OBSERVATORIO SOBRE ENFERMEDADES RARAS

ANALIZAR

ACTUAR

DETECTAR

TRANSFORMAR

REPORT

Analysis: The reality of undiagnosed patients

2019

feder
entidad de utilidad pública
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

OBSER
OBSERVATORIO SOBRE
ENFERMEDADES RARAS

GOBIERNO DE ESPAÑA
MINISTERIO DE SANIDAD, SERVICIOS SOCIALES E IGUALDAD
SECRETARÍA DE ESTADO DE SERVICIOS SOCIALES E IGUALDAD

POR SOLIDARIDAD
OTROS FINES DE INTERÉS SOCIAL

Translated by María Leiras Carmona

INDEX

GROUNDS AND CONTEXTUALIZATION

The 2009 ENSERio study on the situation of socio-health needs of people with rare diseases in Spain was the first comprehensive study to address rare diseases in different social spheres and fields, from a different methodological perspective than previously stated.

There have been previous approaches to the reality of people who suffer from rare diseases. The lack of knowledge regarding these pathologies is related to a delay in diagnosis and thus a delay on getting an appropriate treatment, disorientation in terms of school environment, lack of awareness in terms of the pathology and the people who has to live with it, consequences in the work environment, economic impact and more.

The 2009 ENSERio study was approached from a triangular methodology, both qualitative and quantitative. The aim of this study was getting results which might prove transposable and being able to get conclusions based on quantitative data that did not exists until that moment. This study obtained 715 questionnaires filled by people who suffer from a rare disease and their families, 10 interviews and 4 focus groups.

Seven years later and thanks to the new developments on Spain in terms of investigation, the growth of the association members and the social and regulation changes, an update of the study has been proposed in order to analyze the evolution of this group.

This update was powered by FEDER[1] and CREER[2] dependent on the IMSERSO[3] during the years 2016-2017. This time the sample of the first study was duplicated obtaining a total amount of 1576 questionnaires, 14 interviews and 8 focus groups. This sample came from FEDER, CREER and SpainRDR (Spanish Rare Diseases Registries Research Network). The methodology used was the same as in the first ENSERio study in order to make it possible to compare the results and analyze the possible trends and developments with the first reference work.

In the ENSERio study of 2017 it was shown, in a first approximation to the results, that the group of undiagnosed people faced more obstacles related to the lack of information about their disease. The study stated, undiagnosed people had a more negative perception of their situation. Consequently, their degree of dissatisfaction with the system and health care was one of the highest compared to other groups of minority pathologies. At the same time, they had fewer possibilities to access and maintain employment and suffered more opportunity costs than other diagnosed groups, as well as a greater perception of discrimination.

---

[1] Point of reference for people with rare diseases in Spain.
[2] Care center for people with rare diseases in the region of Burgos, Spain.
[3] Social services for old people in Spain.

Considering all these circumstances and after having all the data from the study, in 2018 we emphasize the importance of keeping the research and analyzing the group of undiagnosed patients, having a bigger sample each time and analyzing the principal aspects of their daily life.

OBJECTIVES

The five key points used to develop the objectives of the ENSERio study are:

- ❖ **Describe the situation and needs** of people who suffer from rare diseases and their families.
- ❖ Know what **resources and services** this group uses as well as the personal support and supporting products they need or have.
- ❖ Identify which **aspects help a person with a rare disease to be socially integrated** as well as knowing those aspects which suppose an obstacle for them.
- ❖ **Compare the results** of this study with the results obtained on previous investigations, especially with the results obtained on the **first ENSERio study**, in order to see if there has been an **evolution**.

This report is focused on undiagnosed patients and their situation. With the aim of adapting the ENSERio study to this group, the objectives of the study are as follow:

- ❖ **Describe the socio-sanitary situation** of people who suffer from rare disease and that had no diagnosis between 2017 and 2018.
- ❖ Know the **resources and services** used by the analyzed group as well as the personal support and supporting products they need or have and that are related to the lack of diagnosis.
- ❖ Identify how the lack of diagnosis affects this group in terms of **social inclusion** as well as knowing their personal barriers.
- ❖ **Compare** the results of the 2017 ENSERio study in its first stage with the global data.

METHODOLOGY

Analyze of quantitative primary data through an online survey provided to people with rare diseases or to the authorized person in the case of minors and people with big limitations.

DIMENSIONS OF THE QUESTIONNAIRE

Socio-demographic data:

- Basic: age, gender, nationality
- Specific: diagnosis, disability degree

Personal assistance and technical assistance:

- Area, frequency and support persons
- Needs and use of technical assistance
- Dependency assessment and use of the System for Personal Autonomy and Care of Dependent Adults (SAAD)
- Other economic benefits

Diagnosis and health care:

- Adequacy of the received treatment
- Medical consultation, examinations and used services
- Knowledge and valuation of the Reference Centers, Services and Units (RCSU) of the National Health System (NHS)
- Displacement to receive healthcare

House: characteristics and adjustments

Education and training:

- Received education
- Actual education (in the case they are still studying)
- Characteristics of the study center
- Technical assistance or educational support

Employment and income:

- Employed, unemployed, inactive…
- Characteristics of the employment situation
- Personal and familiar income
- Disease related costs: percentage and entries

Use of resources:

- Overall
- Specialized
- Organizations membership

Perception of the current situation:

- Situation in the different areas
- Discrimination perception: overall and by different areas.

These points were also analyzed in the first ENSERio study, although new variables were added in order to cover all the objectives mentioned before.

ENSERio UPDATE: FIRST STAGE (2017) AND SECOND STAGE (2018) OF DATA COLLECTION

During the first stage the questionnaires were filled between October 2016 and March 2017. The questionnaires were sent via FEDER, CREER and the SpainRDR through their enterprise networks, meetings, conferences and assemblies.

The questionnaire was distributed online with a link in the websites of the mentioned entities and through the FEDER social network, press release, newsletter, specific workshops and other organizations. This was the main source of questionnaires collection (1552 questionnaires). 49 questionnaires were done via phone call or physically, which means that a total amount of 1606 questionnaires were filled. From the 1601 questionnaires, 25 had to be discarded because they did not fulfill the criteria which left us with a total of 1576 valid questionnaires. From this sample, only 50 questionnaires (3 % of the total) were from undiagnosed people.

In the second stage, from May to September 2018, 28 questionnaires were filled by undiagnosed patients making a **total amount of 78 questionnaires filled by undiagnosed patients**.

The information in the second stage was retrieved by the SpainRDR through the Spanish Rare Disease Research Institute (IIER) and the Biomedical Research Center Rare Diseases Network (CIBERER). The online questionnaire and the associative movement of undiagnosed patients' organizations associated to FEDER was transferred to CIBERER.

- Frequency tables used to detect the basic needs of this group and to have an approximate knowledge of their condition in the different fields of study.
- Bivariate analysis of the detected needs and demographic variables of interest in order to deepen the study.

FACT SHEET: ANALYSIS OF THE 78 UNDIAGNOSED PATIENTS

Because there is no formal register of the people who suffer from rare disease and are lacking diagnosis, the population that we are working with is unknown in different ways. If we accept the population registered at the Spanish Undiagnosed Rare Disease Cases Program (SpainUDP), the total amount is: 112 cases. This means that:
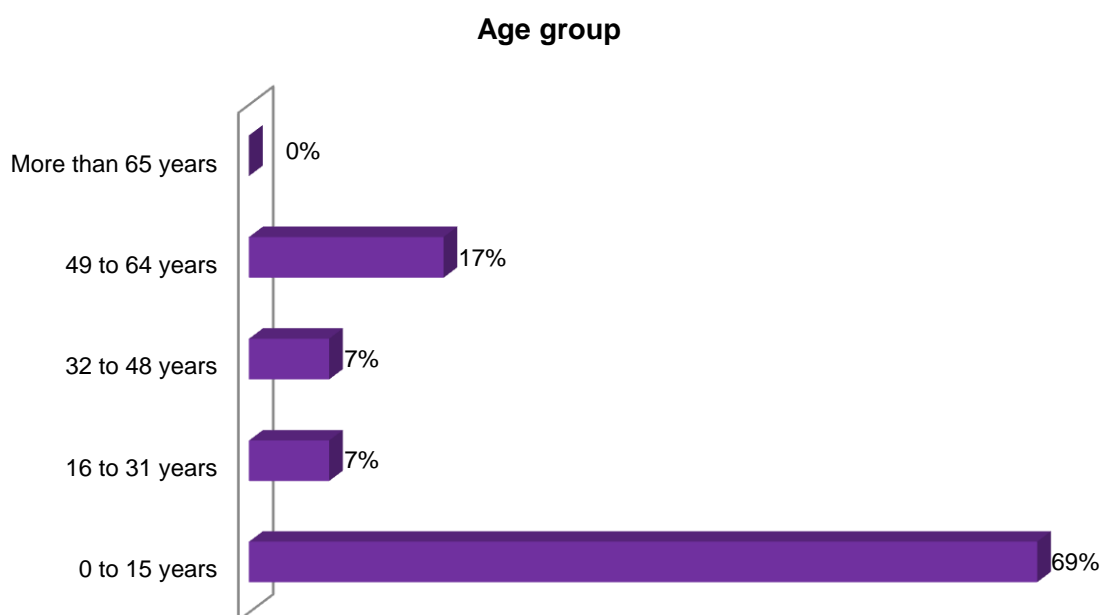
For the 78 collected questionnaires, the study has a 95 % level of trust and a 6,2 % margin of error.

TESTED SAMPLES' PROFILE: UNDIAGNOSED PEOPLE (78 CASES)

Demographic data: age and gender

The sample is divided proportionally between men and women, at 50 %.

Likewise, it is distributed by group of ages, being the majority patients between 0 to 15 years old (69 % of the sample):

Age group

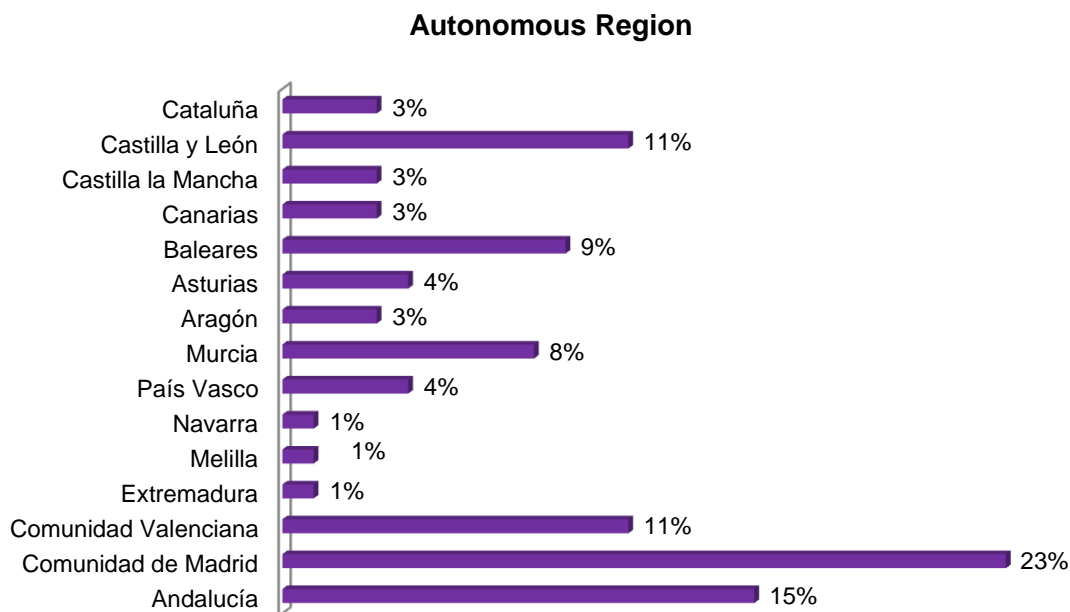| Age group	Percentage
More than 65 years	0%
49 to 64 years	17%
32 to 48 years	7%
16 to 31 years	7%
0 to 15 years	69%

Given the age of the undiagnosed patients, it is common for the survey to be answered primarily by the parents of the person with RD (68% of cases).

In one out of four cases (25%) the person with RD answers by his/her own means, in 3% with the support of an assistant and in another 3% the person who answers is another informant (another relative, tutor, caregiver...).

Nationality and Autonomous Region of residence

97 % of the sample has Spanish nationality, being that 3 % of British nationality. By Autonomous Regions, on the other hand, given that no quota sampling has been carried out, the sample is distributed as follows: with the highest percentage being those people residing in the Community of Madrid, followed by Andalusia, the Valencian Community and Castilla y León.

Autonomous Region

Region	Percentage
Cataluña	3%
Castilla y León	11%
Castilla la Mancha	3%
Canarias	3%
Baleares	9%
Asturias	4%
Aragón	3%
Murcia	8%
País Vasco	4%
Navarra	1%
Melilla	1%
Extremadura	1%
Comunidad Valenciana	11%
Comunidad de Madrid	23%
Andalucía	15%

People who make up the family unit

Almost 50 % of the sample has a four members family. The following highest percentage is around three family members (23 %). The amount of people with just one family member (4 %) or six family members or more (1 %) is a minority.

In these households, 83 % of the sample affirms that just one person suffers from a rare disease. However, there are cases where two people suffer from RD (8 %), three or four people (7 %) and there is a single case where more than six family members suffer from RD (1 %).

UNDIAGNOSED PEOPLES' CONCEPTION

1. SOCIO-HEALTH CARE

1.1.Satisfaction with healthcare

The ENSERio study indicated the importance of adequate health care in improving people's life quality.

In the case of the sample analyzed in the ENSERio study, with all groups of pathologies, half of the people asked (51%) were satisfied with the health care. The percentage of dissatisfaction, on the other hand, was 40%.

Analyzing the perception of the undiagnosed patients, a higher percentage of dissatisfaction is observed (56%) with respect to the overall figures of the group of people with rare diseases, the percentage of satisfaction was lower (28%).

This means, people who lack a diagnosis clearly present fewer positive perceptions about the health care received related to their illness, compared to those perceived by the total or global pathologies of RD analyzed in the ENSERio study.
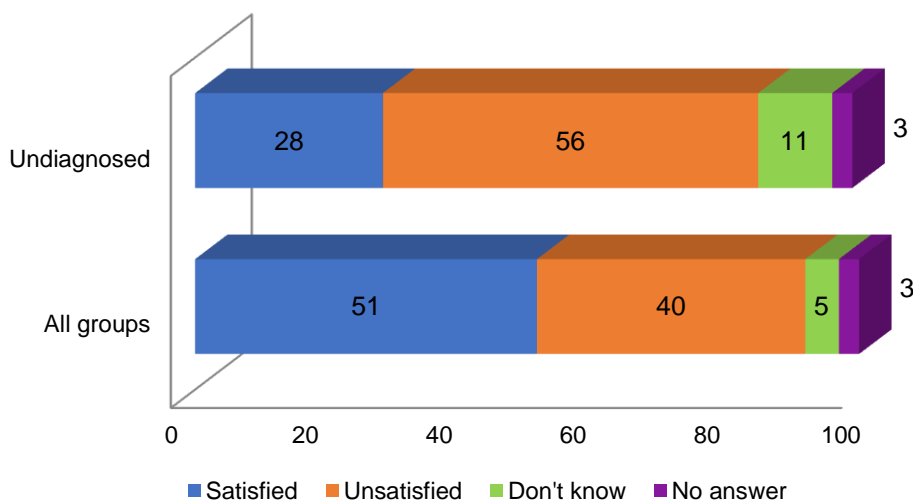

Chart 1. Percentage of satisfaction with healthcare. Comparison between undiagnosed patients and all groups from the ENSERio study.

Source: *ENSERio study.*

Another fundamental aspect for the evaluation of health care, and which is key for people who suffer a pathology, is multidisciplinary coordination and approach.

