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RARE DISEASES IN ARGENTINA: WHAT A SURVEY OF FAMILIES TELL US ABOUT THE DIAGNOSIS PROCESS

Authors: Liljesthröm M., Armando R., Maier B., Robert F., Zelikman D.
Fundación FOP (Fibrodysplasia Ossificans Progressiva) - info@fundacionfop.org.ar

INTRODUCTION

This work about diagnosis process is part of a scientific research entitled "*Rare Diseases in Argentina: a socio-sanitary, legal and scientific approach. Communication and policy proposals for reducing vulnerability*" which is conducted by *Fundación FOP* and supported by the *Agencia Nacional de Promoción Científica y Tecnológica* (part of the Argentinian Ministry of Science, Technology and Productive Innovation) , and two private foundations: *FUNI* and *Gianantonio.*

The research aims to produce information on rare diseases' situation in Argentina for feeding the development of appropriate policies focused both in the particular needs of the target population and the institutional, social, economic and cultural framework in which this collective is inserted.

Three tools have been designed in order to collect primary information,
1) a survey of physicians,
2) a survey of patients/parents of rare diseases organizations, and
3) a survey of families affected by rare diseases.

Having an accurate diagnosis allows interventions. Nevertheless, it is one of the stages which presents big difficulties to people suffering from rare diseases.
This work shows some preliminary results of survey 3), dealing with the diagnosis process from the very first symptoms up to the correct diagnosis.

OBJECTIVES

General (related to the survey of families):

- To produce statistically significant information on the most relevant aspects regarding situations faced by families with a member affected by a rare disease in Argentina, with the purpose of characterizing them according to, among other determinations, place of residence, socioeconomic status, main sources of information and type of health coverage.

Particular of the diagnosis process:

- To know features of the diagnosis process, trying to identify differences according to rare diseases groups and territorial level.

METHODS

The survey of families was conducted amongst those affected by 1 out of 12 selected rare diseases, 6 of them forming a "more rare" group (A) and the other 6 being a "less rare" one. (B)

(A)
Neurofibromatosis type 2
X-linked Agammaglobulinemia
Fibrodysplasia Ossificans Progressiva
Mucopolysaccharidosis type 1H
Gaucher disease
Fanconi Anaemia

(B)
Cystic fibrosis
Tuberous sclerosis
Duchenne muscular dystrophy
Marfan syndrome
Prader Willi syndrome
Crohn´s disease

Argentina´s territorial structure was taken into account when designing the sample. The inclusion of a group located in AMBA (Buenos Aires & its metropolitan area) and another one from the rest of the country keeps the relative population proportion.
The cases to be sampled were constituted with the collaboration of public and private national hospitals, physicians and patient/parents organizations. The research protocol was evaluated by their corresponding ethics committees. The age of the affected person was established between 0 to 30 years old, among other inclusion criteria. The questionnaire focus on perception of families for all issues.

RESULTS

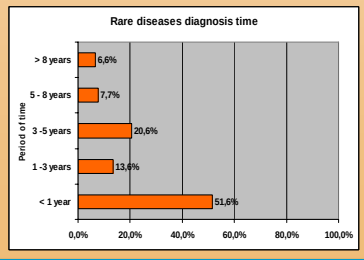
- Universe of sampled cases : 1707 individuals.
- Aleatory sample: 322 cases (keeping the same relative population proportion than in the rest of the country).
- Interviews were mainly performed in every family´s dwellings in:
- 20 provinces out of 24 existing in Argentina. Within them, 86 different sized towns (from less than 5000 to 1,2 million inhabitants, and the metropolitan region of Buenos Aires with 12 millions).

Out of results estimated for the whole sample and groups i) rare diseases, (more and less rare) & ii) territory (AMBA -Metropolitan Area of Buenos Aires- and rest of the country) the most significant are presented hereafter.

1.- Elapsed time when obtaining the diagnosis.

Total mean is higher than 2 years with no significant differences between the two considered groups. Nevertheless, inside each one of them elapsed time until obtaining a diagnosis increases remarkably in the range from 50% to 75% of each population.

