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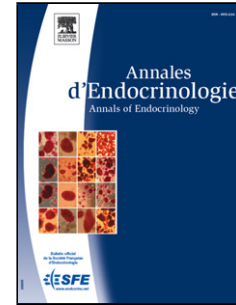
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The experience of diagnosis announcement in rare endocrine diseases:  
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## The experience of diagnosis announcement in rare endocrine diseases: a survey of the French FIRENDO network

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**Short title:** Diagnosis announcement in rare endocrine diseases

**Keywords:** diagnosis announcement, rare disease, endocrine disease, Addison's disease, Congenital Adrenal Hyperplasia, Silver-Russel syndrome, Cushing's syndrome, acromegaly, craniopharyngioma

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## ABSTRACT

### Context

Diagnosis announcement of a chronic disease is a crucial moment for patients as well as for their families and an important step in the management of severe conditions such as rare endocrine diseases. Little is known of how diagnosis is communicated to patients and families. The FIREENDO network was created by the third French Plan for Rare Diseases, to promote autonomy, care and research on rare endocrine diseases.

### Objectives

The aim of this study was to characterize, for the first time, the experience and needs of patients and/or their parents around the announcement of diagnosis to ensure optimal quality of care.

### Methods

A quantitative self-administered survey on diagnosis announcement procedures in rare endocrine diseases was launched in April 2017 by the ad hoc FIREENDO thematic working group in collaboration with its 11 partnering patient associations and support groups. The questionnaire was designed and revised by patient support group representatives, adult and pediatric endocrinologists, psychologists and biologists, all expert in rare endocrine diseases. It was made available on the FIREENDO network website and distributed mainly by email with electronic links on their respective websites to members of all affiliated patient support groups.

### Results

Questionnaires were filled out by 391 patients and 223 parents (median age of patients: 39 years). The following conditions were associated with at least 30 answers: Addison's disease, classical forms of congenital adrenal hyperplasia (CAH), Russell-Silver syndrome, Cushing's syndrome, acromegaly and craniopharyngioma. Overall, some announcement modalities were judged favorably by patients: physician's empathy, availability and use of clear terms, and presence of family at the time of announcement. However, a lack of psychological care and information documents was reported, as well as some inadequate procedures such as postal mail announcements.

### **Conclusion**

This work suggests that better knowledge of the patient's experience is useful for improving the diagnosis announcement of rare endocrine disorders. The main recommendations derived from the survey were the need for several announcement visits, information on patient support groups and reference centers, imperatively avoiding impersonal announcement, and the usefulness of a written accompanying document.

### **Keywords:**

Diagnosis announcement, rare disease, endocrine disease, Addison's disease, congenital adrenal hyperplasia, Russell-Silver syndrome, Cushing's syndrome, acromegaly, craniopharyngioma

### **Introduction**

Information to patients, and particularly the diagnosis announcement of a chronic or rare disease, is complex and never insignificant for the patient or for the clinician (1,2). In Europe, a disease is called "rare" when it affects less than one in two thousand people. In endocrinology, this applies to more than two hundred groups of rare endocrine diseases with more than thirty thousand patients concerned in France. Too frequently, the search for diagnosis can extend over several years (3). The rarity of each disease leads to several specific issues compared to more common diseases: scarcity of information, unavailability of treatments, delays in diagnosis, difficulty in finding a specialist, disability issues, depression and anxiety, social care access and isolation (4,5).

The revelation of a rare disease represents a forever-remembered event for patients and their families. It is an "eternal moment", marking the end of the period of life when the disease was absent (1). The time of the illness begins well before diagnosis. When the patient has been seeking for many years to know what is wrong, the diagnosis is initially experienced by some as a relief but may be experienced by others as a tragedy. In where the disease can be transmitted, such as some genetic diseases, patients or parents may experience a sense of guilt. In addition, the diagnosis of a rare disease in a child is experienced as a cataclysmic event that disrupts parental landmarks and generates an emotional crisis in the family (6,7).

There are several studies on the experience of receiving a diagnosis of common chronic diseases, particularly in cancer (8,9), but much fewer in patients with rare diseases (10,11). In 2009, the European Organization for Rare Diseases carried out two surveys to describe and compare patients' experiences and expectations about diagnosis and access to health services. These surveys provided useful information about 16 diseases in 12,000 patients in 17 countries and aimed to contribute to shaping patient-centered public health policies (12).

Despite some diagnosis-specific studies, little knowledge exists about the diagnosis announcement procedure in rare endocrine diseases as a whole. The diagnosis announcement, however, is a crucial moment for patients and families and an important

step in the management of rare and severe conditions. The aim of the present study, carried out by the French National Healthcare Network for Rare Endocrine Diseases (FIRENDO) as defined by the third National Plan for Rare Diseases (13), was to characterize, for the first time, the experience of patients and/or their relatives around the announcement of diagnosis, and to define their needs, in order to ensure optimal quality of care.