A multidisciplinary [4] approach is the coordination between health services and other institutions. The level of satisfaction for the undiagnosed collective was 24% (who consider this approach "good" or "very good") and the dissatisfaction reaches 58% (who consider this approach "bad" or "very bad"). These dissatisfaction figures are somewhat higher than those presented in the overall ENSERio study, although the difference is not as notable as the data analyzed for satisfaction with health care:
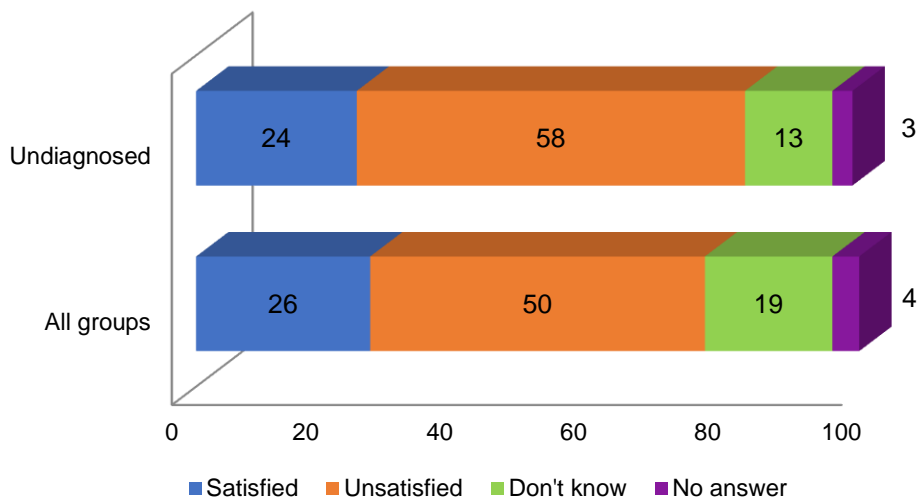

Chart 2. *Percentage of satisfaction with the multidisciplinary approach. Comparison between undiagnosed patients and all groups of the ENSERio study*

Source: *ENSERio study*

Moreover, in the ENSERio study, which compared the different groups of pathologies, this circumstance was also pointed out by identifying the group of undiagnosed patients as the one with the highest proportions of dissatisfaction with the health care received, together with the group of infectious and parasitic diseases.

In order to know more about this, it is key to analyze the satisfaction with the coordination in a concrete way:

- Coordination between different health services

To coordinate different health services, i.e. primary care, specialized care, hospitals, etc., the level of dissatisfaction among people lacking a diagnosis is 63%, compared to 59% in the overall ENSERio data.

---

[4] The ENSERio study considers the multidisciplinary approach as the coordination between the different health services (primary care doctors, specialists, outpatients, hospitals, between the different hospital services and the RCSU, where appropriate), the social services and the educational centers.

On the other hand, the percentage of satisfaction is also lower, with 22% for data on the group without diagnosis and 29% for data collected in the overall ENSERio.
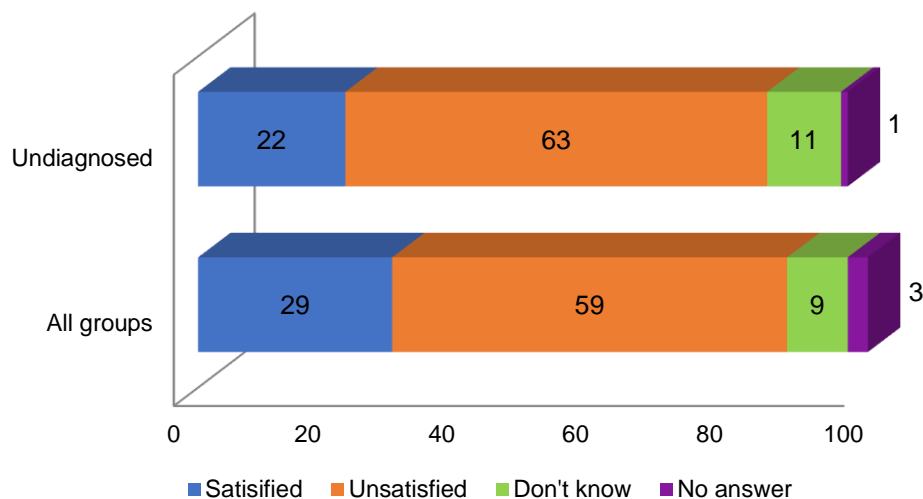

Chart 3. Percentage of satisfaction with the coordination between different health services. Comparison between undiagnosed patients and all groups from the ENSERio study

Source: ENSERio study

- Coordination between the Reference Centers, Services and Units (RCSU) in the National Health System (NHS)

The assessment of coordination between the Reference Centers, Services and Units, which will be analyzed later in this report, is characterized by a high percentage of ignorance ("Don't know" answer option). 31 % of the sample chose this option when assessing the coordination of these services. Of the people who rated coordination, 11.3% rated it positively and 51 % rated it negatively.

This assessment is somewhat more negative than the recorded in the ENSERio. The positive perception of this coordination is 21% and the negative perception 47%. Likewise, 27% of the undiagnosed patients chose the "Don't know" option.
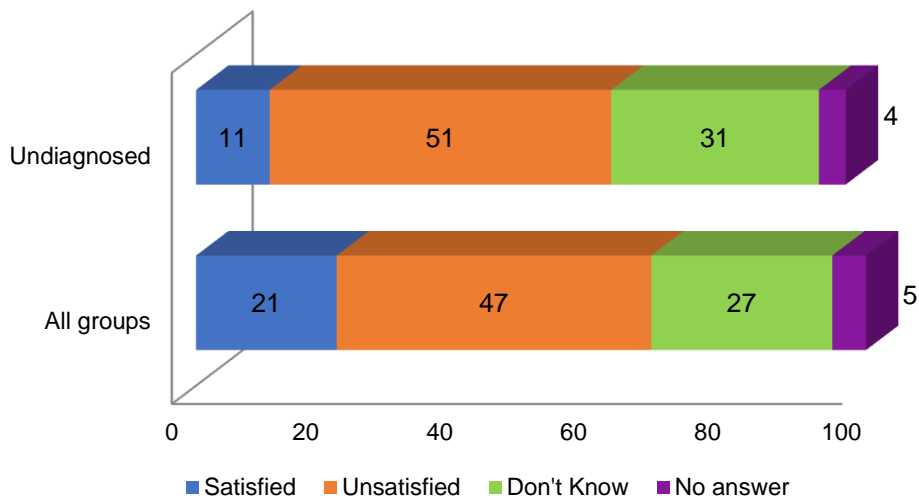
Chart 4: *Percentage of satisfaction with the coordination between the RCSUs. Comparison between undiagnosed patients and all groups from the ENSERio study.*

Source: ENSERio study.

- Coordination between health and social services.

Since networking has been identified from different sources as essential for families, coordination of health services with other services is a key point for analysis.

Thus, in 55 % of the cases in the ENSERio study, for all pathologies, there was a negative assessment of this coordination between health and social services. The undiagnosed people, on the other hand, claimed to have a negative opinion about this coordination in 73 % of the cases, a figure visibly higher than the one presented in the overall pathologies, and being only 9 % of the people in this group who valued it as positive.
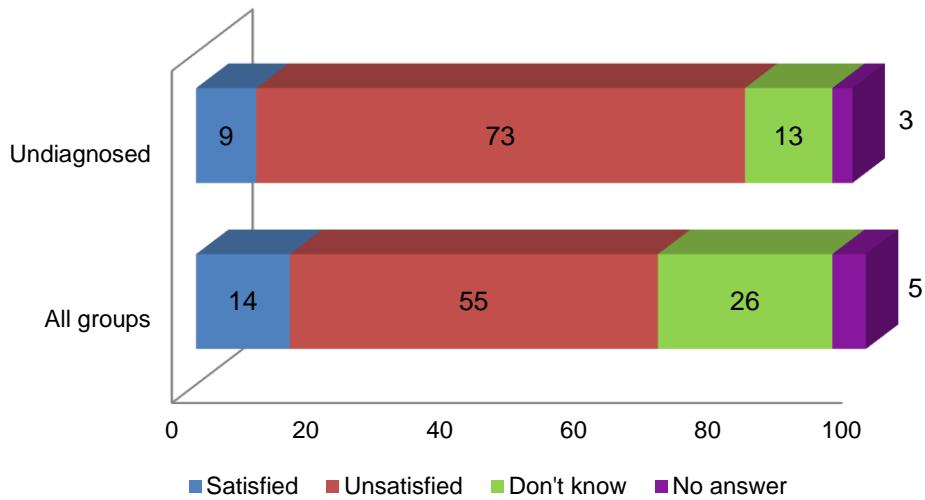
Chart 5. *Percentage of satisfaction with the coordination between health and social services. Comparison between undiagnosed patients and all groups from the ENSERio study*

Source: *ENSERio study*

- Coordination between health and educational services

In this line of evaluation of network work, coordination with educational centers receives the worst assessment, with 60 % of negative perception on the part of undiagnosed patients who participated in the study. On the other hand, the perception is only positive in 17 % of the cases, with no response to the "very good perception" option within this category.

In the ENSERio study, for the global figures for all groups of pathologies, the results also presented negative evaluations, in their majority percentages, collecting a negative perception for half of the sample.
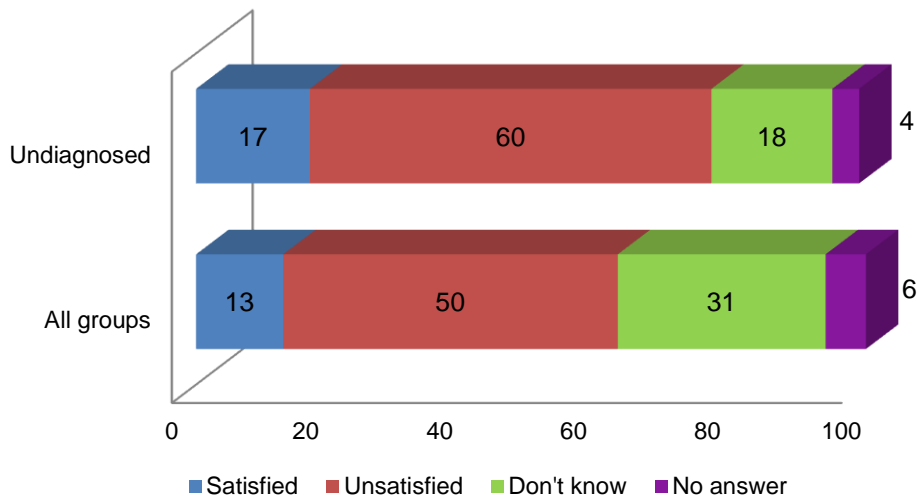
Chart 6. *Percentage of satisfaction with the coordination between health and educational services. Comparison between undiagnosed patients and all groups from the ENSERio study.*

Source: *ENSERio study.*

UNDIAGNOSED PEOPLE HAVE WORSE CONSIDERATIONS ABOUT NETWORKING, AT ALL LEVELS OF COORDINATION.

Percentage of dissatisfaction with networking attention
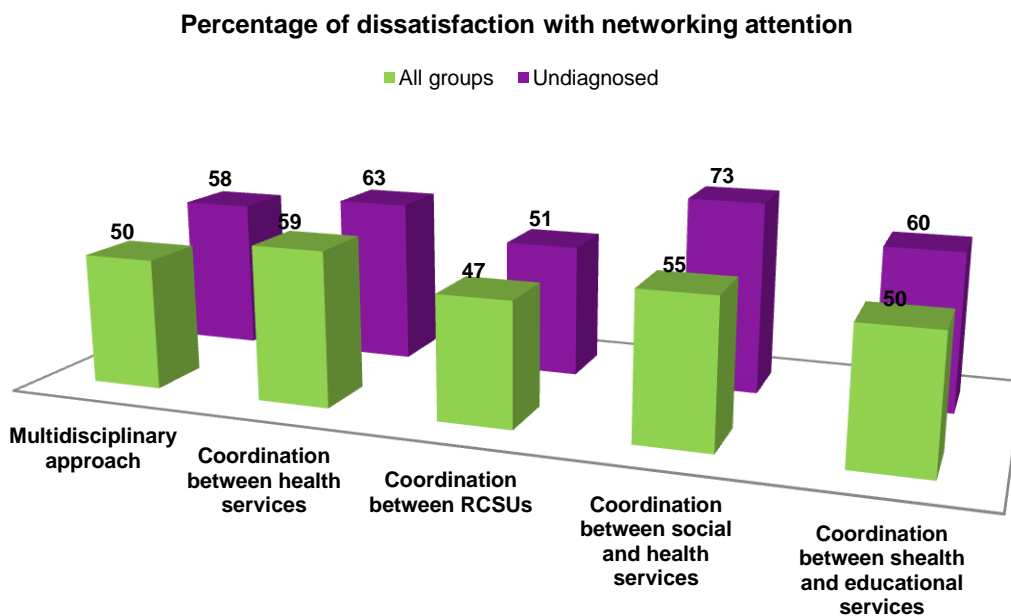
1.2.Diagnosis

1.2.1. Waiting time to get a diagnosis

The people who participated in the study present a diagnosis delay that goes from 6 to 9 years (30 %), although there is a considerable percentage of people who have suffered a diagnostic delay of more than 15 years (29 % of the sample).

One fifth of these people stated that they had a diagnosis delay of 3 and 5 years. And finally, almost a quarter of them had been with symptoms related to a pathology for 10 to 15 years, the diagnosis of which remains unconfirmed.

Late diagnose
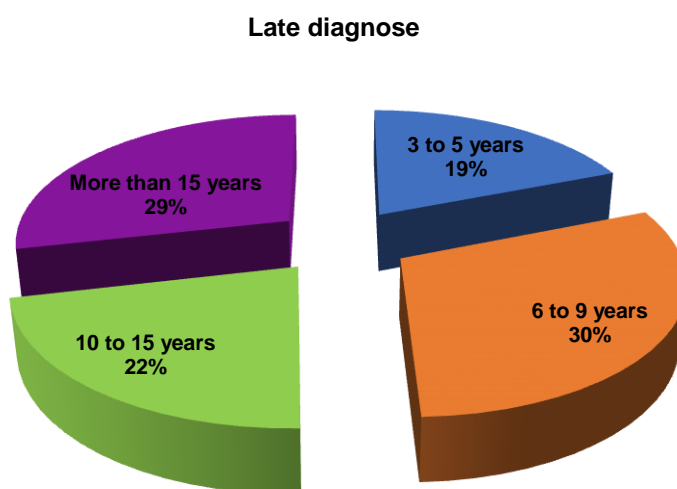
Graphic 1. *percentage of people distributed by time of delay in diagnosis, in groups of years.*

Source: *ENSERio study.*

IDENTIFIED CAUSES

The main identified cause is the lack of knowledge on the pathology.

Causes of late diagnose

- Economic evaluation at the hospital — 7%
- Administrative causes — 3%
- Economic evaluation at the Autonomous Region — 3%
- Lack of knowledge — 87%
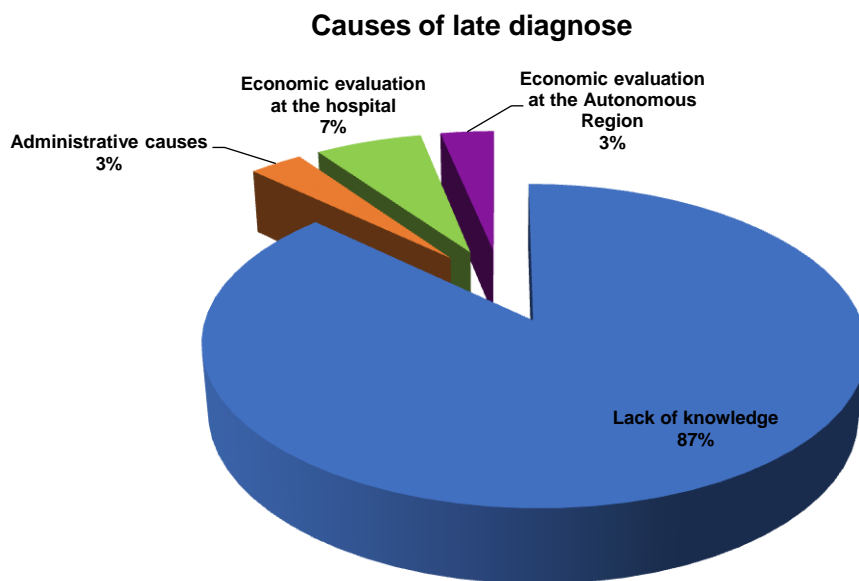
Graphic 2. Percentage of perception of the late diagnosis.