	Rare Diseases diagnosis time		
	Mean (Months)	for 50% of respondents	for 75% of respondents
Total	27,7	11	38
More rare group	28	10	39,75
Less rare group	24,4	16	34

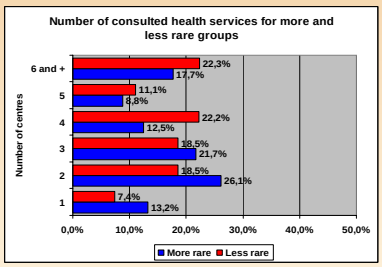


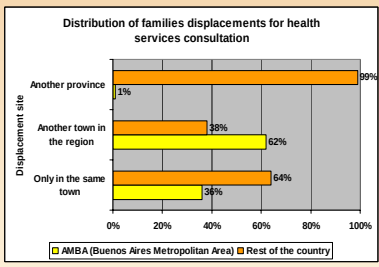
2.- Number and localization of health centres visited when searching diagnosis.

50% of total respondents consulted up to 3 centres, whereas considering 75% of them the number of centres increased to 5. More than 18% of total sample consulted 6 centres and more.

In order to obtain a diagnosis the "Less rare" group consulted more centres than the "More rare" one.

Looking at consulted centres location and corresponding displacements, 28% from the total consulted only same town centres, whereas 44% also traveled to another town in the same province and another 28% moved farther for a consultation in other provinces located centres As reflected in the graphic, 99% of displacements to other province were performed by the rest of the country located group.





3.- Misdiagnoses.

Almost 35% of the total sample expresses having found at least one diagnosis which resulted wrong time after. Most of this group obtained 1 misdiagnosis and a small proportion of it up to 4.

The "Less rare" group got the highest number of misdiagnoses. Additionally, whether more or less rare, time until obtaining a right diagnosis increases significantly in the range from 50 to 75 % of cases of the at least one misdiagnosis group with respect to the no misdiagnosis one.

Number of misdiagnoses/ patient	% of respondents
1	23,60%
2	8,1%
3	2,30%
4	0,70%
Total with misdiagnoses	34,70%
No misdiagnosis	65,30%

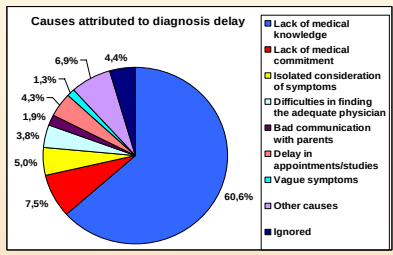
Misdiagnosis presence	More rare	Less rare
Yes	32,20%	59,30%
No	67,80%	40,70%

Diagnosis time for misdiagnosis group		
	for 50% of respondents (months)	for 75% of respondents (months)
Misdiagnosis	18	50
No misdiagnosis	8	32

4.- Feeling of diagnosis delay.

50% of the total sample states having felt elapsed time until obtaining a diagnosis had been excessive. The "Less rare" group gathers the biggest feeling of diagnosis delay -63%- against 48,8% of the "More rare" one.
Average elapsed time when obtaining the diagnosis is 39,18 months for the entire group which felt diagnosis delay.

Diagnosis time for "feeling of excessive delay" group		
Feeling of excessive delay presence	for 50% of respondents (months)	for 75% of respondents (months)
Yes	25	57
No	5	15



More than 75% of respondents consider aspects related to physicians (lack of knowledge, lack of commitment among others) as diagnosis delay causes.

54% of the group which felt diagnosis delay expresses having had related negative consequences.

Negative consequences of diagnosis delay	% respondents
Progress of the disease	47,50%
Birth of another affected child	11,90%
Psychological	32,70%

5.- Ultimate diagnosis obtaining.

63.7% of the sample obtained accurate diagnosis in the Metropolitan Region of Buenos Aires and the other 36.3% did so in the rest of the country.
Only 31% of the sample obtained that diagnosis in the same place of residence.
Only 50% of respondents expresses having had genetic advice after obtaining diagnosis.

Diagnosis obtaining place	% of respondents
Same town	31,1%
Another town in the same region/province	44,4%
Another province	23,9%
Another country	0,6%
Total	100%

CONCLUSIONS

These partial results show us the fact that for a high percentage of respondents, accurate diagnosis obtaining process involves several difficulties, whose extent is comparable to other countries one. Particularly, inhabitants of the interior of Argentina live an inequity situation due to long displacements access to specialized services requires. Regarding the "Less rare" group, whereas significant differences in diagnosis obtaining process times were not registered, they had to consult a higher number of centres and their proportion of misdiagnoses was higher too.

All of it means a great weight not only to affected families but also to the whole health system what, remarkably in countries with scarce resources and persistent health problems linked to social lacks, strengthens the need of making a more efficient system.