THE REALITY OF UNDIAGNOSED "RARE PATIENTS": CLAIMING FOR AN IMPROVEMENT OF THEIR LIFE-QUALITY

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THE REALITY OF UNDIAGNOSED "RARE PATIENTS": CLAIMING FOR AN IMPROVEMENT OF THEIR LIFE-QUALITY

Introduction

Living with a rare disease can be a difficult challenge. Living with a rare undiagnosed disease is even more complicated. There is no yet an official register of the people with a rare disease lacking a diagnosis, although scarce projects worldwide are trying to build a registry of undiagnosed patients (1). Therefore, its representativeness in the group of patients with rare diseases is unknown and it is very complex to carry out an investigation that requires their participation.

With the aim of knowing what is nowadays the psycho-social and working reality of this group and their families in Spain, FEDER (Federación Española de Enfermedades Raras) has done a comprehensive study with this group which has been published recently (2).

This work is a continuation of the previous ones (3, 4) and deepens and broadens several results obtained in the previous studies, mainly those related with the more negative perception that this undiagnosed patient has of their general situation, due to delay diagnostic and lack of information about their disease. This drives to social, psychological and work impacts, which have not been yet adequately studied.

Methodology

The methodology follows those from previous studies (3,4) and analyse quantitative primary data through an online survey provided to people with rare and undiagnosed disease. The questionnaire was distributed mostly online, with a link in the websites of several entities (FEDER; CREER and SpainRDR), press release, newsletter and specific workshops.

From a total amount of 1576 questionnaires answered by patients with rare diseases from the previous studies (4), 50 of them (3,18%) were from undiagnosed patients. Subsequently another 28 undiagnosed patients were added. Therefore, this study is based on the responses obtained from **78 undiagnosed patients**.

Questionnaire were based on Socio-demographic data, personal and technical assistance, diagnosis and health care, education and training, employment and income, household features, use of resources and perception of the current situation.

Results

Results obtained in this study are proportional to the sample size, both from the group of diagnosed patients (n = 1576), and of the not diagnosed (n = 78). Although the sample of 1576 patients include 50 undiagnosed persons, the sampling error is, in any case, negligible. The majority of the patients were between 0-15 years; therefore, the questionnaire was majority answered by their parents (68% of the cases). 97% of the sample belong to Spanish citizens. Almost 50% of the sample has 4 members in the family.

The more important results of this undiagnosed patients related to the group (diagnosed +undiagnosed) from the previous reports has to do with a higher perception of dissatisfaction with: healthcare (56% vs 40%), multidisciplinary approach (58% vs 50%), coordination between health services (63% vs 59%), coordination between health and social services (73% vs 55%), coordination between health and educational services (60% vs 50%) and coordination between the Reference Centres, Services and Units in the National Health System (51% vs 47%).

The people who participated in the study present a diagnosis delay from 6 to 9 years (30%), with a considerable percentage of people who have suffered a diagnostic delay of more than 15 years (29%). The main identified cause by these undiagnosed people was the lack of knowledge of the pathology (87%). Main consequences were not receiving any treatment or support (29%), aggravation of the disease or its symptoms (25%), not receiving an adequate treatment (22%) and need of psychological attention (17%).
Regarding medical specialities, the most consulted by undiagnosed patients is neurology (68% vs 47%) and ophthalmology (62% vs 38%). In this group of patients, the genetic consultation is essential (62% vs 22%) and, in many cases, it is even repeated multiple times until the diagnosis is reached. This study does not show differences in the number of hospitalizations for both groups in the last two years.

Travelling to get a diagnosis (40% vs 25%), access to a treatment (26% vs 20%) or a medication (10% vs 6%) outside their own Autonomous Community, which is not only related with working/familiar situation but also with afforded expenses is a common reality for these undiagnosed patients. On the contrary, the expenses that need to be cover by public administrations is mainly dedicated to travelling to hospital (49% vs 73%), hospital fees (42% vs 75%) and surgery (43% vs 68%). Other expenses which are mainly covered by patients are respite care, hearing aids and glasses and contact lens and nursing home care.

Other difficulties are related with their greater lack of information about the Reference Centres, Services and Units from the National Health System (80% vs 61%) and also to the way to access to medicines (50% vs 35%) and other medical devices (39% vs 31%).

In relation with those medications in special situations, foreigner, orphan and compassionate use, their used is limited to a 4%, 4% and 2%, respectively.

Undiagnosed people have greater percentages of intellectual and motoric disability which leads in higher levels of dependency (53% vs 41%). Lapse time between the dependency assessment request and its real assessment was higher in this group (36% vs 26%), the same for the one between the assessment and the public economic and health assistance (51% vs 47%).

Although access to the System for Autonomy and Care for Dependency service to both group of patients is very similar, undiagnosed patients have greater limitations in the development of basic activities of their daily live (leisure and free time, educational or work activity, self-care, communication with other people, etc.).

A 73% of the undiagnosed people participating in the study claim to have impairments due to their illness to develop basic activities of daily living. For this reason, the family (mainly the mother) assumed their care, being marked as first choice in 70% of the cases studied. This data reflects the same situation as the ENSERio study.

The analysis of the parameter related to their inclusion and social environment, such as the

adaptation of housing, the situation regarding education and training, access to employment, the impact of the disease on the family budget, also show a worse subjective perception of their situation and greater discrimination of the undiagnosed rare disease patients.

Conclusions

Main conclusions of this study show that people lacking a diagnosis clearly express a worse perception about their medical, social, and psychological situation compared to those perceived by the patients with global pathologies of rare diseases analysed in the previous ENSERio study.

This group show greater limitations in the development of basic activities among people without diagnosis, in all of the activities mentioned. Families, especially mothers, are the ones who mostly take care of these patients. This general feminization of health care leads to an increasing gender wage gap and decreasing wage conditions for women.

This enquiry shows a greater degree of ignorance of the Centres of Expertise in the National Health System from undiagnosed patients, which makes it even more difficult for them to access to services that improve their vital situation. These Centres of Expertise not only treats specific pathologies, but also develop specific procedures and techniques. In addition, the delay they go through to reach a diagnosis for their disease, which has been called "'the diagnostic odyssey" (1), clearly impairs their perception and their clinical situation.

There is still much work to be done to improve the well-being of people with rare diseases. This includes advocacy on important public policies as well as educational outreach to medical professionals and students.

Link here for further information

1- Adachi, T., Kawamura, K., Furusawa, Y., Nishizaki, Y., Imanishi, N., Umehara, S., Izumi, K. & Suematsu, M., 2017. Japan's initiative on rare and undiagnosed diseases (IRUD): towards an endo to the diagnostic odyssey. European Journal of Human Genetics 25: 1025-1028.

2- FEDER, 2019a. Analysis: the reality of undiagnosed patients. https://obser.enfermedades-raras.org/wp-content/uploads/2019/04/INFORME-V.Última_II-Fase-Estudio-ENSERio_Sin-diagnóstico.pdf

3- FEDER 2017. Estudio sobre situación de Necesidades Sociosanitarias de las personas con Enfermedades Raras en España. https://obser.enfermedades-raras.org/estudio-enserio-2016-2017/

4- FEDER 2019b. Análisis autonómico de necesidades socio-sanitarias de las personas con enfermedades poco frecuentes: la demora diagnóstica. https://enfermedades-raras.org/images/ESTUDIO_ENSERIO_COMUNIDADES_AUTNOMAS.pdf