Research Square

Preprints are preliminary reports that have not undergone peer review.
They should not be considered conclusive, used to inform clinical practice,
or referenced by the media as validated information.

The psychological care gap in rare diseases: evidence of barriers and inequalities in access in Spain

Laura González-Riesco
Hospital Sant Joan de Déu
Marta Llorens-Capdevila
Hospital Sant Joan de Déu
Emma Prades-Morera
Hospital Sant Joan de Déu
Begonya Nafria-Escalera
begonya.nafria@sjd.es

Institut de Recerca Sant Joan de Déu
Diana Garcia-Ramon
Institut de Recerca Sant Joan de Déu
Helena Martínez-López Amor
Institut de Recerca Sant Joan de Déu
Mireia Llopart-Carrillo
Federación Española de Enfermedades Raras
Mireia Soba-Martínez
Federación Española de Enfermedades Raras
Magdalena Belando-Llor
Federación Española de Enfermedades Raras
Estrella Mayoral-Rivero
Federación Española de Enfermedades Raras
Josep Matalí-Costa
Hospital Sant Joan de Déu

Research Article

Keywords: Rare diseases, psychological care, mental health, healthcare access, referral pathways, health inequalities, patient-centred care, patient organisations, public health system

Posted Date: November 24th, 2026

DOI: https://doi.org/10.21203/rs.3.rs-11660897/v1

License: This work is licensed under a Creative Commons Attribution 4.0 International License.
Read Full License

Additional Declarations: No competing interests reported.

Abstract

Background

Rare diseases (RDs) are associated with unique emotional and psychosocial challenges for affected individuals and their families. Despite increasing recognition of mental health as an important component of comprehensive care for RDs, evidence regarding access to psychological services remains limited. This study examined access to psychological care in RDs in Spain to identify barriers, facilitators, inequalities and unmet needs.

Methods

A national cross-sectional online survey was conducted between December 2023 and October 2024 through the Share4Rare digital platform in partnership with the Spanish Federation of Rare Diseases (FEDER). Adult patients and caregivers of individuals with RDs completed a questionnaire assessing access to psychological care, including referral pathways, perceived barriers, funding sources, satisfaction, adherence, and unmet needs. Descriptive and comparative analyses were performed according to age, gender, diagnosis group, and region of residence.

Results

The 792 responses comprised 56.9% (n = 451) adult patients and 43.1% (n = 341) caregivers. Nearly half of participants (49.7%, n = 394) reported not having received psychological care during the previous year. The most frequent barrier was the absence of a recommendation or referral from a healthcare professional (79,7%, n = 310), followed by financial barriers (13%, n = 54) and logistical constraints (6%, n = 25). Only 33.7% (n = 84) of individual therapy interventions, 24.3% (n = 18) of family interventions, and 31.6% (n = 24) of group interventions were funded through the public healthcare system. Significant inequalities in access to psychological care were observed according to gender, age, diagnostic group, and region of residence. Individual therapy received the highest satisfaction ratings (85.8%, n = 205) and showed greater adherence (70.1%, n = 169 completed all sessions) than family- or group-based interventions. Patient organisations emerged as key facilitators of access by providing psychological services and/or contributing to treatment costs.

Conclusions

One in two people affected by RDs reported not receiving psychological support during the previous year. Lack of referral or recommendation from healthcare professionals emerged as the principal barrier to access, over financial and logistical constraints. These findings suggest that psychological care remains insufficiently integrated into RD care pathways and highlight the need for systematic psychological

assessment, routine referral mechanisms, equitable public funding, and stronger integration of mental health services within RDs care models.

1-INTRODUCTION

Rare diseases (RDs) comprise more than 7,000 heterogeneous conditions that, individually, affect a very small number of people, but which collectively represent a significant public health challenge. In the European Union, RDs are defined as conditions that affect fewer than 5 people per 10,000 inhabitants [1]. Collectively, they affect approximately 5% of the population, with more than 30 million people living with a rare disease across Europe [2]. Many of these diseases are chronic, multisystemic, and disabling, often genetic in origin and with onset during childhood. Furthermore, they are often associated with delays in diagnosis, limited access to specialised clinical care, lack of specific treatments, and considerable uncertainty regarding prognosis. These characteristics create complex care needs and are associated with a poorer quality of life and a significant psychosocial burden for both affected individuals and their families [3–5]. Consequently, RDs require integrated, multidisciplinary, and person-centred care models that respond in a coordinated manner to the medical and psychosocial needs of patients and their families.