Source: ENSERio Study.

Moreover, some people point out the lack of referral to a specialist, for not assessing their symptoms according to their severity and the reluctance to request tests from the genetic service. However, all these causes could be the result of the lack of knowledge of the disease, its symptoms and causes.

In the ENSERio study, although opinions were more distributed among the different causes, the main cause identified for the diagnostic delay was the lack of knowledge about the disease (73 %).

1.2.2. Consequences of a delayed diagnosis

What are the consequences of these delayed diagnosiss and what impact is it having on people without a confirmed diagnosis?

Consequences of a late diagnose
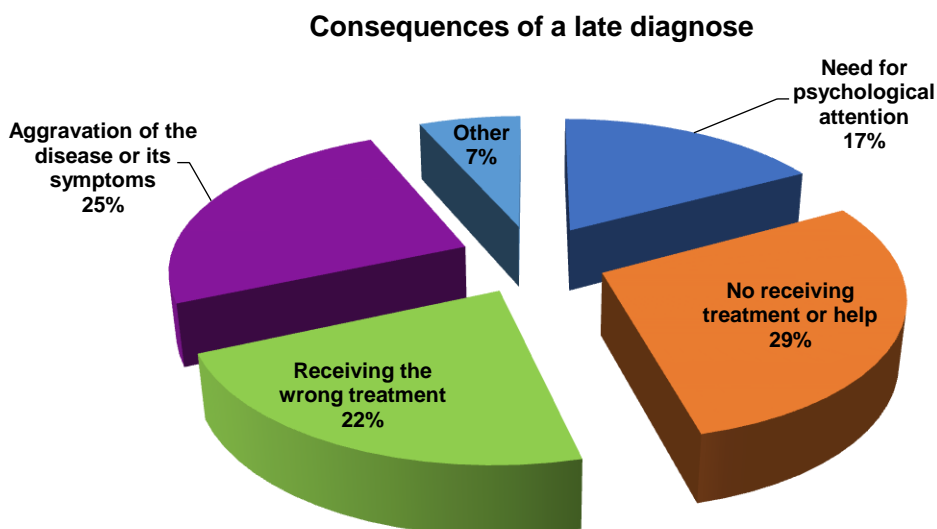
Graphic 3. *Consequences of a late diagnosis.*

Source: ENSERio Study.

Unlike what happened in the ENSERio study, for people without a confirmed diagnosis participating in this report, the main consequence is not receiving any treatment or support (29 %). The second consequence is the aggravation of the disease or its symptoms, in one out of four cases (25 %), followed by 22 % of the cases that does not receive an adequate treatment and, finally, the need for psychological attention in 17 % of the cases.

Taking these data into account, and making a comparative analysis with the global data provided by the 2017 ENSERio study, it can be concluded that people without a confirmed diagnosis claim to need or to have needed psychological attention to a greater extent (17 % as opposed to 15 % reflected in the ENSERio) and have a perception that delayed diagnosis creates obstacles for them to access support or adequate treatment, as opposed to other consequences (51 % as opposed to 47 %).

1.3. Treatment

Access to appropriate treatment for the disease is closely related to obtaining a confirmed diagnosis. This way, the group of undiagnosed people, although they coincide with the overall figures in the percentage of people who do not have treatment or who have it, but it is inadequate (49 % and 47 %, respectively), differ from the total pathologies in that a much lower percentage of people who claim to have access to the treatment they need (10 % as opposed to 34 % of the ENSERio).

Following the same path, the percentage of people who answered, "Don't know" (33 %, one out of three people) didn't even knew if they had an adequate treatment, which is linked to the absence of a diagnosis.

I have the correct treatment
I don't have the correct treatment
I have a treatment, but I don't think is the correct one
Don't know
No answer

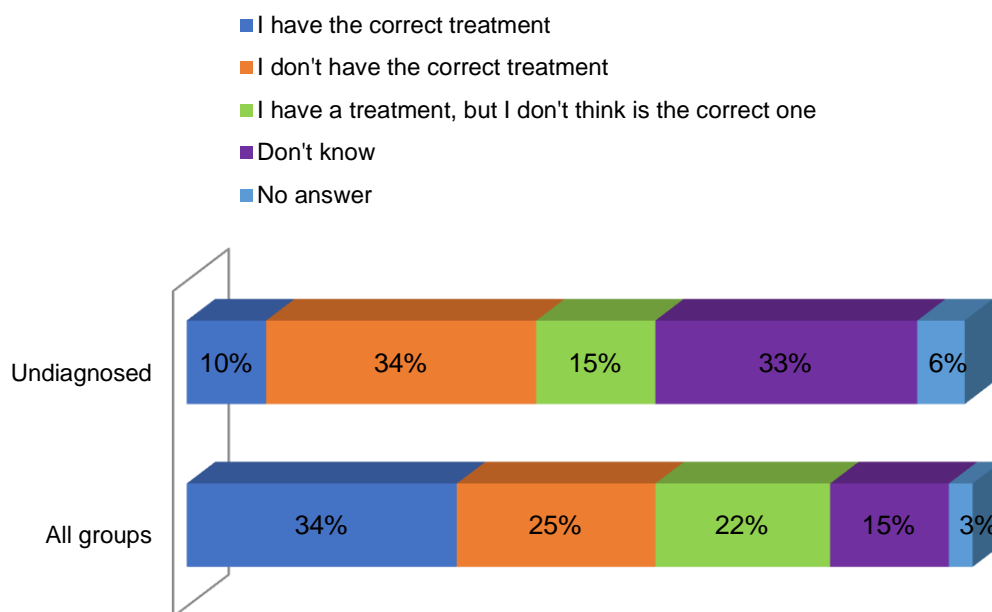|	I have the correct treatment	I don't have the correct treatment	I have a treatment, but I don't think is the correct one	Don't know	No answer
Undiagnosed	10%	34%	15%	33%	6%
All groups	34%	25%	22%	15%	3%

Chart 7. *Percentage of Access to treatment. Comparison between undiagnosed people and all the groups from the ENSERio study.*

Source: *ENSERio study.*

For those who receive a treatment, it is mainly through the hospital, followed by the private specialist and, finally, by the family doctor and/or pediatrician.

Health center that gives the treatment
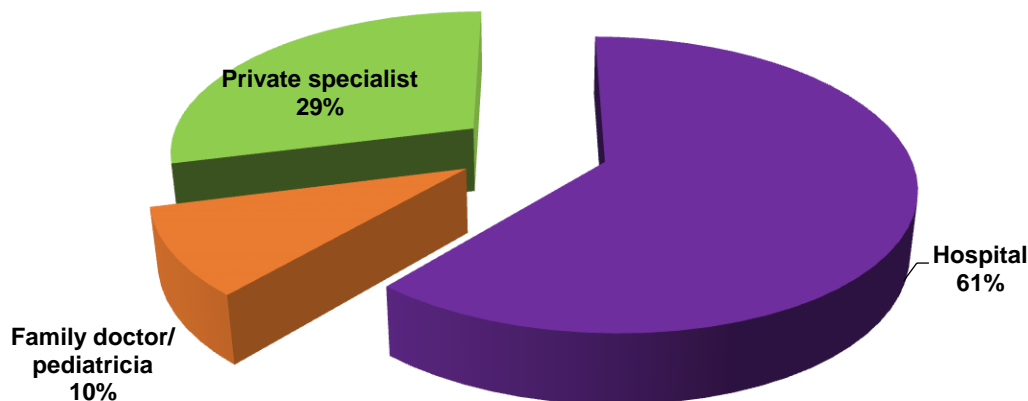
Graphic 4. *Percentage of the sample that receives treatment and the health center where they receive it.*

Source*: ENSERio study.*

In the ENSERio study, patients obtained the treatment first from the hospital (in 63 % of cases), followed by the family doctor and/or pediatrician (in 20 %) and finally from the private specialist (17 %).

In the case of people without a confirmed diagnosis, treatment is mainly obtained through private specialists, although the main facilitating center in both cases is the hospital.

Regarding to the type of treatment, the majority percentage, both in the ENSERio study and in the specific study for people without a diagnosis in the ENSERio study the percentage is higher (in 70 % of cases for people without a confirmed diagnosis and in 86 % of cases in the total group of pathologies in the ENSERio study).

Specialties attended at least once in the last two years

The specialties most consulted or used in the last two years by people without confirmed diagnosis are: neurology and ophthalmology, coinciding with the main consultations obtained in the ENSERio, which also obtained a big percentage for genetics (62 % for undiagnosed people compared to 22 % for all the pathologies).

There is also a higher percentage of people who declare to have used the services of traumatology and orthopedic surgery and rehabilitation. Regarding to the global data of the ENSERio, the percentages are similar, adding pediatrics (41 %).

All groups

Specialty	%
Neurology	47%
Ophthalmology	38%
Emergency services	34%
Rehabilitation	31%
Cardiology	29%
Orthopedic surgery and traumatology	25%
Endocrinology	23%
Pulmonology	22%
Genetics	22%
Dermatology	21%

Undiagnosed

Specialty	%
Neurology	68%
Genetics	62%
Ophthalmology	46%
Pediatrics	41%
Rehabilitation	37%
Orthopedic surgery and traumatology	32%
Emergency services	28%
Otolaryngology	22%

Charts 8 and 9. Specialties attended in the last two years. Charts for all groups of the ENSERio study and undiagnosed patients

Source: ENSERio study.

Tests and hospitalizations

Regarding to the number of tests performed in the last two years, undiagnosed persons coincide with the overall number of rare pathologies in which the majority is concentrated in those persons who have had between 1 and 10 tests.

Particularly for undiagnosed people, 49 %, i.e. approximately half of the sample analyzed, claimed to have had between 1 and 5 tests, 32 % between 6 and 10 tests and 5 % between 11 and 15 tests. Finally, only 14 % stated that no tests had been carried out.

Tests in the last two years

■ Number of tests

Category	Percentage
1 to 5	31%
6 to 10	49%
11 to 15	5%
None	14%
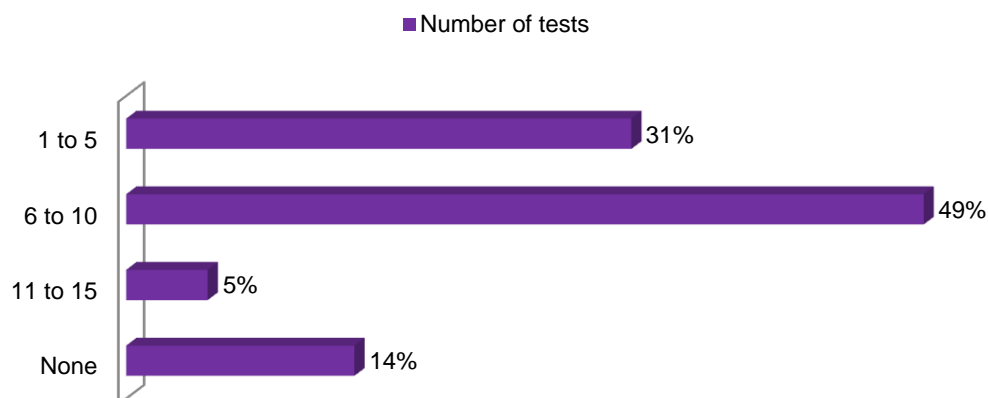
Chart 10. Tests taken by undiagnosed patients in the last two years.

Source: ENSERio study.

On their behalf, people without a confirmed diagnosis claim to have been hospitalized more times than the total number of pathologies in the ENSERio study. Thus, while in the ENSERio study there were 64 % of people who had not been hospitalized at all in the last two years, the specific data for undiagnosed patients reveal 60 %, a slightly lower percentage. Moreover, in the ENSERio study 24 % of people declared to have been hospitalized 1 or 2 times and the data for this specific report for the group of undiagnosed people are 30 % of people who have been hospitalized once or twice in the last two years.

Hospitalizations in the last two years

■ Undiagnosed   ■ All groups

Category	Undiagnosed	All groups
More than 10	1%	2%
6 or 10	0	2%
3 or 5	8%	8%
1 or 2	29%	24%
None	60%	64%

1.4. Commuting

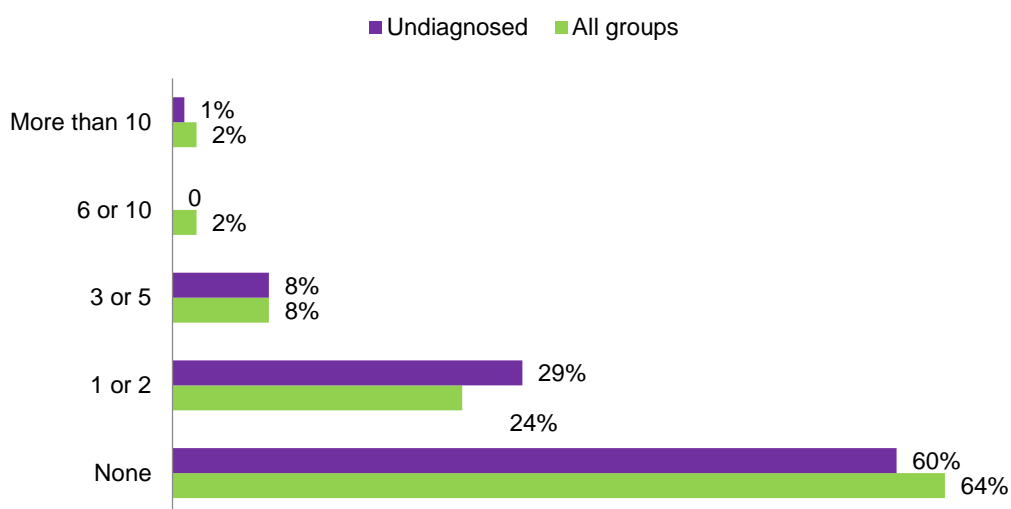59 % of the answers about commute claim to have had to travel to get a diagnosis outside their province in the last two years. Specifically, 40 % have moved 1 to 4 times and 19 % have had to move 5 or more times.

On the other hand, it is also important to point out that 8 % of the people surveyed stated that they had needed to commute but had not been able to. Finally, 21 %, approximately 1 out of 5 cases, did not need to travel to try to obtain a diagnosis.

In order to access treatment, the percentage of commuting is reduced to 22 % (13 % 5 or more times and 9 % between 1 and 4), although 13 % needed to travel and had not been able to. The amount of people who did not need to travel in the last two years to access treatment is 41 %.

Finally, in order to have access to medication, 10 % travelled in the last two years (7 % travelled 5 or more times and 3 % between 1 and 4), 7 % needed it although they were finally unable to do so and the percentage of people who did not need to travel for this access is the highest of all: 57 %.

Therefore, the highest percentage of commute takes place with the aim of obtaining a diagnosis for this group. More than half of the people who answered about these commuting had to move in the last two years for this reason.
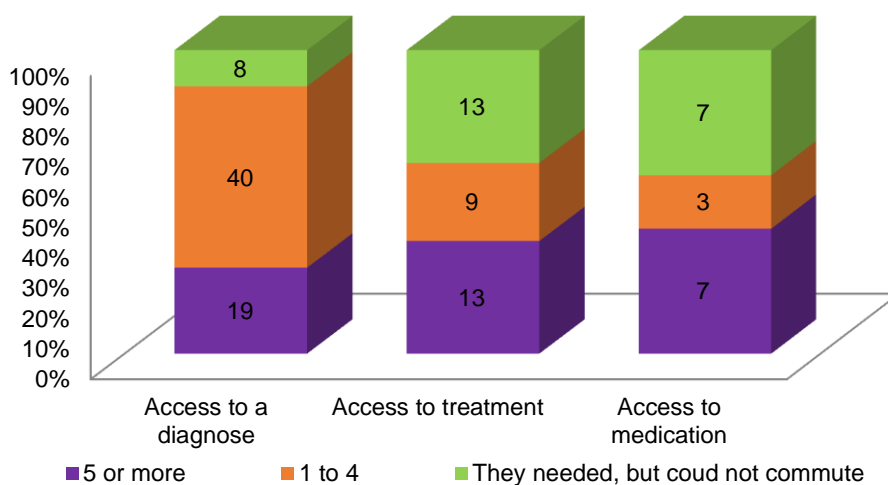
Chart 12. Number of times undiagnosed patients have had to commute to another Autonomous Region in order to get healthcare.