## 1. Methods

### 1.1 Design

One of the thematic groups of the FIRENDO network devoted to relationships with patient associations had identified diagnosis announcement modalities and their consequences on disease management as a main topic of interest that should be addressed in priority. Patient associations (n=11) are active members of the French FIRENDO healthcare network. A quantitative self-administered survey on diagnosis announcement procedures in rare endocrine diseases was launched from April 1st, 2017 to November 1, 2017, under the auspices of the FIRENDO network in collaboration with its partnering patient support groups. The questionnaire was designed and revised by patient support group representatives, adult and pediatric endocrinologists, psychologists and biologists, all expert in rare endocrine diseases. The questionnaire comprised 22 questions; open comments/suggestions fields were also included. Quantitative assessments were based on scales from 1 to 10. Qualitative assessments and verbatims were assessed and summarized by the same group of experts who participated in the questionnaire design. It was made available on the FIRENDO network website and publicized mainly by email to members of patient support groups affiliated to the network, via electronic links on their respective websites (survey available as supplementary data).

### 1.2 Participants

All participants (patients or parents of patients) were volunteers. They were informed about the aim of the study, and that the questionnaires were anonymous. All patients with a rare endocrine disease or their relatives (for patients under 18 years old), having received the information from the FIRENDO network website or from the patient support groups, were eligible to take part in the study. The 11 participating patient support groups were: the Association of Chromosomal Abnormalities Carriers (*Association de Porteurs d'Anomalies Chromosomiques*: Valentin APAC), McCune-Albright Syndrome and Bone Fibrous Dysplasia Patients Association (*Association des Malades Porteurs du Syndrome de McCune-Albright, de Dysplasie Fibreuse des Os*: Assymcal), Association of Patients Suffering from Infertility Problems (*Association de patients confrontés à l'infertilité*: MAIA), Rokitansky Syndrome Association (*Association Syndrome de Rokitansky*: MRKH), Craniopharyngioma Solidarity Association (*Association Crâniopharyngiome Solidarité*), Acromegalic Patients Association (*Acromégales Pas Seulement*: APS), Wolfram Syndrome Association (*Association Syndrome de Wolfram*), French Association of Families Affected by Silver Russell Syndrome (*Association Française des Familles touchées par le syndrome de Silver Russell et des Personnes Nées Petites pour l'Age Gestationnel (PAG) et Leurs Amis*: AFIF SSR/PAG), Association for Patients with Growth Disorders (*Association Grandir*), French Adrenal Diseases Association (*Association Surrénales*), and Association for Patients with Thyroid Dysfunction (*Association Vivre sans Thyroïde*).

### 1.3 Data analysis

Only diseases with  $\geq 30$  responses from patients or parents of patients with the corresponding diagnosis were analyzed for quantitative data, while the whole cohort was taken into account for qualitative assessment. Continuous variables were compared on

Wilcoxon test. Comparison of several continuous variables (>2) used Kruskal-Wallis test. Comparison of qualitative variables used chi<sup>2</sup> test (or Fisher as appropriate). The significance threshold set at  $p < 0.05$  (5% alpha threshold).

## 2. Results

### 2.1. Characteristics of respondents

Among the 614 collected responses, 391 respondents were patients (63.7%) and 223 were patient's parents (36.3%). Median age was 39 years. The groups of pathologies (n=35) were diverse and representative of the whole spectrum of rare endocrine diseases covered by all relevant reference centers as defined by the National Plan for Rare Diseases, including diseases of the thyroid, hormone receptors, pituitary gland, sex development from fetus to adult, insulin secretion and action, gynecological disorders, growth and development disorders, and adrenal gland diseases. Median age at diagnosis was 27 years (Table 1).

### 2.1 Experience of the diagnosis

#### 2.1.1 Experience according to pathology

Only pathologies with  $\geq 30$  responses were analyzed. Seven distinct disease categories, corresponding to a total of 425 patients (69.2% of the whole cohort), reached this threshold, as detailed in Table 1: Addison's disease (18% of respondents), congenital adrenal hyperplasia (CAH) (13%), Silver-Russel syndrome (12%), acromegaly (11%), Cushing's syndrome (6%), Cushing's disease (5%) and craniopharyngioma (5%). Patient experience differed significantly (Fig. 1) between the 7 pathologies ( $p < 0.0001$ ). CAH, craniopharyngioma and acromegaly were the most poorly experienced pathologies (median 2, 2 and 3, respectively).

Some comparisons were made (Table 2) between disease categories sharing common features: pituitary adenoma (acromegaly vs. Cushing's disease), and adrenal insufficiency (CAH vs. Addison's disease). Diagnosis was made significantly later in patients with acromegaly than in patients with Cushing's disease (median 5 years vs. 3 years). Similarly, there was a tendency for acromegaly patients to have a poorer experience with the announcement, and by patients with CAH than with Addison's disease (median 2 vs. 5).

#### 2.1.2 Patients vs Parents

Overall, the diagnosis announcement was not well experienced, and parents had a significantly poorer experience than patients (median 3 vs 5 respectively).