Among the many challenges associated with RDs, diagnostic uncertainty constitutes one of the most significant sources of emotional vulnerability. Many patients and families experience a prolonged "diagnostic odyssey", characterised by repeated medical consultations, delayed recognition of the condition, misdiagnosis, and difficulties accessing specialised expertise. Recent European evidence indicates that the average time to diagnosis remains close to five years for many people living with a rare disease [6]. In Spain, data from a national rare disease registry show that individuals who experience diagnostic delay report greater psychosocial impact at the time of diagnosis compared with those diagnosed within the first year, highlighting the diagnostic process as a period of particular psychological vulnerability [5, 6].

Beyond diagnosis, living with a RD is associated with a sustained psychological burden. Studies conducted across different rare chronic diseases have identified clinically relevant levels of anxiety and depression, associated not only with diagnostic category but also with perceived symptom burden, illness perceptions, coping strategies, and social support [7]. Consistent evidence indicates that people living with RDs experience higher levels of psychological distress than the general population, including elevated rates of anxiety and depression [8]. Furthermore, recent findings suggest that nearly one quarter of patients (22.9%) meet diagnostic criteria for a mental health disorder attributable to the consequences of their rare disease. These findings are consistent with the 2017 EURORDIS report "*Juggling care and daily life"*, which found that 37% of respondents reported unhappiness or depression "often" or "very often" [9].

The psychosocial impact of RDs extends beyond the affected individual and must be understood from a systemic perspective. Given that approximately 80% of RDs have a genetic origin and many first appear

during childhood, their consequences profoundly affect parents, siblings, and other family members. Furthermore, the burden associated with RDs appears to be distributed unevenly within families. Several studies have reported greater emotional distress and caregiver burden among mothers, who experience higher levels of anxiety, depression, and stress than fathers [10].

More broadly, parents, family members, and informal caregivers often take on complex caregiving responsibilities while coping with uncertainty, fragmented healthcare systems, financial difficulties, and changes in family roles. A systematic review identified increased psychological distress, poorer quality of life, greater caregiver burden, and disruptions in social support networks among parents and family members of people with RDs [11]. Similarly, a scoping review focused on caregivers highlighted challenges related to diagnosis, high care demands, financial costs, and barriers to accessing healthcare as factors that significantly affect their quality of life [12].

Taken together, these findings suggest that the psychosocial needs associated with RDs are not an isolated or temporary problem, but rather a cross-cutting aspect of the illness experience that requires attention throughout the entire care trajectory.

Scientific literature has consistently emphasised the importance of providing psychological support from the early stages of the diagnostic process, ensuring empathetic communication, effective interdisciplinary collaboration, and family-centred care [10, 13]. Consequently, psychological care should be considered an essential component of comprehensive care for RDs, rather than an optional or complementary resource. For the purposes of this study, psychological care refers to assessment, counselling, psychotherapy (including individual, family, and group formats), psychoeducation, and other interventions delivered by mental health professionals to address the emotional, behavioural, social, and adaptation-related needs associated with living with a RD.

Consistent with this perspective, a recent European expert consensus recognised psychosocial vulnerability as a fundamental dimension of RD care and recommended the systematic integration of psychosocial interventions into routine healthcare pathways [14]. Similarly, although United Nations resolutions have urged Member States to promote accessible mental health programs for the entire population [15], psychosocial support is far from being a standard component of routine care for RDs.

Current evidence also suggests that integrating mental health professionals into multidisciplinary teams can improve both patient experience and health outcomes [16]. However, despite the growing recognition of psychosocial vulnerability as a core dimension of RD care, relatively little is known about whether patients and families can access appropriate psychological support in routine clinical practice.

In Spain, available evidence suggests that people affected by RDs and their families experience substantial unmet psychosocial needs, with consequences across social, occupational, and emotional domains [17]. However, much less is known about the extent to which these needs translate into actual access to psychological care. In particular, evidence remains scarce regarding referral to psychological services, barriers to access, funding pathways, and potential inequalities according to age, gender,

diagnosis, or autonomous community of residence. This knowledge gap is especially relevant in a decentralised healthcare system such as Spain's, where the availability of specialised services, referral pathways, and public funding mechanisms may vary substantially across autonomous communities. In the absence of integrated psychological care pathways for rare diseases, access may depend on individual professional awareness, patients' and families' capacity to seek help, financial resources, or support from patient organisations.