Source: ENSERio study.

In addition, these undiagnosed people recorded higher percentages of people who needed to move, for the three reasons: diagnosis, treatment and medication[5], although at the end they could not.
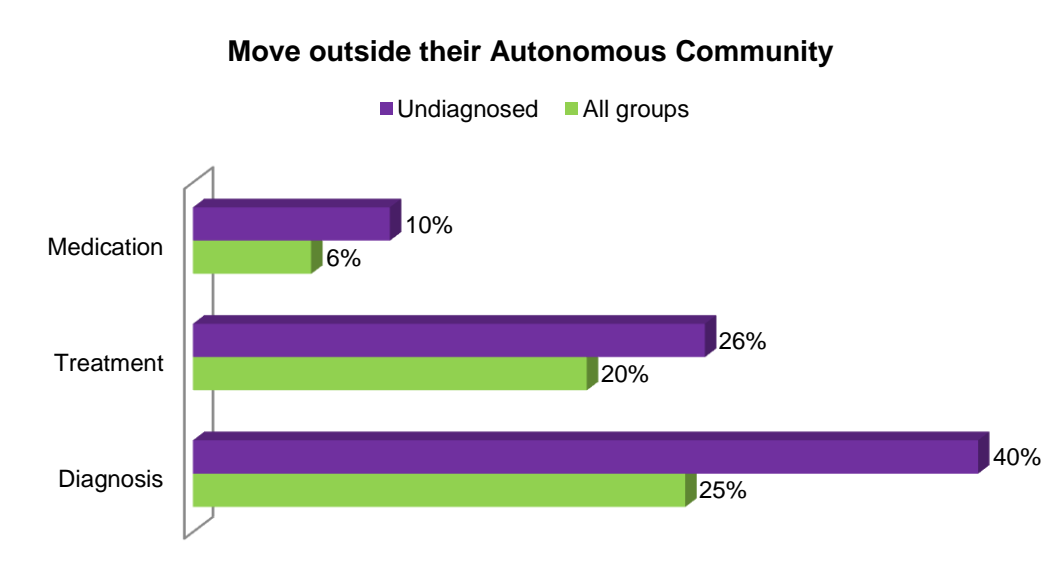
For commutes outside the Autonomous Community in order to access a diagnosis, the data show that it reaches 40 % of the total sample analyzed, a percentage notably higher than the percentages for all the groups of pathologies in the ENSERio study (25 %).

The main Autonomous Communities that act as a destination for obtaining the diagnosis are: Community of Madrid (44 % of cases) and Catalonia (34 %).

In order to access treatment, 26 % of the total sample commuted outside their Autonomous Community, and the ENSERio study showed a slightly lower percentage (20 %). The main destinations are Catalonia and the Community of Madrid, coinciding with the main destinations for obtaining a diagnosis (in 29 % of cases each) and Andalusia is added in 15 % of cases.

Finally, in order to have access to a specific medicine, they have moved outside their Autonomous Community in the last two years: 10% of the total sample analyzed, coinciding with movements outside the province. This percentage is also somewhat higher than that presented in the ENSERio study (6%).

That is, proportionally, people without a diagnosis move to a greater extent, especially in order to obtain a diagnosis, outside their Autonomous Community.

Move outside their Autonomous Community

■ Undiagnosed ■ All groups

	Undiagnosed	All groups
Medication	10%	6%
Treatment	26%	20%
Diagnosis	40%	25%

---

[5] For the previous ENSERio study no one answered "I couldn't even though I needed it" for the diagnose option. For the treatment option, the last study registered and 8% while the current study registers 13%. For medication it is 5% against 7%.
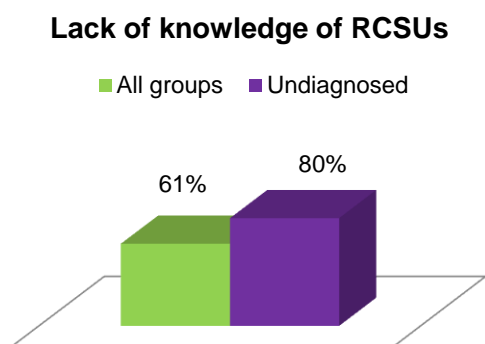
Chart 13. Patients who had to commute to another Autonomous Region. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

1.5. Reference Centers, Services and Units (RCSU) in the National Health System (NHS)

The degree of ignorance of the RCSUs in the case of undiagnosed participants of this study amounts to 80%, combining the percentages of "Isn't aware of," and "Doesn't know". In general, the percentages of ignorance are higher than those included in the ENSERio study, with 61% of people who did not know a RCSUs for their pathology.

Lack of knowledge of RCSUs

■ All groups ■ Undiagnosed

61%   80%

This data is closely linked to the lack of confirmation of a diagnosis for the pathology that these people have, which makes it difficult for them to identify those specific centers that attend to a technique, technology or procedure that can be used in the diagnosis or treatment of their disease.

Chart 15. Amount of people who don't know about a RCSU. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

On the other hand, of the 20% of people who know a Centre, Service and Reference Unit that serves them for certain procedures even if they do not know or have their pathology confirmed, 56%, that is to say, more than half, have a positive evaluation of these centers. However, this percentage is lower than that presented in the ENSERio study, with 86% satisfaction with the RCSUs.

It should be noted that there is also greater ignorance about how it works (13% say they do not know how it works so as to give an assessment compared to 5% of the ENSERio). In this aspect, the same thing happens as with treatment, in some cases not knowing the specific disease makes it difficult to assess whether the center or treatment works correctly.

Degree of satisfaction with RCSU

■ Undiagnosed ■ All groups

- **Unsatisfied**: 12% (Undiagnosed), 7% (All groups)
- **Satisfied**: 56% (Undiagnosed), 86% (All groups)
- **Don't know**: 13% (Undiagnosed), 5% (All groups)
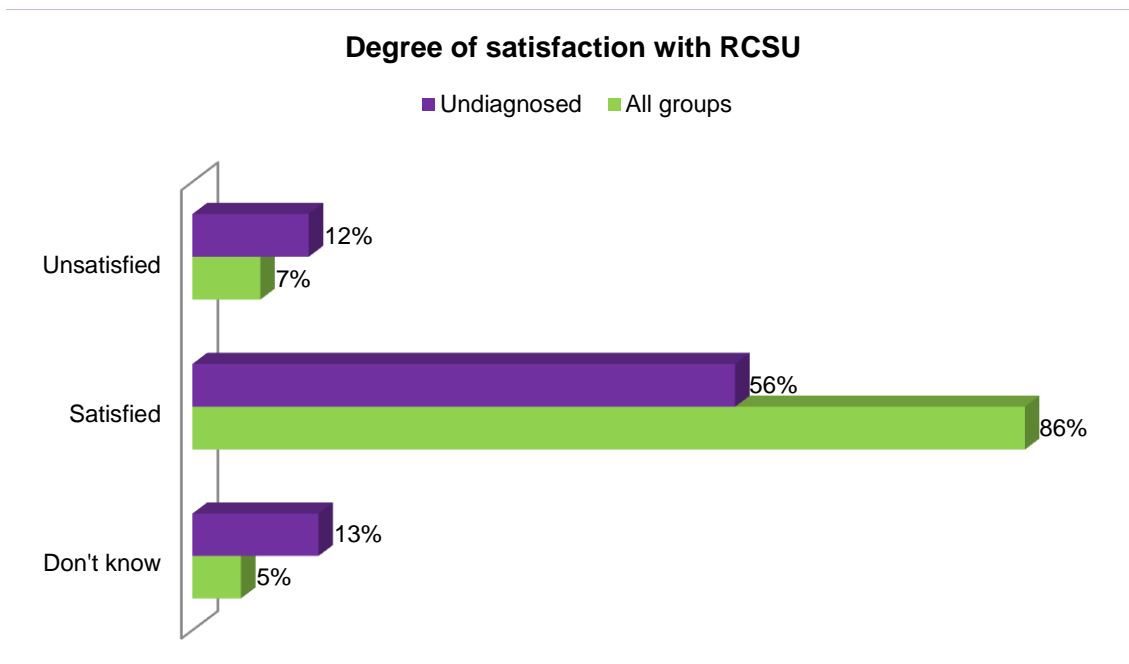
Chart 15. Percentage of satisfaction with the RCSU. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

1.6. Access to medicines and other medical devices

Medical devices

Study participants report that 39% of them do not have access to the health products they need, or that this access is difficult for them. Following the line of ignorance of their situation linked to the lack of diagnosis, 16% of cases did not know if they really had the health products necessary for their illness.

One out of three cases, 33%, could also access without difficulty, compared to 45% of people participating in the ENSERio study, who stated that they did not perceive access to these products as difficult.

Access to medical devices

■ Undiagnosed ■ All groups

Chart 16. Access level to medical devices. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

Access to medicines

The lack of knowledge about whether adequate treatment is being accessed, as on previous occasions analyzed in the report, is a remarkable result due to the percentage of people who do not know whether they have access to the treatment they need for their disease (more than a quarter of the sample analyzed, 26%, compared to 10% represented by this option in the ENSERio study).

Approximately another quarter of this population, 24%, claim to have difficulty accessing treatment or even not being able to access it.

On the other hand, although consistent with the foregoing, the percentage of people in the ENSERio study who claimed to have access without difficulty was 50%, half of the sample analyzed, whereas in the present report, the undiagnosed people studied perceive to a lesser extent that they can access the treatment they need without difficulty (35% of cases).

Access to medicine

■ Undiagnosed ■ All groups

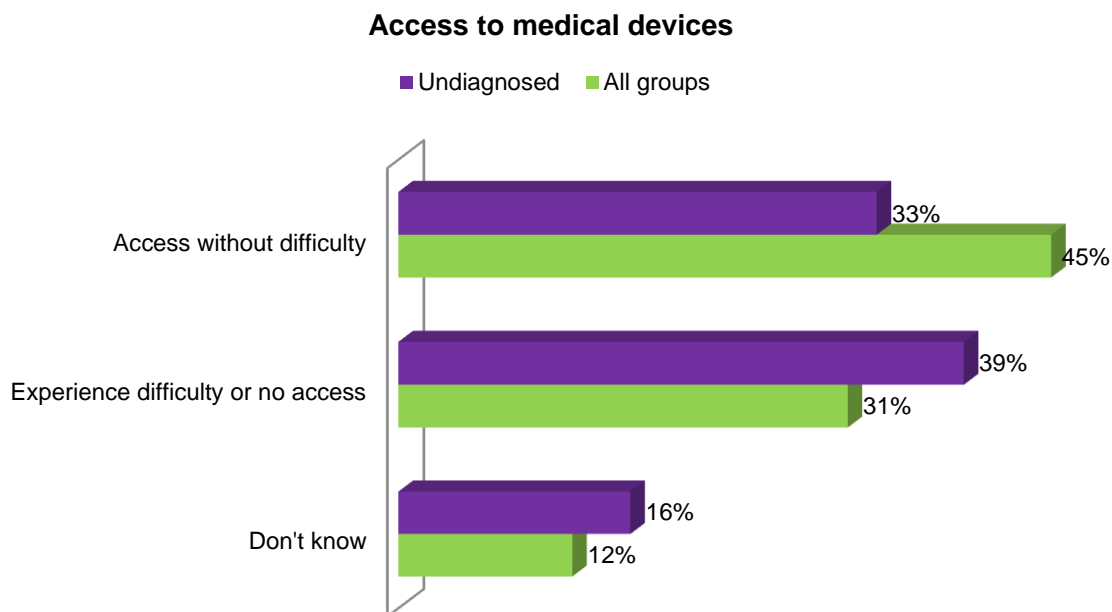|	Undiagnosed	All groups
Access without difficulty	35%	50%
Experience difficulty or no access	24%	31%
Don't know	26%	10%
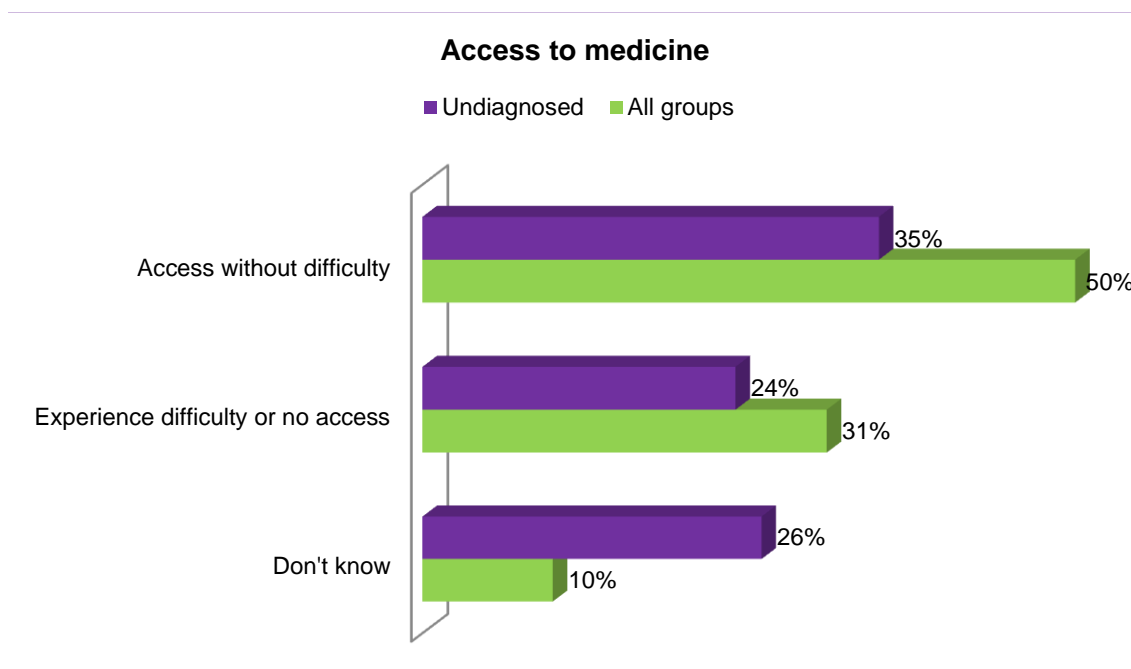
Chart 16. Level of Access to medication. *Comparison between all groups of ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

Finally, with regard to public coverage of health products and medicines, the most frequently reported options were those related to partial coverage, as was the case with the ENSERio study (44% for health products and 81% for undiagnosed persons studied).

Medications in Special Situations

Exclusively 11% of people have responded to any of the options for the use of medicines in special situations (foreigner, orphan, compassionate use). Of these, 4% indicate that they have ever used an orphan drug (a specific drug for rare diseases) for a disease that has a foreign drug (not authorized in Spain but authorized in other countries), another 4% have used an orphan drug (specific drug for rare diseases) and, to a lesser extent, 2% have used a drug for compassionate use (investigational drugs for patients outside a clinical trial and without therapeutic alternative). On the other hand, no one has indicated that they have used off-label medicines (use of medicines in conditions different from those authorized).

Medication in special situations

4% 4%

2%

Foreigner Orphan Compassionate use
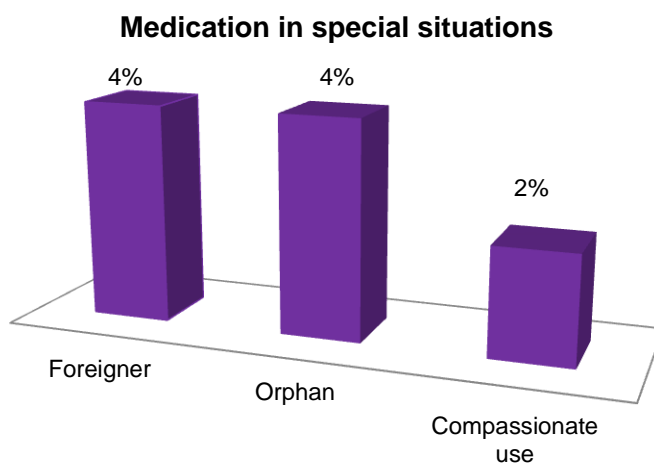
Chart 17. Use of medication for people in special situations.

Source: ENSERio study.