In describing how they learned about the diagnosis, participants talked about difficulties with the way information was communicated to them. Representative verbatim samples selected by the data review group included the following. One patient said "I had trouble understanding the nature of my illness. The endocrinologist gave me explanations too quickly, with complex vocabulary". Another said: "At last! They believed me". One parent reported: "Having no knowledge about the disease, I thought the sky was falling on my head". Another parent said, "While in apparent good health, we were told that our child is carrying a serious congenital illness, which generated misunderstanding". Additional quotes are given in Appendix 2.

#### 2.1.3 Context of diagnosis announcement.

For 80% of patients, diagnosis was communicated during an outpatient visit, 9% by phone, and 2% by postal mail, and mainly by endocrinologists rather than other health care professionals. The rating given to announcement significantly differed depending on the context ( $p < 0.0001$ , Fig. 2A). The diagnosis announcement was generally well experienced when made during an outpatient visit (median 7), or by phone (median 6), while postal mail announcements received the lowest score (median 1.5). On the other hand, the



announcement experience did not significantly differ according to where it was made (community office or hospital setting).

The announcement for patients with early diagnosis ( $\leq 1$  year between symptom onset and diagnosis) was significantly less well experienced than for those with later diagnosis (Fig. 2B).

#### **2.1.4 Communicating additional information on the disease.**

There was no significant difference between the scores of patients who were told that the disease was rare versus those who were not told (median: 4). 69% of respondents considered that the vocabulary used was appropriate, and 66% were able to ask any questions about the disease.

For about 62% of patients, diagnoses were given without enough information on the disease, while 36% considered the information to be sufficient and 3% considered they got too much information (Fig. 3A). Diagnosis announcement was well experienced when sufficient information was given, whereas patients and parents had a more negative experience of diagnosis when they were given too much information (Fig. 3B).

At the diagnosis announcement, 68% of respondents reported that they were not given a document summarizing their pathology, although the announcement experience did not significantly differ between those who had been given a document and those who had not. There was also no significant difference between the experience of patients who were well informed about follow-up care (number and rate of outpatient visits or hospital admissions) vs. those who were not (both, median: 4).

#### **2.1.5 Who informs the patient and family circle about the disease?**

Asked who provided the most information, 47% of respondents cited a specialist physician, 38% a patient organization and 37% the Internet (Fig. 4A). Patients reported supplementary learning through Internet searches, but that they needed guidance from health care providers to evaluate the quality of Internet health information sources. At the announcement, 39% of respondents considered that they were not helped by anyone, 33% were helped by the medical doctor, 3.7% by the hospital nurse, and 1.3% by the psychologist. About 71% of patients reported that the psychologist's help was not proposed. Unexpectedly, the diagnosis announcement was less well experienced (median 3 versus 5;  $p=0.011$ ) when the psychologist's help was proposed (Fig. 4B).

### **3. Discussion**

Rare endocrine diseases are most often chronic, can be progressive and can sometimes lead to life-threatening emergency situations. One of the most difficult moments for a patient with a rare disease is the diagnosis announcement. He or she may have waited a long time before putting a name to their medical condition, but the announcement is so overwhelming that the patient finds it very difficult to understand, accept and be able to explain this diagnosis to relatives (1). The announcement of a rare disease and providing adequate information is never easy. It is experienced as a tragedy and often considered as an "eternal moment". However, the announcement also puts an end to a period of uncertainty, especially if the quest for a diagnosis has been particularly long; in that case, the diagnosis is initially experienced as a relief, and the patient finally knows more precisely what must be fought against.

Owing to the structured management pathway for rare diseases in France (13), this study was able to generate data about diagnosis experience based on a survey carried out in a large population of patients with rare endocrine diseases within the FIREENDO network and its partnering patient support associations. Results from 614 patients and relatives covered 35 different diseases. Participants represented a panel of FIREENDO users and patient association website users, and therefore may not be fully representative of all endocrine

rare disease patients in France. Out of all the quantitative data from the online questionnaire, a subset of 425 patients, in 7 disease categories with at least 30 respondents each, was more particularly analyzed in the present study.

Among the different rare endocrine diseases selected and analyzed in this study, patients' experiences differed according to pathology, as previously found in other studies (14,15). In the present study, CAH, craniopharyngioma and acromegaly were the most poorly experienced diagnoses, probably for various reasons pertaining to the specificities of each (16, 17, 18). It is well established that a diagnosis of CAH in newborns is not well experienced by parents. Parents often feel overwhelmed by the prospect of caring for a child with a severe illness, and usually have limited ability to absorb the information provided, particularly when special care is needed and an interdisciplinary team is required (19,20). Parents highlight the need to communicate clearly explained health information, addressing emotional needs and providing social support. The poor diagnosis announcement experience of patients with acromegaly or craniopharyngioma may partly be related to the impaired quality of life (QoL), daily life and specific comorbidities of these patients. As reported in several studies, QoL is impaired in all pituitary diseases, marked by factors such as anxiety, delayed diagnosis, physical appearance, social isolation and ongoing medical therapy (21–23).