The present study aimed to examine access to psychological care among people affected by RDs in Spain, including referral pathways, perceived barriers, cost coverage, satisfaction, adherence, and unmet needs. It also examined potential inequalities according to age, gender, diagnostic groups, and autonomous community of residence.

By investigating psychological care through the lens of access and equity, this study seeks to generate evidence that can inform the development of more integrated and equitable models of RD care for affected individuals and their families.

2-METHODS

2.1 Study design and participants

A national cross-sectional online survey was conducted between December 2023 and October 2024 to assess access to psychological care among individuals affected by RDs in Spain. The study was developed through a collaboration between the FEDER, the Share4Rare digital platform, the Patient Engagement in Research Area of the Sant Joan de Déu (SJD) Research Institute, and the Mental Health Department of SJD Barcelona Children's Hospital.

The survey was specifically developed for this study by a multidisciplinary team including clinical psychologists, patient-engagement researchers, professionals from the FEDER, and patient representatives from RDs organisations. The online survey was disseminated nationwide via FEDER communication channels, patient organisations, newsletters, social media, and the Share4Rare community. Data were collected anonymously.

Two groups of respondents were targeted: (1) adults living with an RD and (2) caregivers of children, adolescents, or dependent adults affected by an RD. Eligible participants were individuals aged 18 years or older residing in Spain who were either patients with an RD or caregivers responding on behalf of an affected person. Responses in which the initial sociodemographic section was not completed were considered invalid and excluded from the analysis. This data curation process was performed by the project manager of Share4Rare through inspection of the responses in the Excel file downloaded from the platform.

The unit of analysis was the patient, regardless of whether the questionnaire was completed directly by the patient or by a caregiver.

The study was approved by the Research Ethics Committee of Fundació Sant Joan de Déu on 14 September 2023, with assigned code PIC-127-17. All participants provided informed consent electronically before starting the questionnaire. Participation was voluntary.

2.2 Measures

The questionnaire consisted of up to 63 questions, including items to assess: (1) access to psychological care, defined as having received any intervention delivered by a mental health professional during the previous 12 months. Psychological care included psychological assessment, counselling, psychotherapy, psychoeducation, and other psychological support interventions; (2) referral and prescription pathways, including whether psychological care had been recommended or prescribed by a healthcare professional and the specialty of the referring professional; (3) barriers to accessing clinical psychology services, including lack of professional recommendation, financial difficulties, logistical barriers, and voluntary refusal; (4) reasons for psychological intervention, including coping with the disease, the impact of diagnosis, management of uncertainty, and other reasons; (5) characteristics of psychological care, including the type of intervention (individual, family, or group therapy), the place of care, waiting time from referral to treatment initiation, recommended and actual session frequency, and treatment adherence; (6) funding pathways, including public coverage, private funding, and financial support provided by patient organizations; (7) patient experience, including perceived usefulness, satisfaction with care, adequacy of session frequency, adherence to treatment, and the extent to which psychological support met participants' needs, and (8) sociodemographic and clinical characteristics, including age, gender, region of residence (autonomous community), educational level, employment status, diagnostic group, and membership to patient organisations.

2.3 Data analysis

Data were analysed using descriptive and inferential statistical methods. Statistical analyses were performed using IBM SPSS Statistics.

Categorical variables are presented as frequencies and percentages, and continuous variables as means and standard deviations or ranges, as appropriate. Percentages were calculated based on available data for each variable; therefore, denominators may vary due to missing responses.

Comparisons between groups were performed using chi-square tests or Fisher's exact tests when expected frequencies were small. Exploratory analyses were conducted according to age group (paediatric: 0–18 years; adult: ≥19 years), gender, diagnostic category, and autonomous community of residence.

To further explore potential inequalities in access to psychological care and public funding, logistic regression models were fitted, and results are presented as odds ratios (ORs) with 95% confidence intervals (CIs). For territorial analyses, Catalonia was used as the reference region because it contributed the largest number of participants, providing the most statistically stable comparison group.

Because participation was voluntary and recruitment occurred primarily through patient organisations and online communities, territorial post-stratification weighting was performed as a sensitivity analysis to assess the potential impact of regional over- or under-representation on aggregate estimates.