What needs due to the disease are covered by the public health system (in the last two years) and to what extent?

Need coverage by public administrations

■All groups ■Undiagnosed

Category	Undiagnosed	All groups
Apoyos (silla de ruedas, adaptación de la…	8%	21%
Commuting	0	13%
Respite care	0	3%
At-home care services	6%	31%
Treatment and medication	34%	53%
Tests	51%	66%
Hospital admission	42%	75%
Going to the hospital	49%	73%
Occupational therapy	6%	25%
Rehabilitation	12%	32%
Eye retraining	7%	13%
Psychomotor education	7%	17%
Psychology	25%	26%
Protheses	22%	27%
Speech therapy	12%	21%
Injections	33%	67%
Surgery	43%	68%
Glasses/contact lenses	0	3%
physical therapy	13%	16%
Dental care	10%	14%
At-home nursing care	3%	9%
Hearing aids	0	12%
Early care	31%	49%

Chart 18. Percentage of people who receives coverage by the public administration. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source*: ENSERio study.*

Some needs are covered to a greater or lesser extent by the public system, however, the people participating in the study state that they cover part of their needs through more sources, either

their own or through patient associations. Those needs that are not perceived to be covered by the public health system are: hearing aids, glasses, contact lenses and, in general, visual aids, respite care and transportation.

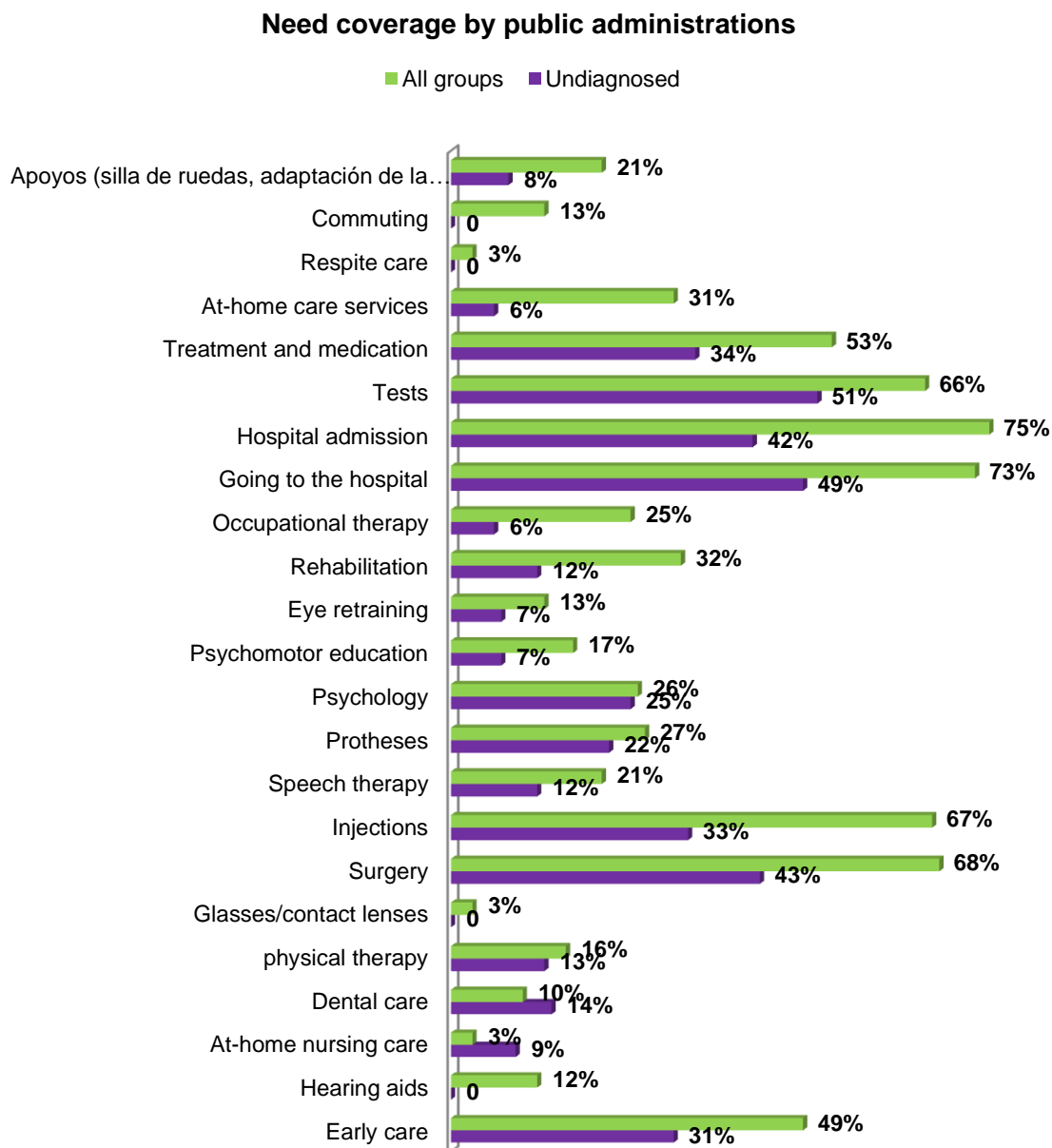On the other hand, some needs that are perceived as covered in less than 10% of the sample studied are: visual/Orto-optical re-education, occupational therapy, supports such as wheelchairs and/or housing adaptation, psychomotricity and home care services and home nursing care.

Those needs, on the other hand, most perceived as being covered by the public health system, are those that are most basic or urgent: whether they are surgical interventions, appointments with health professionals, hospital admissions and tests and evaluations. The above needs are perceived as covered in less than half of the cases, despite representing the needs most frequently valued as covered, with the exception of tests and evaluations, which reaches half of the people who answered this question (51% of the cases).

Finally, in approximately one out of every three cases, the following needs are considered covered by the public health system: treatment and medication, injections/perfusions and early care. In one out of four cases, psychological needs and internal and external prostheses are covered by the public system. And in less than one in five cases, rehabilitation, physiotherapy and dental care are publicly covered.

Percentageately, the figures for all the groups of pathologies included in the ENSERio study show greater coverage of all the needs included, with the exception of dental care, with a very similar percentage of coverage (10% in the case of ENSERio and 14% in the case of the specific report for undiagnosed persons).

1.8. Assessment of disability and dependency.
1.8.1.   Assessment of disability

88% of people without a confirmed diagnosis claim to have some form of disability.

Considering that these people can live with various types of disability, 69% of the sample claim to have a motor disability, half have intellectual disability, 15% have vision disability, 8% have hearing impairment and, finally, 9% of the people studied have psychiatric disability.

In addition, undiagnosed people have higher percentages of disability, especially in relation to intellectual disability, in which the percentage difference is greater.
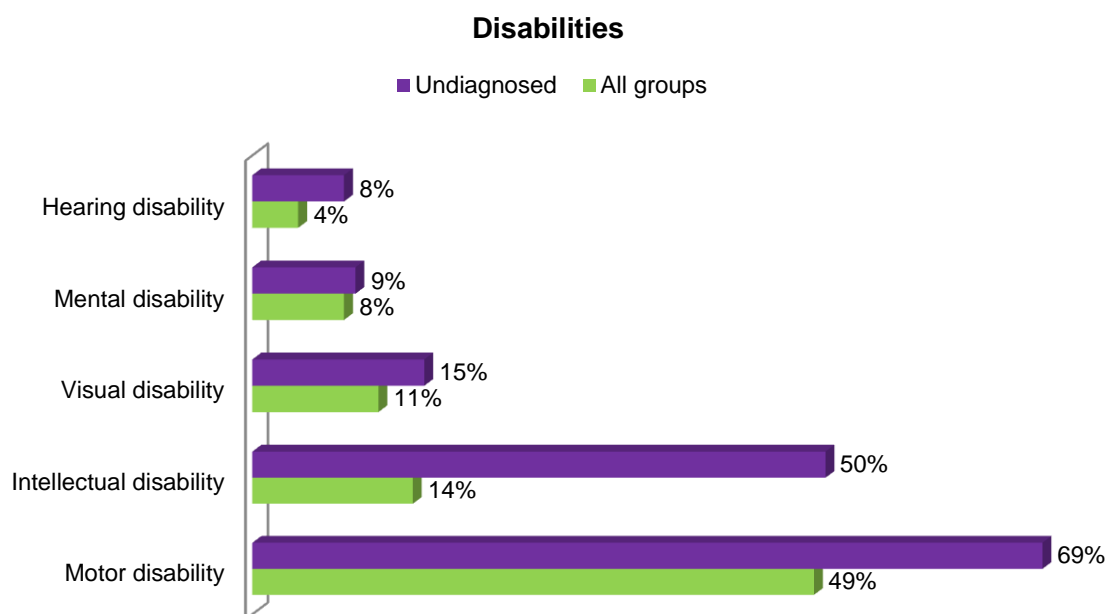
Disabilities

Chart 19. People who has a disability, one person can mark more than one answer. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

Of the people with disabilities participating in the Study, 84% have recognized the disability through the official certificate. Of these persons with official disability certificate, 39% had a disability from 33% to 64%.

The percentages collected in the ENSERio study point to lower degrees of disability, 51% as opposed to the 39% indicated in the specific report of persons without a diagnosis who had a recognized disability of 33% to 64%.

Along the same lines, the report for persons without a diagnosis includes higher percentages of disability. In this way, the group with a recognized disability of more than 75% is, in the report for undiagnosed persons, 34% compared to 24% in the ENSERio study for all groups of pathologies, ten percentage points higher.

Disability degree

■ 33-34% ■ 65-74% ■ More than 75%

	33-34%	65-74%	More than 75%
Undiagnosed	39%	22%	34%
All groups	51%	23%	24%

Chart 20. Disability degree. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

In general terms, there is concurrence with the degree of disability received. Non-diagnosed persons stated that they were satisfied in 61% of cases, the category most frequently indicated being a higher percentage than that included in the ENSERio study. Nevertheless, it is important to point out that 22% of the sample, more than one in five people, disagree with their degree of disability.

Concurrence with the degree of disability

■ Undiagnosed ■ All groups

	Undiagnosed	All groups
Yes	61%	51%
No	22%	36%
Don't know	14%	11%
No answer	3%	2%
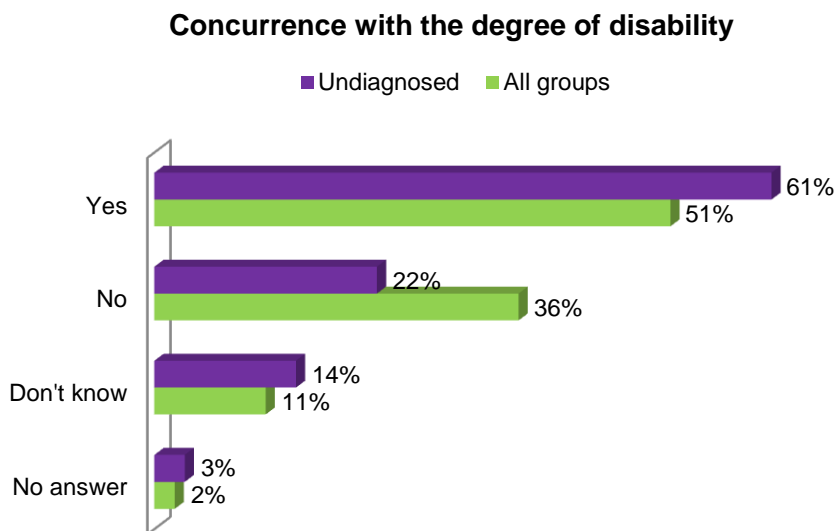
Chart 21. Concurrence with the disability degree. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Soruce: ENSERio study.

On the other hand, the application process for disability assessment lasted less than six months in 48%, that is, almost half of the cases studied.

In one out of three cases, 34%, it took between six months and a year. And it lasted for more than a year in 14%.

Thus, the percentages for delay of more than six months for undiagnosed persons (approximately half of the cases studied) are lower than the percentages of the ENSERio study for total pathologies: 60%.

Time between the application for the disability degree and the actual assessment

■ All groups ■ Undiagnosed
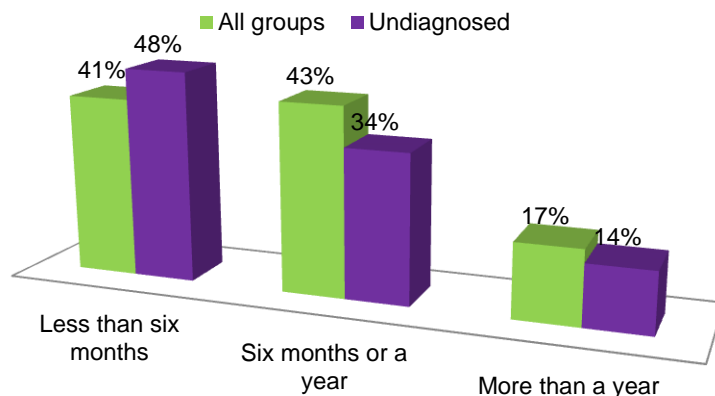
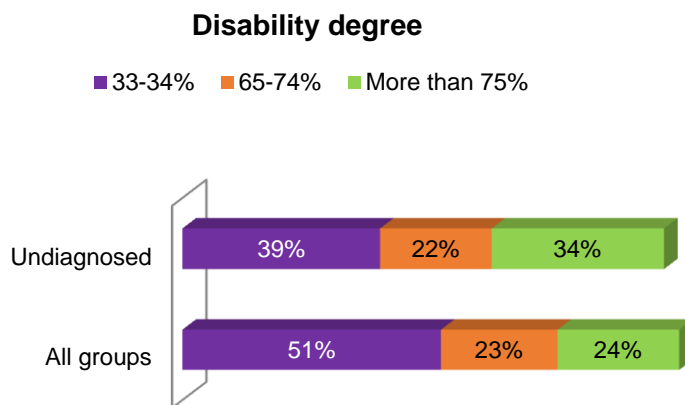**Chart 22**. Time patients had to wait since the first time they applied for the disability degree until actually getting the disability assessment. *Comparison between all groups of the NESERio study and undiagnosed patients.*

Source: ENSERio study.

On the other hand, from the time this assessment was received to the time the aid was received, 40% of the people studied spent less than six months. The delay of between six months and one year to receive aid since the assessment was experienced by 44%, the most frequent option, and the delay of more than one year by 13% of the population analyzed, data very similar to those of the ENSERio.

Waiting time between the assessment and the assistance

■ All groups ■ Undiagnosed
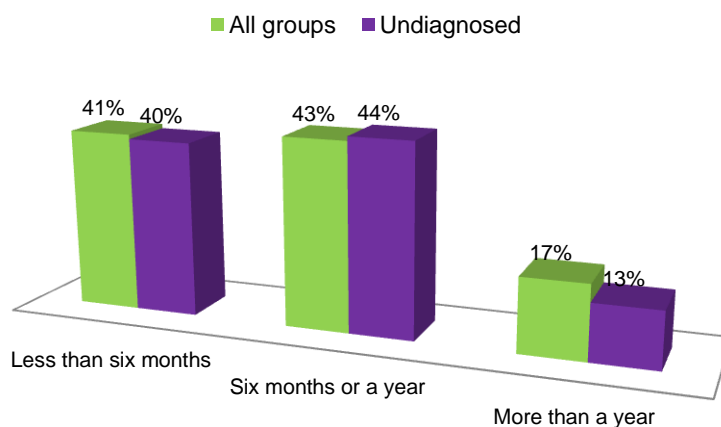
Chart 23. Time patients had to wait since they got their disability assessment until receiving assistance. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

1.8.2. *Assessment of dependency*

The most frequent thing among the undiagnosed persons studied is that they have applied for dependency and have obtained it (43% of the cases) and 14% are in the process of processing after applying for it.

However, those who have not applied for dependency intend to do so at some time in 29% of the cases studied, the second most frequent option, and only 1% do not intend to do so.

Among those who have been assessed, 7% of them obtained grade I. The evaluations among the persons participating in the Study were mainly grade III, in more than half of the cases (53%) and grade II in a third of the cases (33%). Therefore, among the undiagnosed persons studied, the assessments were in more than half of the cases assessed, for the highest degree of dependence.