Patients in this study reported that the diagnosis announcement was often poorly experienced, with poorer experience scores in parents than in patients themselves. For families, receiving a diagnosis of a rare, chronic and complex disease for their child is often devastating, representing a life-changing event that implies profound adaptive challenges (24–26). Parents often feel socially isolated, anxious and stressed, especially by delays in treatment (27,28). Representative verbatim samples in this study (Appendix 2) were in agreement with previous studies, where patients and relatives' testimonies reported delayed diagnosis, and a feeling of not being heard and believed (29,30). In the European Organization for Rare Diseases survey, correlations were observed between longer time to diagnosis and prior misdiagnosis, and between shorter time to diagnosis and personal initiatives of rare disease patients, such as finding a diagnostic structure themselves (12).

The context of diagnosis announcement is never insignificant, as seen in the present study where announcement was better experienced during patient visits than by postal mail. It must observe some rules of temporality in its enunciation, and take into account the wishes of patients considered as full actors of their health.

As demonstrated by our results, diagnosis announcement is better experienced when sufficient information is given. Patients wish to be informed about their disease and treatments, and always look for any missing information via dedicated websites or patient associations, if it has not been sufficiently clear and complete with the doctor. In addition to verbal communication, patients request written information, as they often find information difficult to remember. The diagnosis announcement and the way it is conducted have an impact on patients' experiences and their ability to apprehend the outcome of their illness, because patients' psychological profiles differ according to individual differences and cultural environment. Medical providers must take into account each patient's capacity to absorb complex and novel information, which is affected by emotional state, volume of information, and health literacy (8,31). The announcement of bad news is part of a relationship of care, where trust constitutes the start of a whole therapeutic act. While some patients are relieved and start fighting, others totally collapse or even conceal the issue. Patients emphasized the importance of knowing and understanding what the endocrinologist or other medical provider is trying to explain. Knowledge has been described as a means to feel empowered, to regain control and to partner with health care providers (32, 33). The medical announcement of a serious illness, whether chronic or

rare, must include the fundamental elements of “fair, clear and appropriate” information (Article 35 - Health care) but also the need for progressive and respectful communication. Health professionals’ lack of knowledge of the patient’s specific rare disease was reported several times in patients’ testimonies. Patients’ perception of medical care showed that health professionals, in particular outside of the hospital, did not appear to have enough knowledge about rare pathologies. This perception correlated with the delay in diagnosis (34). Rare diseases involve a great variety of medical specialists and all of them should have some training in rare diseases. Therefore, the initial and continuing training of physicians should be strengthened to guide the patient’s pathway. Addressing this aspect is a major concern of the Third National Rare Diseases Plan (13) involving the French FIRENDO network, together with reference and skills centers.

Receiving a diagnosis of a rare chronic and complex disease is a life-changing event for many patients and their families. Further, beyond disease and symptom burden, the rarity of the condition can result in patients and families feeling marginalized. In addition, the psychosocial impact of perceived invisibility, isolation and a sense of helplessness can have significant deleterious impact on quality of life. The fact that psychological support was likely proposed to patients with the most severe prognoses may be a bias that can explain the paradoxical finding of an inverse relationship between proposal for a psychologist’s intervention and poorer announcement experience. Various needs and issues were reported in this study: psychological support seemed to be inconsistently proposed and should certainly be taken in consideration: several patients highlighted the need to embed psychological counsellors in all departments where severe diagnoses are announced. The French health care planners and providers should consider strategies to increase the provision of psychological support to all patients and families who have recently received or are soon to receive a diagnosis such as that of a rare endocrine disease.

### **Conclusion**

Patients affected by rare diseases are dispersed and face a variety of challenges: lack of specialized care, delays in diagnosis, negative social consequences, and other psychosocial burdens. The present study confirmed that better knowledge of the patient’s experience is useful for improving the diagnosis announcement in rare endocrine disorders. The main recommendations derived from the survey were the need for several announcement visits, and for providing information on patient support groups and reference centers, imperatively avoiding impersonal announcement, and the usefulness of a written accompanying document.

### **The experience of diagnosis announcement in rare endocrine diseases: a survey of the French FIRENDO network**

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### **ETHICAL STATEMENT**

**Declarations of interest:** The authors have no conflicts of interest to declare in relation with the present article

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**Legends to Figures**

Fig. 1. Patient's experience scores in the 7 endocrine rare diseases with 30 respondents or more. Patient experience significantly differed between these pathologies ( $p < 0.0001$ ). The numbers of patients in each disease group are given in Table 1.

Fig. 2. (A) Rating given to announcement depending on context. Patient experience significantly differed between these distinct modalities ( $p < 0.0001$ ). (B) Rating given to announcement experience in patients diagnosed within 1 year of first symptoms (early diagnosis) or after a longer delay. Patient experience score was significantly better in patients with an early diagnosis vs. patients with a longer time to diagnosis ( $p < 0.0001$ ).

Fig. 3. (A) Patients' or parents' evaluation of the amount of information provided at diagnosis announcement. (B) Patients or parents' diagnosis announcement experience according to the amount of information provided. The difference between the 3 categories (not enough, sufficient or too much information) was significant ( $p < 0.0001$ ).

Figure 4: (A) Origin of the information on the disease provided to the patient and family. (B) Patient's experience whether a psychologist's intervention had been proposed.

