which has not been included in this proposal, but it is also part of the researcher's doctoral thesis.

The researcher is not a G2MC member, but has the advice and support of Dr. Gabriela Repetto, Chile, who is a G2MC member. The project also involves working with patient organizations such as FECHER and FENPOF, and the mentoring of researchers from the Social Studies in Health Program of Universidad del Desarrollo, Chile, and Universidad de Ciencias Sociales y Empresariales, Buenos Aires Argentina.

Future funding:

The remaining activities correspond to analysis and integration of the information, so no additional funds are required, since they will be carried out as part of the researcher's doctoral thesis project. In addition, a quantitative component will be included that is not part of this proposal and is being developed within the framework of the project "Solving the unsolved: an interdisciplinary evaluation of personal, social and health system effects of the use of genomic strategies for rare diagnosis disorder", directed by Dr. Gabriela Repetto and financed by the National Fund for Scientific and Technological Development of Chile (Fondecyt).

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Budget

Budget requested = USD\$ 26,775

Program budget:

Categories	Description	Hourly rate per activity/unit value (USD)	Quantity	Total (USD)
Personnel	2 interviewers, for 20 hours each	\$75	40	\$3000
	2 transcriptionists, for 30 hours each	\$30	60	\$1800
	2 analysts, for 30 hours each	\$75	80	\$4500
	2 researchers for literature search and review, 50 hours each	\$75	100	\$7500
	2 researchers for literature systematization and analysis for 50 hours each	\$75	100	\$7500
Materials and supplies	Gift for interviewees	\$20	20	\$400
Travel and meeting support	4 trips for interviews are contemplated, with an approximate value of US\$ 200 each.	200	4	\$800
Miscellaneous costs	Contingencies (5%)	---	---	\$1275
Total				\$26775