In the ENSERio study, on the other hand, the grade III dependency assessments were percentage lower, as was the higher percentage of grade I assessments.

Dependency level

■ All groups ■ Undiagnosed

Chart 24. Dependency level. *Comparisong between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

As was the case in the ENSERio study, positive results were observed for compliance with the degree of dependency received, in 62% of cases in the ENSERio and 64% among undiagnosed persons, specifically.

Among the 20% of people dissatisfied with their degree of dependence, the majority consider that they have a lower percentage than the adequate one, and in one of the cases it points specifically to the lack of diagnosis.
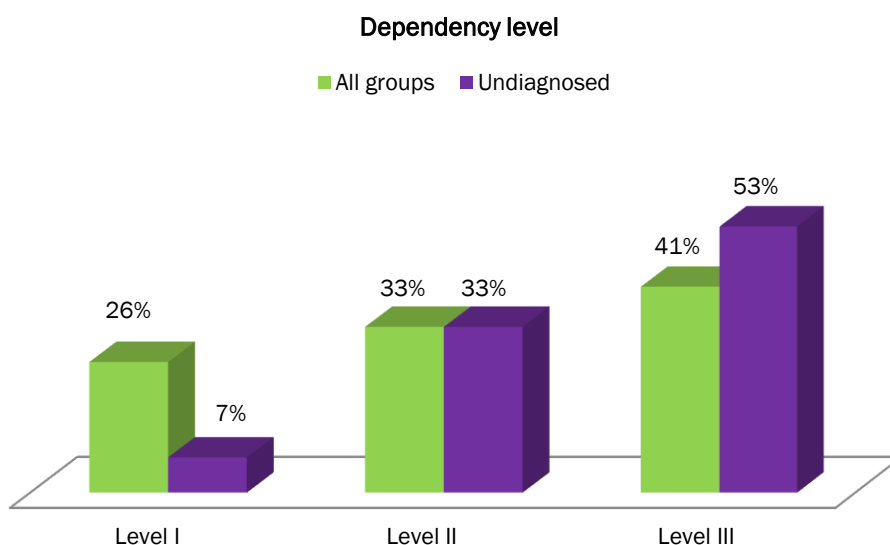
In relation to the waiting time between the application and the assessment and between the assessment and the receipt of the aid, a greater percentage of people with respect to disability who took more than a year to obtain the assessment (36%) is observed, a percentage that is equivalent to less than six months.

Likewise, and along the same line, 51% of the sample stated that it had taken more than a year to receive the aid since the assessment of dependency. Therefore, the process of obtaining dependency, and the aid linked to it, is longer in time.

More than a year of delay

■ All groups   ■ Undiagnosed
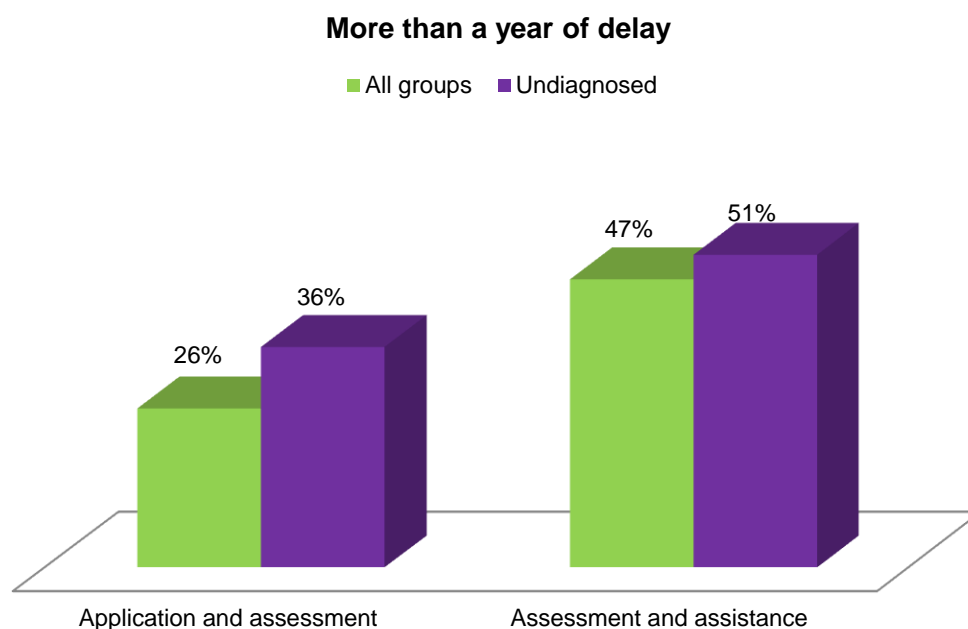
Chart 25. People who waited more tan a year for the disability procedures. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

Related to this assessment and obtaining aid is the System for Autonomy and Care for Dependency (SAAD) which, as defined in the ENSERio study, is the set of services and economic benefits aimed at promoting personal autonomy, care and protection for dependent persons, through public and private services duly accredited and concerted and contributes to improving the living conditions of citizens.

As was the case in the ENSERio study, all services show very low figures for the percentages of coverage of the related needs in each of them, with the exception of adapted transport and schools, where the percentages of coverage of these services are similar to those of people who claim not to have coverage or to have it partially, but it is not sufficient. Therefore, among this

specific group there is also a majority perception of a lack of coverage of their needs in relation to these services.

Access to the service and coverage, in addition, of needs only reaches a quarter of the population analyzed in these two items (school and adapted transport) for the undiagnosed. In the case of the ENSERio study, this only occurs in the case of the adapted school and in remote assistance.

Access to SAAD services and needs coverage

■ Undiagnosed ■ All groups

Service	Undiagnosed	All groups
Refund (treatment, medical consultation, transport…)	18%	18%
Social benefits	19%	15%
Tax reduction	22%	20%
Adapted transport	37%	28%
School adaptation	30%	30%
Household adaptation	7%	8%
Public holiday care	8%	8%
Respite care	8%	7%
Long-term care	7%	8%
Medical consultation	21%	39%
Rehabilitation service	20%	11%
Psychological assistance	11%	16%
Economic benefit for personal assistance	17%	7%
Economic benefit for care in the family environment and support for non-professional carers	22%	14%
Economic benefit linked to the service	22%	11%
Residential service	8%	12%
Day or night center service	14%	19%
At-home care service	19%	12%
Telecare service	7%	34%
Service for the promotion of the personal autonomy	11%	14%
Prevention service of dependendy situations	10%	10%

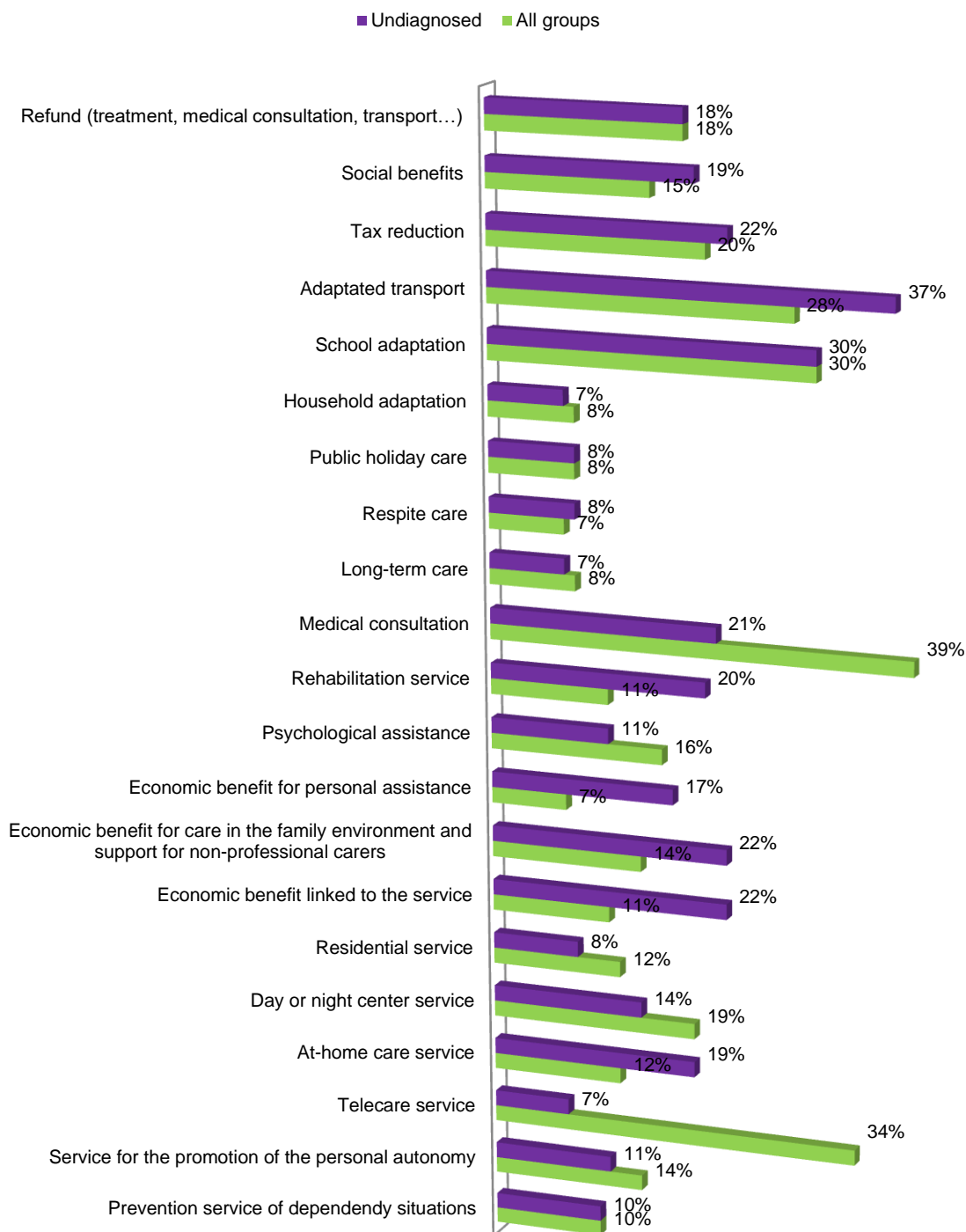**Chart 26**. People with a positive perception of the need coverage and the access to SAAD services. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

1.9.Personal assistance and support products

73% of the undiagnosed people participating in the study claim to have impairments due to their illness to develop basic activities of daily living.

It is worth mentioning that the perception of almost half of the participants (45%) is that they have much limitation to develop these activities.

Impairments on the development of daily activities

■ Undiagnosed ■ All groups

Activity	Undiagnosed	All groups
Hearing	8%	4%
Behavioral aspects (self-harm…)	19%	13%
Other	8%	14%
Eyesight	17%	15%
Administrating the money and economy	40%	18%
Communication	62%	20%
Learning and applying knowledge	67%	26%
Interpersonal relationship	47%	26%
Selfcare	63%	36%
Educational or laboral activities	62%	36%
Movements (change positions, move the limbs…)	56%	41%
Free time	65%	43%
Transport and commuting	54%	43%
Feelings	40%	46%
Domestic life	58%	53%

Chart 27. People with impairments in their daily life activities. *Comparison between all groups fo the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

As can be seen in the graph above and analyzing the percentages for each of the samples studied, there are greater limitations in the development of basic activities among people without diagnosis, in all of the activities mentioned.

They present difficulties especially in the learning and application of knowledge, leisure and free time, educational or work activity, self-care, communication with other people.

41

1.9.1. Personal care and assistance

As reflected in the ENSERio study, the data for people without diagnosis also confirm that in most cases it is the family that assumes the support for the different tasks of care and assistance, with 80% of the people analyzed indicating that this type of assistance was always assumed (49%) or many times (31%) by relatives. In the same line, the persons who indicated that the families attended them few times or none constituted 18% of the sample.

The highest percentage was reflected in the family, for personal assistance, as opposed to other profiles such as specialized professionals, non-specialized professionals such as household employees, or other persons.

Specifically, the profile that most assumed the care of people with undiagnosed HBS was the mother, being marked as first choice in 70% of the cases studied. This data reflects the same situation as the ENSERio study, which observed a general feminization of care.

Unlike the ENSERio study, however, the profile most marked as the second option is the father (in the ENSERio study it was the couple).

Finally, and related to the percentages of disability and dependency (higher in the persons studied without diagnosis), higher percentages are also reflected with respect to the ENSERio study in the periods of more hours of care per day of the person with HBS.

Thus, higher percentages are observed in persons who claim to dedicate more than 6 hours to the care of the person with HBS (57% compared to 34% in the ENSERio study). However, in both cases, the use of more than six hours for personal assistance was the most frequent option.

What are the implications for the caregiver of this circumstance?

Likewise, the main consequence pointed out among the undiagnosed people, for the people who take charge of this support is to have less free time/leisure, followed by the reduction of the working day, and the resentment of their relationship as a couple.

The consequences indicated above are the three main repercussions, also pointed out in the ENSERio study.
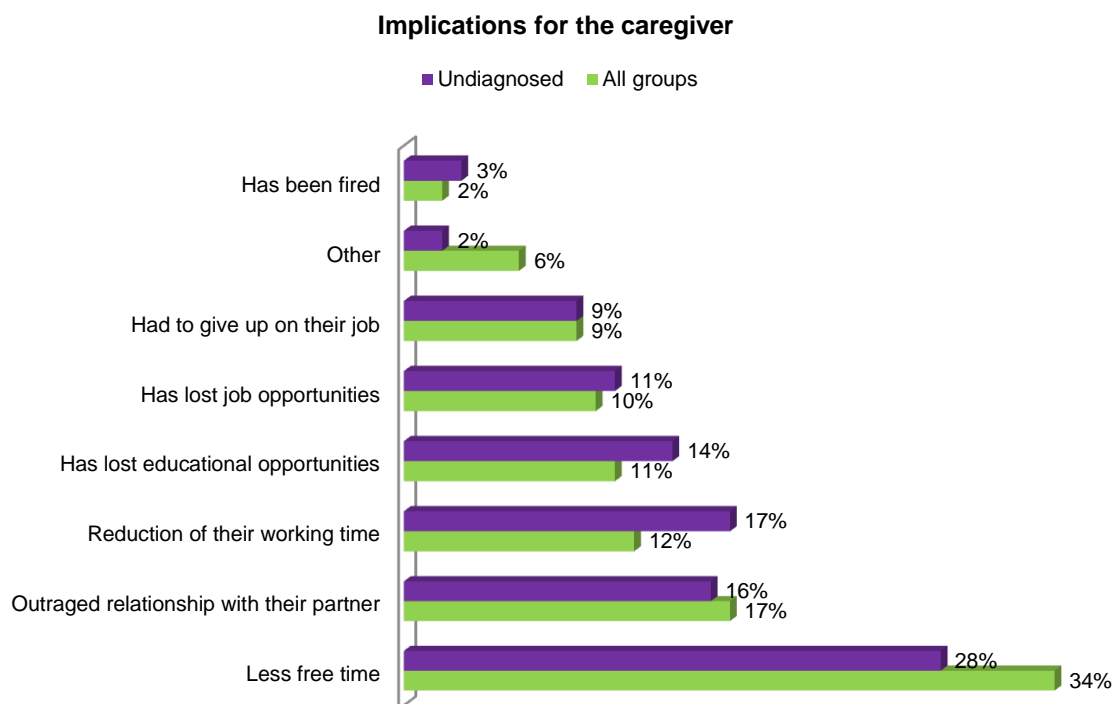
Implications for the caregiver

■ Undiagnosed   ■ All groups

Has been fired	3% / 2%
Other	2% / 6%
Had to give up on their job	9% / 9%
Has lost job opportunities	11% / 10%
Has lost educational opportunities	14% / 11%
Reduction of their working time	17% / 12%
Outraged relationship with their partner	16% / 17%
Less free time	28% / 34%

Chart 28. Answers for each of the implications given. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

1.9.2.   The use of supportive products

The people studied claimed to use supportive products in 25% of the cases, one in four. At the same time, approximately another quarter of the study population, 24% need, but do not have, assistive devices. The latter figure is 10% higher than in the ENSERio study.