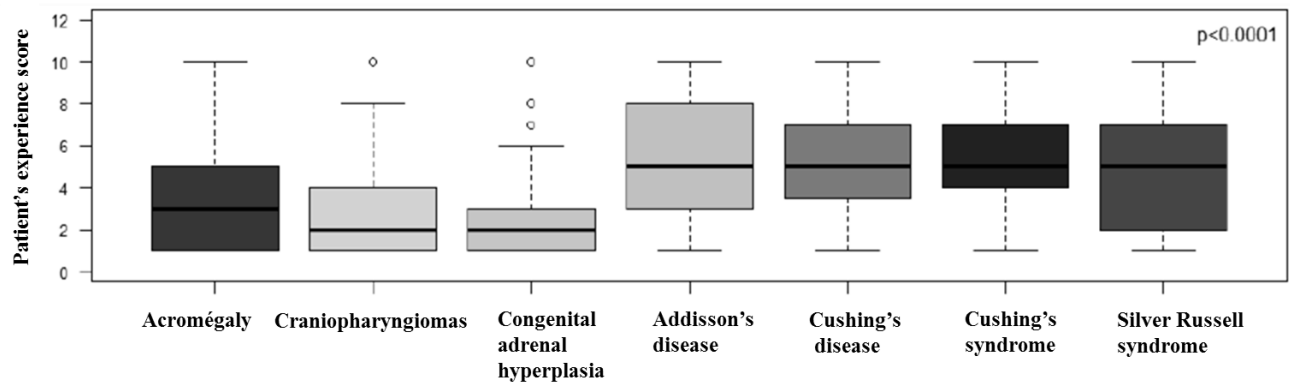
**Legends to Tables**

Table 1: Characteristics of respondents ( $n = 614$ ). IQR: Interquartile Range, represents the range of the middle 50% of the sample.

Table 2: Experience according to pathology (acromegaly vs Cushing's disease, CAH vs Addison's disease)



Figures  
Figure 1:



