



SUMMARY: Until now, there have been several studies carried out by FEDER (Federación Española de Enfermedades Raras) on the group of people with a rare disease (1,2). We have gone one step further by investigating what is the socio-health care and their inclusion and social involvement reality of undiagnosed patients. Any study with this group of people is complex, given their representativeness in the group of patients with rare diseases is unknown, due to the lack of records. This comprehensive study (3) is a continuation of those previous ones and deepens and broadens several results obtained in them, mainly those related with the greater negative perception that this undiagnosed patient has of their general situation due to the delay in their diagnosis and the lack of information about their disease. 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. This analysis show that people lacking a diagnosis clearly express a worse perception about their medical, social, and psychological situation; they show greater limitations in the development of basic activities and a greater degree of ignorance of the networks and Centres of Expertise in the Spanish National Health System.

METHODOLOGY: Following the methodology from previous studies done with the overall group of people with rare disease (diagnosed +undiagnosed) we analyse quantitative primary data through questionnaires obtained between October 2016 to September 2018, through phone call, face-to-face or online surveys provided from 78 undiagnosed rare diseases patients (or to the authorized person in the case of minors). Results obtained in this study are proportional to the sample size, both from the group of diagnosed patients (n = 1526), and of the not diagnosed (n = 78). We show results comparing undiagnosed patients vs. the group of diagnosed patients from the previous reports (1,2). Taking into account the registration in the Spanish Program of Cases of Undiagnosed Rare Diseases (SpainUDP), which has registered a total of 112 cases, this means that this study has a confidence level of 95% and a margin of error of 6,2%.

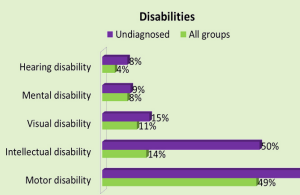
- » The majority of the patients were between 0-15 years (69% of the cases)
- » 97% of the sample belong to Spanish citizens
- » Almost 50% of the sample has 4 members in the family.
- » 83 % of the sample correspond to families with just one person undiagnosed.

RESULTS:

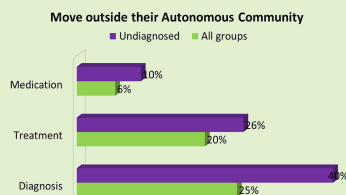
• **Early diagnosis:** In the case of undiagnosed patients 81% have a diagnostic delay of more than 6 years, mainly due to the lack of knowledge about the disease (87%). Main consequences of this late diagnostic were:

- » Not receiving any treatment or support (29%)
- » Aggravation of the disease or its symptoms (25%)
- » Not receiving an adequate treatment (22%)
- » Need of psychological attention (17%).

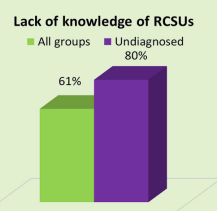
• **Disability:** undiagnosed people have higher percentages of disability, especially in relation to intellectual disability:



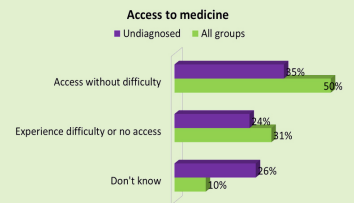
• **Travelling:** undiagnosed patients must travel further to obtain a diagnosis, treatment, or a medication.



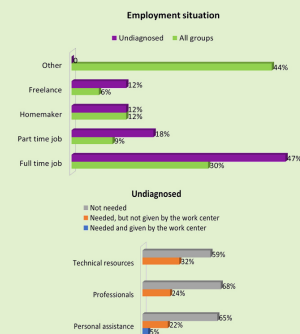
• **Centres of Expertise:** lack of knowledge of undiagnosed patients about this Centres is very high.



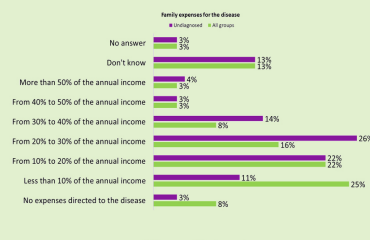
• **Access to medicines and to medication in special situations:** half of the undiagnosed patients have difficulties accessing treatment or do not know what to do for it. Scarce use of medicines in special situations.



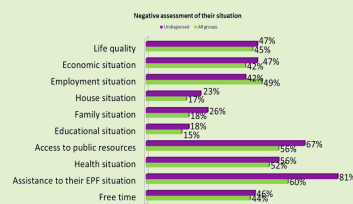
• **Employment:** because 69% of the cases are 0-15 years old, most of them still continue their training. People working mostly state the need of personal/product support (but not provided).



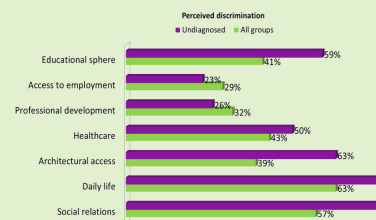
• **Expenses:** p82% of undiagnosed people studied value high or very high the cost of their rare disease.



• **Perception of dissatisfaction:** worst perception has to do with their public health care and access to public resources. More than half of the people express their dissatisfaction with Government services



• **Discrimination:** Higher discrimination is perceived with daily life and social relations, followed by architectural access



CONCLUSIONS:

This is the first study aimed at finding out the social, health and employment situation of undiagnosed people in Spain. It should be noted that more than half of the participants are minors, so important answers, such as perception of dissatisfaction, come from their parents or guardians.

Delay to reach a diagnosis is still very high, which clearly impairs their perception and their clinical situation. This entails a greater number of trips in search of a definitive diagnosis and treatments, a greater expense in clinical and familiar treatments, increased hospitalizations, surgeries, difficulties to access to medicines, clinical trials, medication in special situations and so on. This group also express greater limitations in the development of basic activities and a worse perception about their medical, social, and psychological situation. In addition, they express a greater feeling of discrimination compared to that perceived by the patients with global pathologies of rare diseases analysed in the previous ENSERio study (2).

This comprehensive study confirms that the key factor that would definitively improve the quality of life and clinical status of people without a diagnosis lies in reducing the time frame for achieving a diagnosis of their disease. All the necessary efforts must be directed towards this.

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REFERENCES:

- 1- 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/>
- 2- FEDER, 2019a. Análisis autonómico de necesidades socio-sanitarias de las personas con enfermedades poco frecuentes: la demora diagnóstica. https://enfermedades-raras.org/imagenes/ESTUDIO_ENSERIO_COMUNIDADES_AUTONOMAS.pdf
- 3- FEDER, 2019b. Analysis: the reality of undiagnosed patients. https://obser.enfermedades-raras.org/wp-content/uploads/2019/04/INFORME-VÚltima_Il-Jase-Estudio-ENSERIO_Sin-diagnóstico.pdf