1.10.   Benefits and other resources on grounds of disability

A majority of the persons who answered this question received benefit for a dependent child with a disability, approximately half of the sample studied, 46%. The next highest benefit received, well behind the family allowance for a disabled dependent child, was the non-contributory invalidity pension (in 8% of cases). Finally, the persons who did not receive any benefit were 34%. The main difference with the rest of the groups of pathologies is related to this last data, the persons in the ENSERio study who did not receive benefit were more than half (55%), as opposed to 34% of the undiagnosed persons.

2. INCLUSION AND SOCIAL INVOLVEMENT

In addition to social and health care, the ENSERio study analyses the extent to which people with EPF could exercise their citizenship under full conditions, i.e. "if they have real access to rights recognized by law".

Therefore, this report will specifically study for the undiagnosed people analyzed: the adaptation of housing, the situation regarding education and training, access to employment, the impact of the disease on the family budget, the subjective perception of their situation in different spheres and the discrimination perceived by these people.

2.1. Adaptation of housing

The most frequent option of undiagnosed people participating in the Study about housing adaptation is that which indicates their difficulty in assuming the expenses related to housing adaptation (21%). In the ENSERio study, the percentage of people who had difficulties in assuming expenses related to access to housing was higher (27%).

It is worth mentioning that for the SAAD aids regarding the adaptation of the home, however, only 7% indicated receiving them.

Expenses for the house adaptation

Category	Percentage
No answer	18%
Don't know	18%
No, it is covered by the health care	6%
Yes, but I can't pay for it	12%
Yes and it is difficoult to undertake	21%
Yes, but I can't undertake the expenses	15%

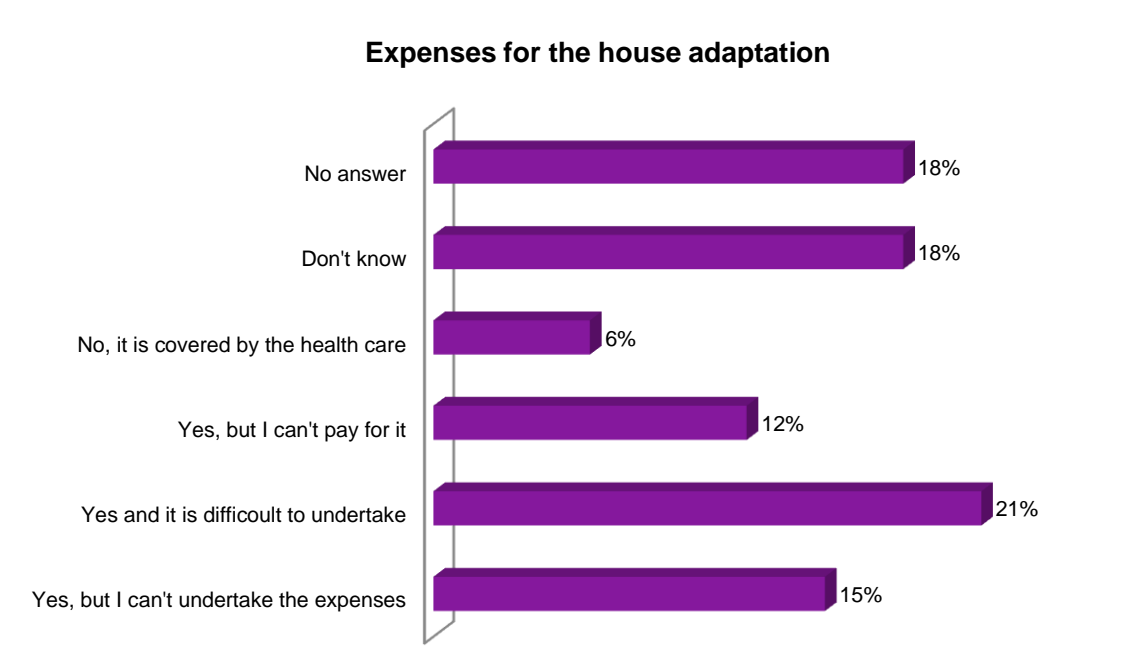**Chart 29.** Undiagnosed patients' answers regarding to the difficulties of undertaking the expenses related to house adaptation.

Source: ENSERio study.

2.2. Education and training

For the over 18 year old participants, only 13% stated that they had reached university studies, a reality that contrasts with the 56% of people who stated in the ENSERio study that they had reached this level.

44

Among the people who continue their training, 36% are in an ordinary center with support, the same figure that claims to be in a special education center, the most frequently indicated centers.
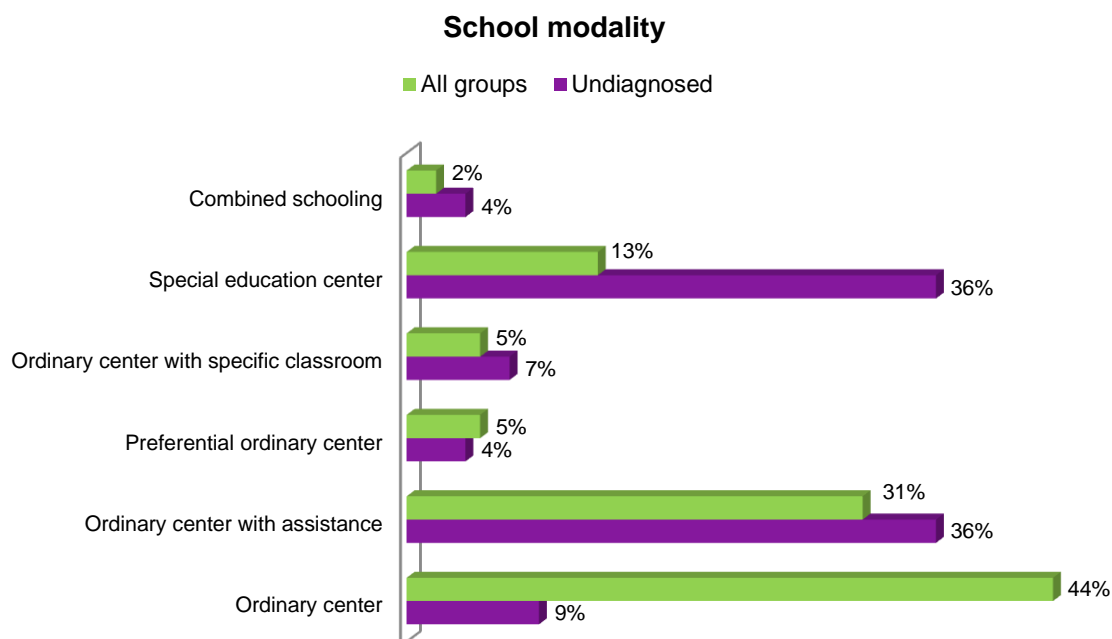
School modality

■ All groups ■ Undiagnosed

Chart 30. School modality of the sample who is still studying. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

2.3. Access to employment

Of those undiagnosed people who indicate that they are not working, 21% state that it is due to their rare disease, as they cannot work permanently as a result of it (compared to 31% indicated in the ENSERio Study for all pathologies). The other reason indicated, and according to the fact that the sample is in a high percentage of minors, is that they continue their training, they are studying (76%).

On the other hand, people of working age who indicated that they had an official certificate recognizing their incapacity for work (15%), indicated that they had a partial type of incapacity for work in 17% of the cases, the same as those who declared that they had a type of total incapacity for work. It is more frequent, among people with recognized incapacity for work, to have absolute incapacity for work or great incapacity, both a third of the sample (33%).

Among those who indicate that they are working (30% of people of working age, a percentage similar to the ENSERio, 33%), almost half (47%) are full-time, a higher percentage than in the ENSERio study (30%). 18% of them have a part-time working day (compared to 9% in the ENSERio study for all groups of pathologies).

For their part, 12% are housewives and there is the same percentage of self-employed. In this last percentage, moreover, there is also a relevant difference with respect to the total number of pathologies, with a lower number of self-employed in the ENSERio study (6%, half).

Therefore, the group without diagnosis studied presents higher percentages of partiality in employment and more frequency of people who dedicate themselves to employment as self-employed persons. However, they also present a higher percentage of full-time workers, especially related to the fact that in the ENSERio study there was a high percentage of people (44%), almost half, who indicated the option "Others" to describe their employment situation.

Employment situation
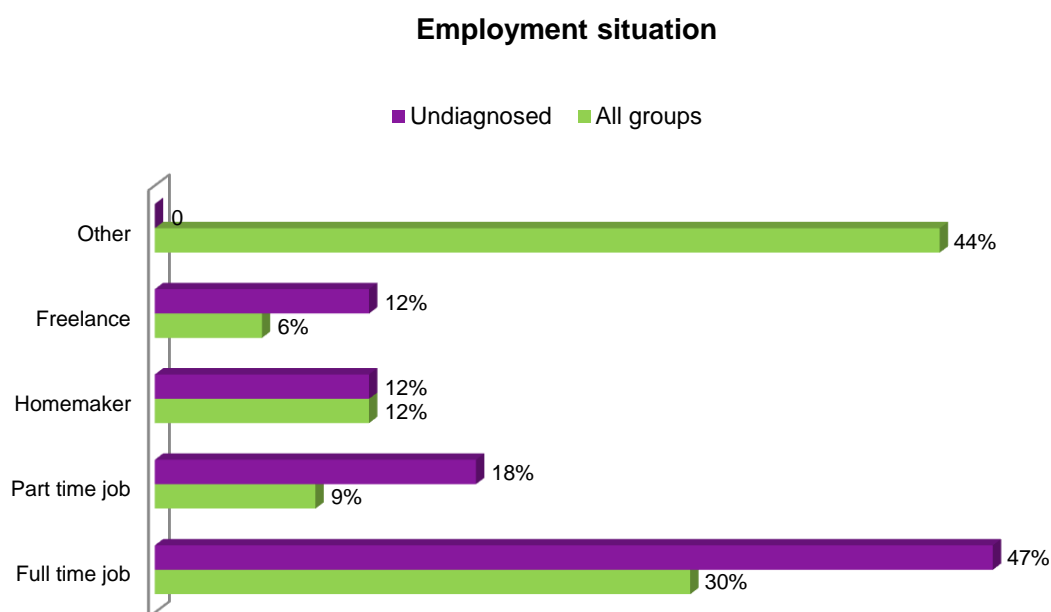
Chart 31. Employment situtation of those who had a job. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

Moreover, they also state to a greater extent that they need personal support and/or product support. The undiagnosed group studied has higher percentages of people who indicate that they need personal assistance, technical resources (technical support and accessibility) and specialized professionals.

In addition, among the people who claim to need it, there are more those who indicate that it is not provided by the workplace, as was the case in the ENSERio study for all groups of pathologies, although the group without diagnosis also has higher percentages in this sense.

Undiagnosed

- ■ Not needed
- ■ Needed, but not given by the work center
- ■ Needed and given by the work center

Technical resources 59% / 32%

Professionals 68% / 24%

Personal assistance 65% / 22% / 5%

All groups

- ■ Not needed
- ■ Needed, but not given by the work center
- ■ Needed and given by the work center

Technical resources 68% / 22% / 10%

Professionals 79% / 15% / 5%

Personal assistance 79% / 16% / 5%
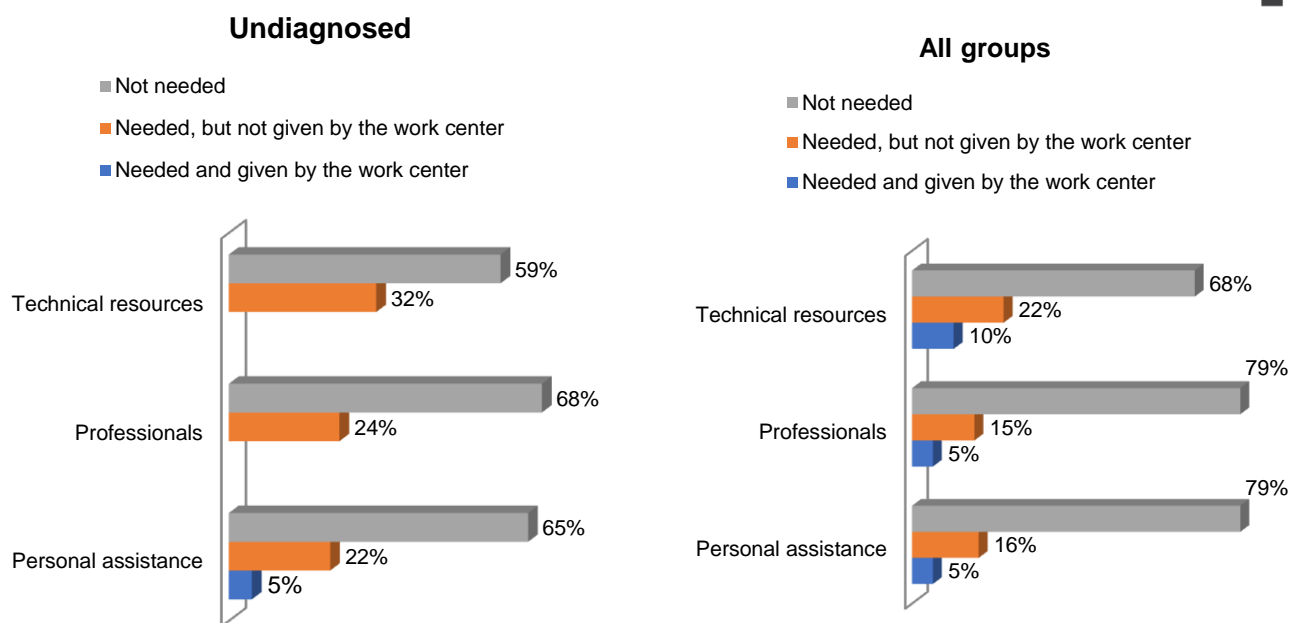
Charts 32 and 33. Needs of assistance at the work place. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

2.4.Impact of the disease on the family budget

What impact does the disease have for the undiagnosed group studied on the budget of the family unit?

The highest percentage, 26% of the participants, state that between 20% and 30% of annual family income is spent on the costs of the illness. Only 2.5% indicate that the disease has no costs, the least indicated response option by the people studied.

These data contrast with those collected for the total number of pathologies in the ENSERio study, the majority of which (25%, a quarter of the sample) claimed to allocate less than 10% of annual income, and 7% claimed not to allocate income as the disease did not involve any expenditure.
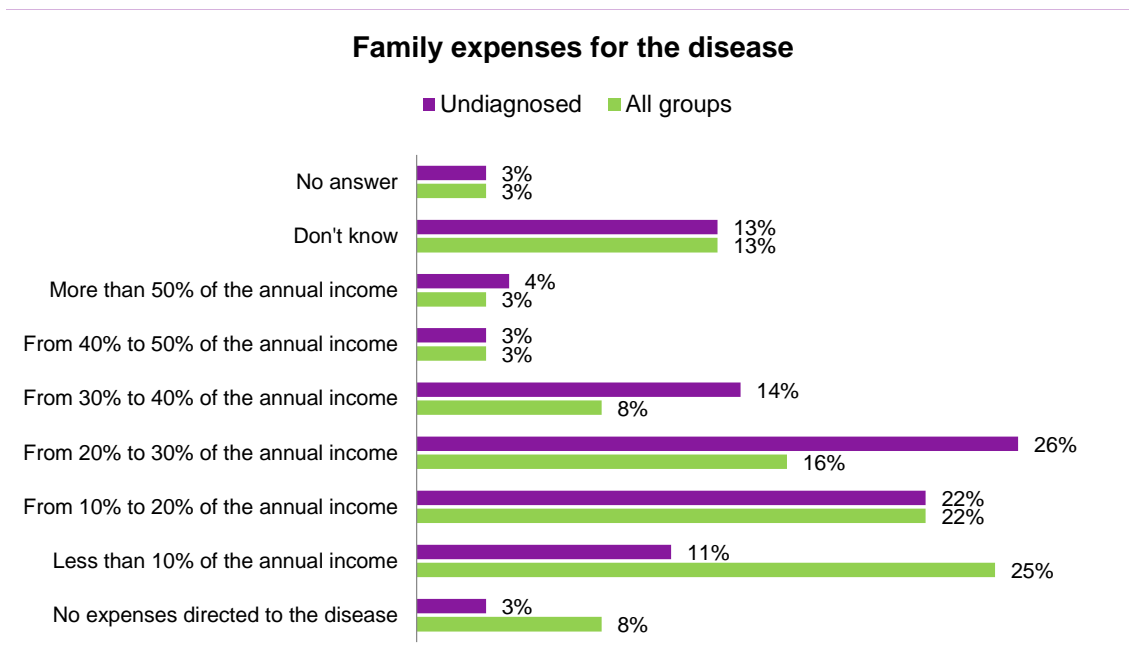
Family expenses for the disease

■ Undiagnosed ■ All groups

Category	Undiagnosed	All groups
No answer	3%	3%
Don't know	13%	13%
More than 50% of the annual income	4%	3%
From 40% to 50% of the annual income	3%	3%
From 30% to 40% of the annual income	14%	8%
From 20% to 30% of the annual income	26%	16%
From 10% to 20% of the annual income	22%	22%
Less than 10% of the annual income	11%	25%
No expenses directed to the disease	3%	8%

Chart 34. Family expenses for the disease. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

In the same vein, 82% of undiagnosed people studied value high or very high the cost of their rare disease, compared to 71% of the ENSERio study for total pathologies.
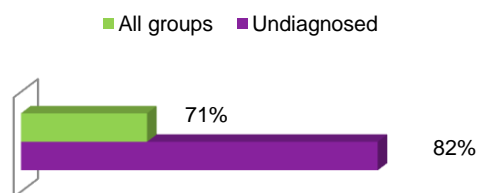
■ All groups ■ Undiagnosed

All groups: 71%
Undiagnosed: 82%

Chart 35. Percentage of people who assessed the expenses as high or really high. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: ENSERio study.