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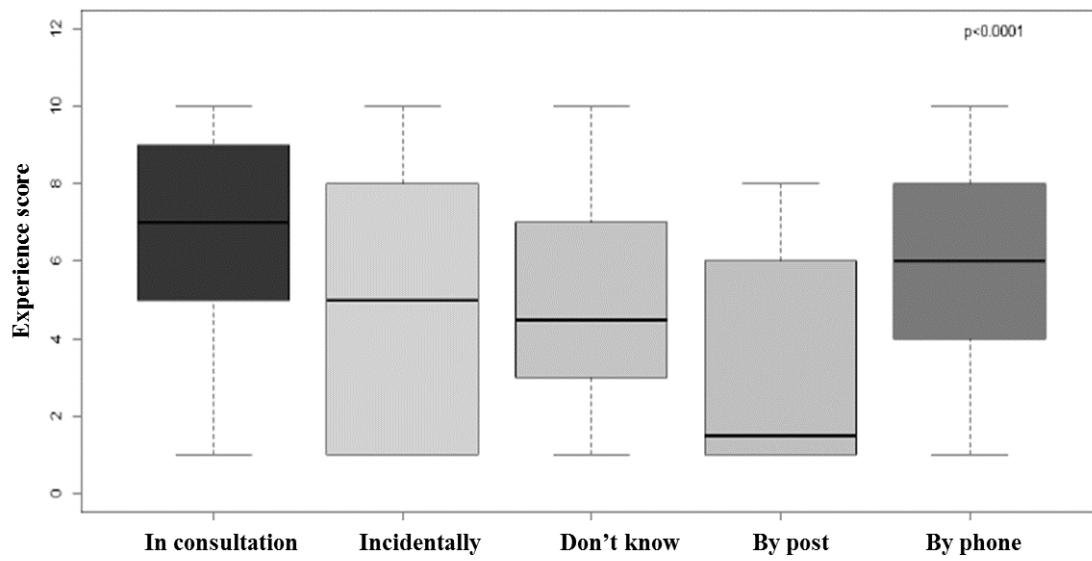
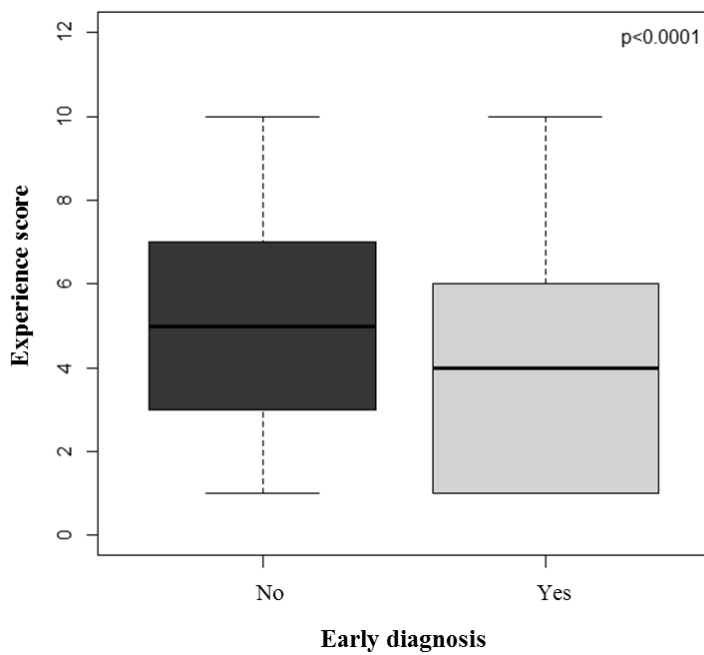
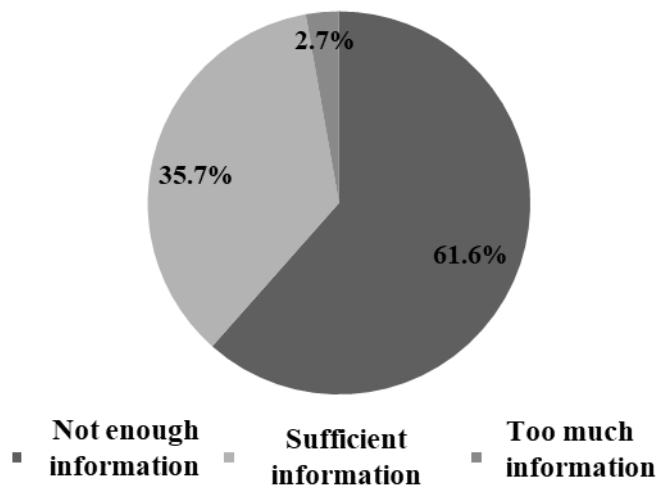
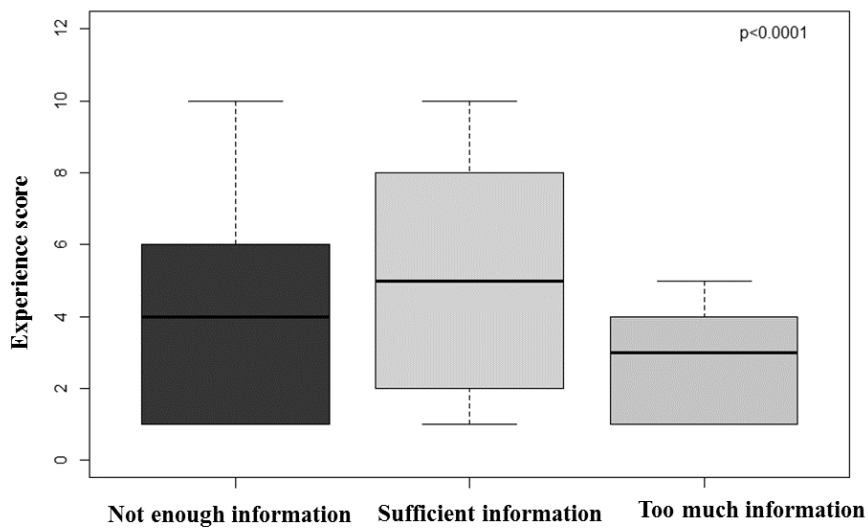
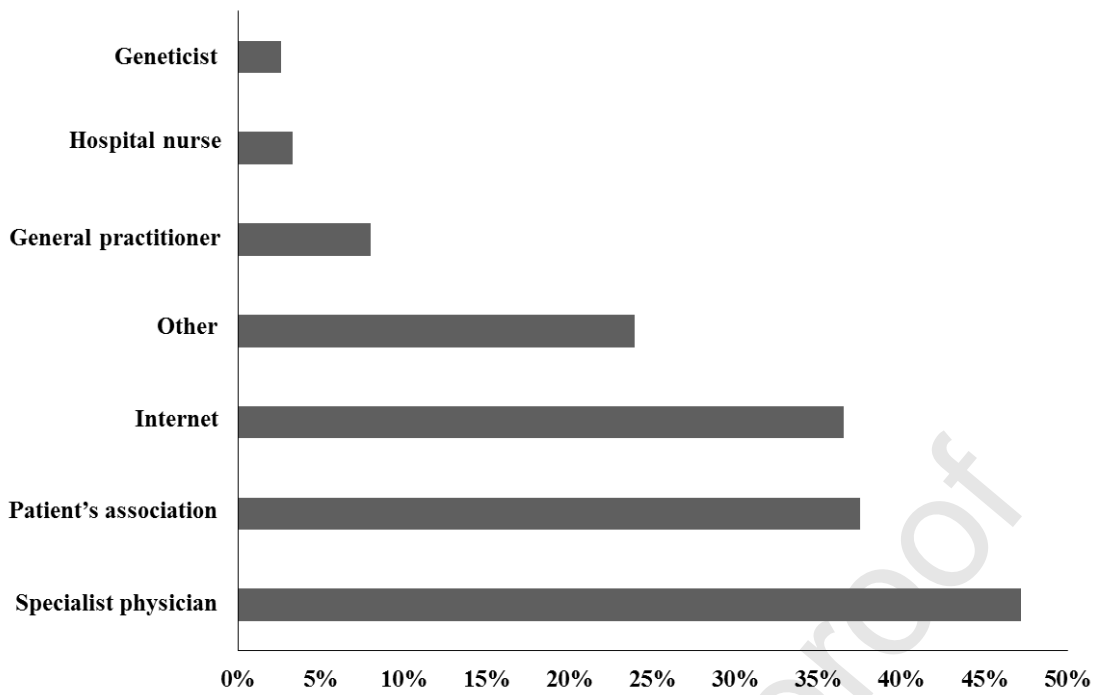
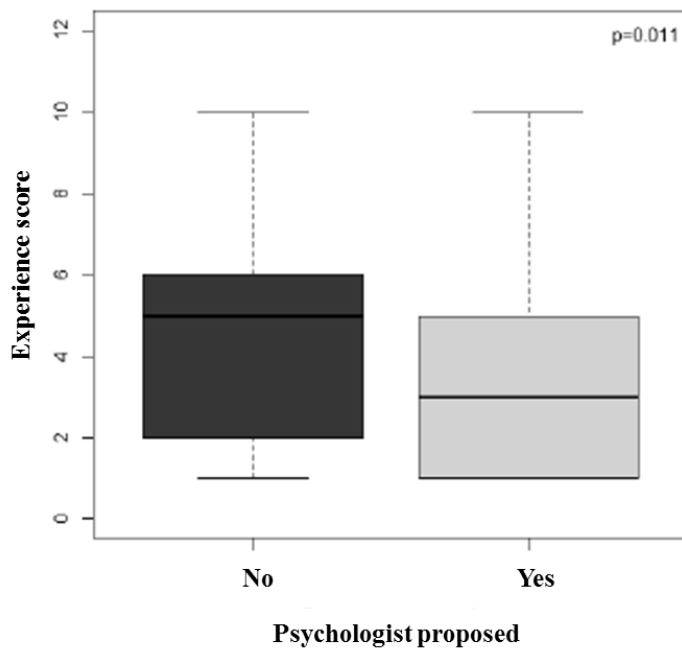
**Figure 2:**  
**A****B**