Related to this circumstance, 69% of people consider it quite or very difficult to assume, eight percentage points above those who identified the same difficulty in the ENSERio study.

By budgetary items, the people studied identified more difficulties in all of those indicated, except for medicines and other health products, in which the difficulty in assuming the expense is similar to the overall results of the ENSERio study.

They identified difficulties especially in physiotherapeutic, psychological, educational support, personal assistance and technical aids, in order of greatest to least difficulty identified.

Difficulty in assuming the expenses by items

■ All groups ■ Undiagnosed

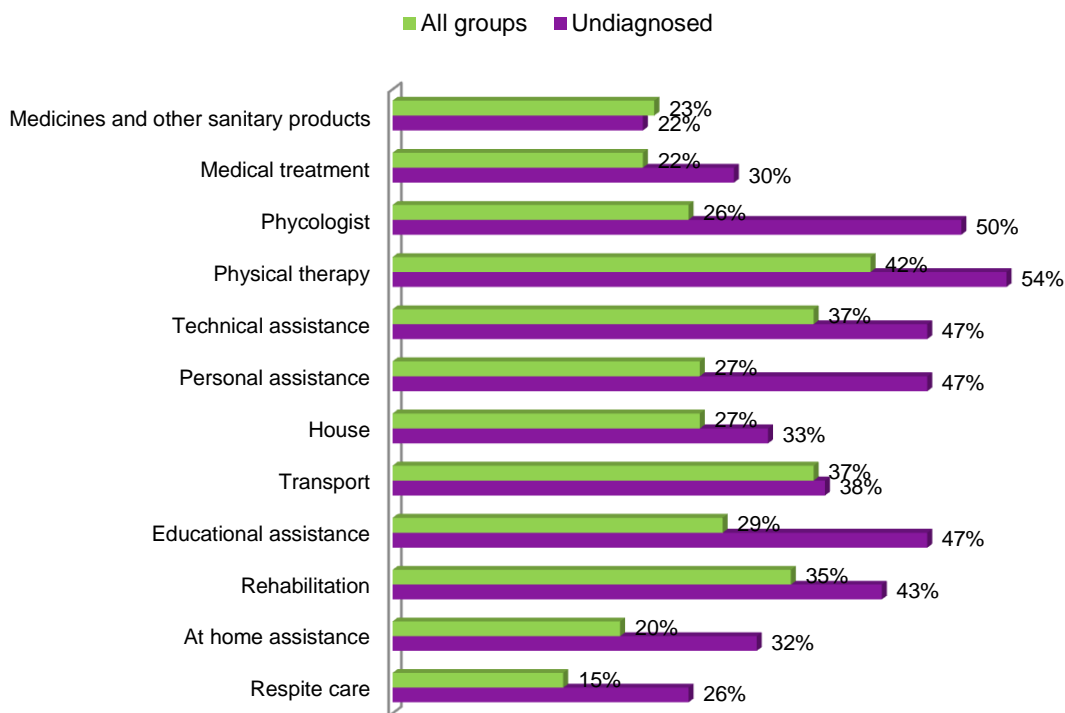| Item	All groups	Undiagnosed
Medicines and other sanitary products	23%	22%
Medical treatment	22%	30%
Phycologist	26%	50%
Physical therapy	42%	54%
Technical assistance	37%	47%
Personal assistance	27%	47%
House	27%	33%
Transport	37%	38%
Educational assistance	29%	47%
Rehabilitation	35%	43%
At home assistance	20%	32%
Respite care	15%	26%

Chart 36. Percentage of people who has difficulties to afford the expenses. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

2.5. Subjective perception of your situation

In general, people without a confirmed diagnosis analyzed, and with respect to the sample of the ENSERio study for the global pathologies, present more negative opinions regarding their situation in different areas.

Negative assessment of their situation

■ Undiagnosed ■ All groups

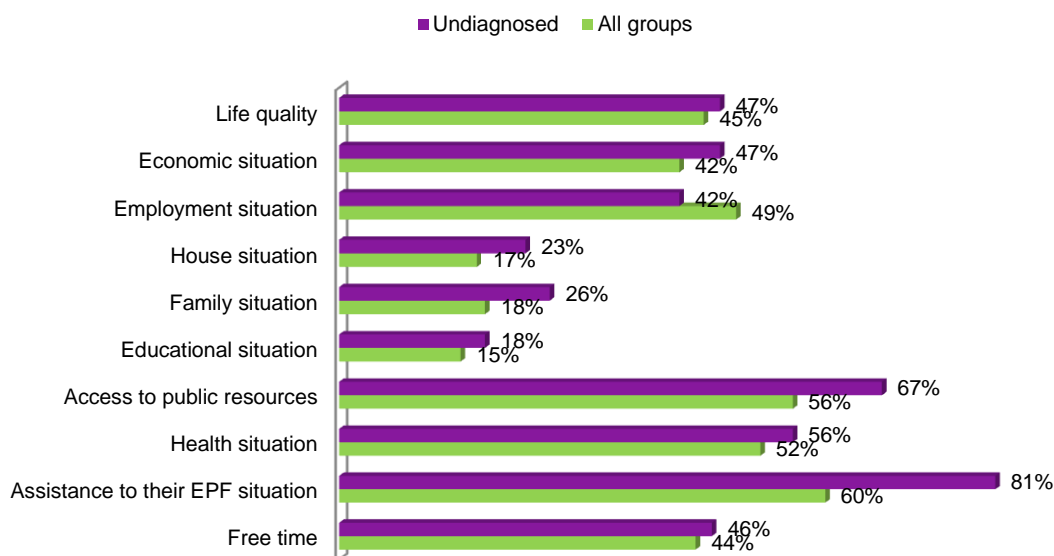|	Undiagnosed	All groups
Life quality	47%	45%
Economic situation	47%	42%
Employment situation	42%	49%
House situation	23%	17%
Family situation	26%	18%
Educational situation	18%	15%
Access to public resources	67%	56%
Health situation	56%	52%
Assistance to their EPF situation	81%	60%
Free time	46%	44%

Tabla 37. Percentage of people who assess their situation as negative. *Comparison between all groups of the ENSERio study and undiagnosed patients.*

Source: *ENSERio study.*

There is a similarity in the percentage of people who value their situation negatively between the two studies, with the exception of the situation in the care of illness, where there is a percentage difference of 21 points, with people without a diagnosis being those who perceive this situation most negatively.

The worst valued situations, in general terms, are attention to infrequent illness, access to public resources and the health situation, in order of highest to lowest percentage of negative perception.

2.6. Perceived discrimination.

With regard to the discrimination perceived by the group without diagnosis, it should be noted that the areas where discrimination has been perceived most are those related to social relations and daily life (in both cases, 81% have felt discriminated against or discriminated against on some occasion), followed by architectural access (63% of cases).

Likewise, and with respect to the ENSERio study, the undiagnosed persons have presented significantly higher figures of discrimination in the educational field (18 percentage points), in architectural access, daily life and in social relations.
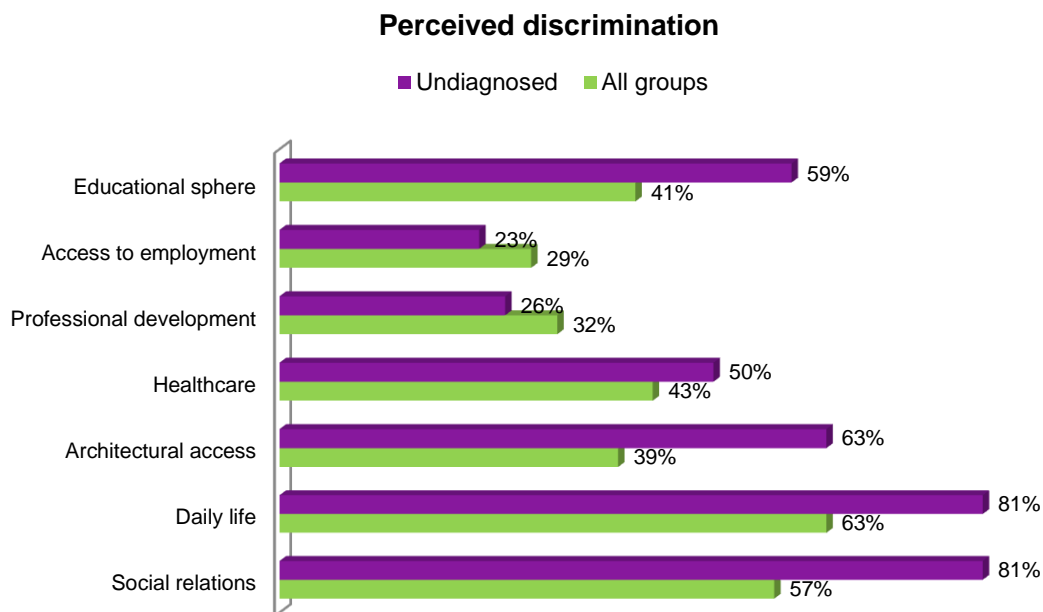
50

Perceived discrimination

■ Undiagnosed ■ All groups

Category	Undiagnosed	All groups
Educational sphere	59%	41%
Access to employment	23%	29%
Professional development	26%	32%
Healthcare	50%	43%
Architectural access	63%	39%
Daily life	81%	63%
Social relations	81%	57%

Chart 38. Percentage of people who has feel discriminated. *Comparison between all groups of the ENSERio study and undiagnosed patients*

Source: *ENSERio study.*

For its part, it is important to point out that, while the average discrimination perceived in the ENSERio study was 43.38%, among the undiagnosed people analyzed it was 54.7%, 11 percentage points higher.

3. CONCLUSION

People who do not have a diagnosis have fewer positive perceptions about the health care received related to their illness, compared to those with different RE pathologies analyzed in the ENSERio study.

In addition, this group has worse considerations about networking, and the multidisciplinary approach, at all levels of coordination, although especially in that connection between health services and other instances, such as social services or educational centers.

Half of the sample, 51%, have been waiting for a confirmed diagnosis for more than 10 years. The main cause of this delay in diagnosis identified by the people directly affected is the lack of knowledge about this type of pathology. In addition, the first consequence identified by more than one in four of the people is not receiving any support or treatment, the aggravation of the disease being the main consequence of the diagnostic delay identified in the ENSERio study.

Given that access to treatment is closely linked to obtaining a diagnosis, people who lack it, in comparison to the ENSERio study, claim to have access to the treatment they need to a lesser extent (24 percentage points below).

In addition, proportionally they register higher percentages of people who, needing to travel to obtain diagnosis, treatment and medication, were finally unable to do so.

Along the same lines, coverage by the public health system for treatments is generally lower than that perceived by the people who participated in the ENSERio study. In this way, the specific study of people without diagnosis reveals a lower perception of coverage in the services of: Early care, hearing aids, home nursing, physiotherapy, eyeglasses, contact lenses/visual aid, surgery/surgery, injections/perfusions, speech therapy, internal and external prostheses, psychology, psychomotricity, visual/Orto-optical re-education, rehabilitation, occupational therapy, health professional appointments, hospital admissions, tests and evaluations, treatment and medication, home care, respite care, transportation and supports such as wheelchairs or housing adaptation.

This figure needs to be considered especially in light of the results about disability and dependency. There is a higher percentage of undiagnosed people who claim to have some type of disability. In addition, among the persons with disabilities studied, those with undiagnosed disabilities report having higher percentages of recognized disability, with a difference of ten percentage points for disability greater than 75%.

 With regard to the certificate of dependency, it has been requested and obtained by almost half of the cases studied (43%). Among those who have been assessed, the assessments were, in more than half of the cases, for the highest degree of dependency.

In the ENSERio study, on the other hand, the grade III dependency ratings were percentage lower, as was the higher percentage of grade I ratings.

Along the same line, 73% of the undiagnosed people participating in the study claim to have impairments due to their illness to develop basic activities of daily life. It is worth mentioning that the perception of almost half of the people studied (45%) is that they have much limitation to develop these activities.

There are greater limitations in the development of basic activities among people without diagnosis, in all the analyzed daily activities. They present difficulties especially in the learning and application of knowledge, leisure and free time, educational or work activity, self-care and communication with other people.

Likewise, higher percentages with respect to the ENSERio study are reflected in the stretches of more hours of care per day of the person with a rare disease. Thus, higher percentages are observed in people who claim to dedicate more than 6 hours to the care of the person with HBS (57% compared to 34% in the ENSERio study). However, in both cases, the use of more than six hours for personal assistance was the most frequent option.

To deal with these difficulties, the people studied reported using assistive devices in one out of every four cases. It is important to note, moreover, that a similar percentage do not have these products, although they are needed, with a figure 10 percentage points higher than that collected in the ENSERio study. In addition to social and health care, the ENSERio study analyses the extent to which people with EPF could exercise their citizenship under full conditions, i.e. "if they have real access to rights recognized by law". In order to do so, its studies: housing, education and training, access to employment, the economic impact of the disease on the family budget and the subjective perception of their situation and discrimination.

The most significant data, on a comparative level, are those referring to the employment situation, the economic impact and the negative perception of their situation and discrimination.

Thus, among the people who indicate that they are working (30% of people of working age, a percentage similar to that of the ENSERio, 33%), there are higher percentages of bias in employment and more frequency of people who are self-employed than in the ENSERio study for total pathologies.

In addition, they also state to a greater extent that they need personal support and/or support products. The group without a diagnosis studied presents higher percentages of people who indicate that they need personal assistance, technical resources (technical support and accessibility) and specialized professionals. In addition, among the people who claim to need it, there are more those who indicate that it is not provided by the workplace, as was the case in the

ENSERio study for all groups of pathologies, although the group without diagnosis also has higher percentages in this sense.

In relation to the impact of the disease on the family economy, the highest percentage, approximately one in four people, claims to allocate between 20% and 30% of annual family income to the expenses of the disease, percentages notably higher than in the ENSERio study, whose most frequent response option, also one in four, was to allocate less than 10% of annual income.

Along the same line, 82% of undiagnosed people studied value the cost of their rare disease high or very high, 11 percentage points higher than that collected in the ENSERio study. Linked to this circumstance, undiagnosed persons also present higher percentages of difficulty in assuming the expense, eight percentage points above those who identified the same difficulty in the ENSERio study.

These data also coincide with a more negative assessment of their situation and the perception of greater discrimination in different spheres. The area or situation worst valued, with 21 percentage points of difference with respect to the percentage of people who value this situation negatively, is attention to illness.

In this sense, while the average discrimination perceived in the ENSERio study was 43.38%, among the undiagnosed people analyzed it was 54.7%, 11 percentage points higher. Especially in the field of education (18 percentage points), architectural access, daily life and social relations.