Figure 3:  
A



B



**Figure 4:**  
**A****B**

## Tables

Table 1

	Number	percent
Patient's parent(s)	223	36.3
Patient	391	63.7
	<b>Median [IQR]</b>	<b>Range</b>
Year of patient's birth	1978 [1964-2002]	(1934-2016)
Patient's age (years)	39 [15-53]	(1-83)
Year of first symptoms	2007 [1999-2012]	(1953-2017)
Age of patient at onset of symptoms (years)	22 [0-37.5]	(0-75)
Year of disease diagnosis	2009 [2002-2014]	(1964-2017)
Age of patient at diagnosis (years)	27 [2-42]	(0-75)
<b>Relevant Pathology</b>	<b>Number</b>	<b>percent</b>
Addison's disease	108	17.6
Congenital adrenal hyperplasia: classic 21 hydroxylase deficiency	81	13.2
Silver Russell syndrome	74	12.1
Acromegaly	66	10.7
Cushing's disease	36	5.9
Craniopharyngioma	30	4.9
Cushing's syndrome	30	4.9
Other rare endocrine disease	24	3.9
Non-functional pituitary adenoma	14	2.3
Pheochromocytoma	13	2.1
Isolated congenital growth hormone deficiency	12	2
Hypopituitarism (pituitary insufficiency)	12	2
Acquired pituitary insufficiency	12	2
Other: my disease is not on this list	11	1.8
Secondary adrenal insufficiency (corticotropic insufficiency)	11	1.8
McCune Albright syndrome	10	1.6
Congenital adrenal hyperplasia: non-classical 21 hydroxylase deficiency	9	1.5
Congenital adrenal hyperplasia without 21 hydroxylase deficiency	8	1.3
Panhypopituitarism	8	1.3
Mixed pituitary adenoma GH+PRL	5	0.8
Prolactin adenoma	4	0.7
Conn's adenoma	4	0.7
Acquired central diabetes insipidus	4	0.7
Gonadotropic adenoma	3	0.5
Pituitary apoplexy	3	0.5
Carney complex	3	0.5
Multiple congenital pituitary deficiency (without diabetes insipidus)	3	0.5

Congenital hypothyroidism with abnormal development of the thyroid gland	2	0.3
Ectopic posterior pituitary and pituitary stalk abnormality	2	0.3
Without precise diagnosis	2	0.3
XY/XO gonadal dysgenesis	1	0.2
Germinoma	1	0.2
Primary hyperaldosteronism	1	0.2
Congenital adrenal hypoplasia	1	0.2
Infiltrative pituitary insufficiency (histiocytosis, sarcoidosis, beta-thalassemia)	1	0.2
Isolated hereditary adrenal insufficiency	1	0.2
Non-rare disease	1	0.2
Paraganglioma	1	0.2
Glucocorticoid resistance	1	0.2
Turner's syndrome	1	0.2

**Table 2**

	<b>Congenital adrenal hyperplasia</b>	<b>Addison's disease</b>	<b>p-value</b>	<b>Cushing's disease</b>	<b>Acromegaly</b>	<b>p-value</b>
	<b>Median [IQR]</b>			<b>Median [IQR]</b>		
Diagnostic delay (years)	NA	NA		3 [1-6]	5 [2-10]	0.016
Announcement experience	2 [1-3]	5 [3-8]	<0.0001	5 [3.8-7]	3 [1.2-5]	0.05

## Appendix 1 Survey on diagnosis announcement procedures in rare endocrine diseases.

1. You are  
parent(s) of the patient  
patient
2. Patient's year of birth:
3. Year of onset of first symptoms:
4. Diagnosis:  
Year of disease diagnosis:  
Year of diagnosis announcement if different:
5. Pathology involved:
6. Before the diagnosis  
Before the diagnosis was formally made by a physician, did you suspect that you or your child had a rare disease?  
No  
Yes  
Do you have any doubts about the diagnosis?  
No  
Yes
7. Who told you the diagnosis?  
General practitioner  
Pediatrician  
Endocrinologist and/or diabetologist  
Pediatric endocrinologist  
Geneticist  
Gynecologist  
Hospital intern  
Ophthalmologist  
Don't know  
Other
8. In which context was the announcement made?  
In consultation  
By telephone  
By mail  
Incidentally  
Don't know
9. Place of diagnosis  
Hospital  
Community office  
Don't know  
Other
10. How do you think this announcement was made to you?  
Tactlessly 1 2 3 4 5 6 7 8 9 10  
Comment:
11. Overall, how did you experience this announcement?  
Very badly 1 2 3 4 5 6 7 8 9 10 very well  
Why:
12. Was a psychologist's help offered to you?  
Yes  
No  
Don't remember
13. Who helped you most at the diagnosis announcement?

General practitioner

Medical specialist

Hospital nurse

Hospital psychologist

Person

Other (comment below)

14. Did you have the opportunity to ask any questions?

Yes

No

Don't remember

15. Did you find the vocabulary used appropriate?

Yes

No

Don't remember

16. Were you told that it was a rare disease?

Yes

No

Don't remember

17. Were you aware of the next step in the care process?

The people you were going to meet?

Yes

No

The number of consultations and at what frequency?

Yes

No

The number of hospital admissions and at what frequency?

Yes

No

I am not concerned

Was time taken to talk to you/your child (depending on age) about the disease/syndrome?

Yes

No

I am not concerned

18. Did you receive a document summarizing the information?

Yes

No

19. Were there several visits?

Yes

No

Don't remember

20. Did you find that there was:

Not enough information

Enough information

Too much information

21. Who provided you with the most information about the disease?

General practitioner

Medical specialist

Hospital nurse

Geneticist

Patient association



Internet

Other

22. How do you think the diagnosis announcement of a rare disease can be improved?

Announcement in the presence of several healthcare providers

Yes

No

In the presence of relatives

Yes

No

Several interviews

Yes

No

Your suggestions

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**Annex 2 Representative quotes from patients selected from the qualitative study.**

Theme	Representative quotes
The way diagnosis was given	<p>“We expected it, it was only a confirmation for us”</p> <p>“I was considered as a person, an actor of my own disease, in dialogue with the doctor who established the diagnosis; doctor that I felt invested in the study of my case; I felt considered and in confidence with this doctor through the dialogue, the attitude, the chosen words and the determination shown by the practitioner to follow me and not to just turn me loose.”</p> <p>“Minimalist and ultra de-dramatized information on the pathology. On the doctor's face, we could feel that it is not a cold and I "threw myself" on the Internet for hours as soon as I left the consultation.”</p> <p>“Very simplistic announcement, as if it was some kind of disease. I only understood the seriousness of my illness later, by being sick and especially in the therapeutic education workshops.”</p> <p>“Given the vital urgency of the situation, a brutal but pragmatic announcement! During a scheduled "routine" consultation, after 2 weeks”</p>
Experience when receiving diagnosis	<p>“At last they believed me”</p> <p>“For me, it was a real earthquake and nothing less than the end of my world.”</p> <p>“I felt like the ground was giving way under me. No doubt because of the late diagnosis and the eleven years of suffering. Then, a few seconds after this announcement, a certain peace: a name had just been put to what I had been experiencing for too long. Nobody could think that it was just "in my head" anymore...”</p> <p>“Very bad but at the same time a real relief to know that it's something!”</p> <p>“We feel guilty for having transmitted the disease to our child”</p> <p>“In good health we are told that our child has a serious congenital disease. Incomprehension, fear of losing our child and then fear about his future life”</p>

Suggestions	<p>“Referral to associations and a link to reference centers.”</p> <p>“A psychologist could be proposed to digest the announcement of the disease.”</p> <p>“Suggest other interviews after the diagnosis is announced so that all or some of the questions that come up in the aftermath of the diagnosis can be asked. “</p> <p>“With hindsight, I find that everything that medicine and its practitioners have done for me is remarkable. Caring support is essential. Thank you”</p> <p>“It would be good to systematically give an information document to the patient because, once the shock of the announcement has passed, we forget 95% of the information that was given to us”</p> <p>“The most difficult thing is not the announcement but afterwards. Managing work or studies, to get your relatives to understand your daily life”</p>
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