Qualitative analyses were based on responses to one open-ended question completed by participants who had received psychological care: “Please indicate which aspects of the therapy have been useful.” A total of 305 responses were analysed. Microsoft Copilot was used to support data organisation and theme generation, while final coding, theme selection, and interpretation were performed by the research team. Themes were identified and organised into three overarching categories: reported benefits, reported shortcomings, and recommendations for improvement. Final theme selection and interpretation were reviewed by the research team.

No correction for multiple testing was applied. Consequently, all subgroup and territorial comparisons should be considered exploratory and hypothesis-generating rather than confirmatory.

Percentages were calculated based on available data for each variable; therefore, denominators may vary due to missing responses.

3-RESULTS

3.1-Sample

A total of 792 valid responses were included in the analysis. Of these, 56.9% (n = 451) were completed directly by adult patients and 43.1% (n = 341) by caregivers of children, adolescents, or adults living with an RD.

Overall, 64.4% (n = 510) patients were adults aged 19 years or older, and 35.6% (n = 282) were children or adolescents aged 0–18 years. Of the participants, 67.6% (n = 535) identified as female and 31.6% (n = 250) identified as male. A substantial proportion of the sample (78.7%; n = 623) reported belonging to a patient organisation. The most frequent diagnostic categories were genetic or mitochondrial diseases (28.4%; n = 225) and neurological or neuromuscular diseases (22.1%; n = 175), although diagnostic categories were not mutually exclusive because participants could select more than one disease group. The most represented regions were Catalonia (22,6%; n = 179), Madrid (19,3%; n = 153), and Andalusia (14.0%; n = 111). Table 1 provides a summary of the full sample.

Table 1
Characteristics of the study sample (N = 792)

Variable	n (%)
Respondent	
Adult patient	451 (56.9)
Caregiver*	341 (43.1)
Patient age	
Paediatric (0–18 years)	282 (35.6)
Ault (≥ 19 years)	510 (64.4)
Patient gender	
Female	535 (67.6)
Male	250 (31.6)
Belongs to a patient organisation	
Yes	623 (78.7)
Not	169 (21.3)
Group or RD**	
Genetic or mitochondrial diseases	225 (28.4)
Neurological or neuromuscular diseases	175 (22.1)
Other diagnostic group	225 (28.4)
No diagnosis	26 (3.3)
Geographical distribution of the sample ***	
Catalonia	179 (22.6)
Madrid	153 (19.3)
Andalusia	111 (14.0)

* The group of caregivers consists mainly of parents or other family members, both for paediatric patients and for adult patients who required assistance in completing the questionnaire.

**Diagnostic group was a multiple-response item; therefore, diagnostic categories are not mutually exclusive.

*** Only the three autonomous communities with the largest number of participants are presented. The supplementary material section contains the data for the remaining autonomous communities

3.2-Access, referral and barriers to psychological care

Overall, 50.3% (n = 398) of participants reported having received psychological care during the previous year. Conversely, 49.7% (n = 394) of participants had not received psychological care during the same period. Figure 1 and Table 2 summarize the overall distribution of access to psychological care, referral pathways, and barriers to care.

Referral data were available for 374 participants who had received psychological care. Of these, 56.1% (n = 210) reported accessing care following a recommendation or referral from a healthcare professional, whereas 43.9% (n = 164) accessed care without a professional recommendation.

Among the 384 participants who provided information on reasons for seeking psychological care, multiple responses were permitted. The most frequently reported reason was adaptation to and coping with the disease (63.3%, n = 243), followed by other psychological or contextual difficulties not directly related to the disease process (38.0%, n = 146), the emotional impact of diagnosis (19.8%, n = 76), and management of uncertainty (16.4%, n = 63).

Among participants who had not received psychological care during the previous year (49.7%, n = 394), data on barriers to access were available for 389 participants. The absence of a professional recommendation or referral was the most frequently reported barrier (79.7%, n = 310), followed by financial barriers (13.9%, n = 54), voluntary refusal of therapy (4.4%, n = 17), scheduling incompatibility (3.3%, n = 13), and logistical or transportation barriers (3.1%, n = 12). Multiple barriers could be reported.

Table 2
Access to psychological care: referral recommendation, reasons and barriers.

Variable	Participants, n/N (%)
Received psychological care during the previous year	
Yes	398/792 (50.3)
Not	394/792 (49.7)
Recommendation/referral by a healthcare professional among psychological care users	
Yes	210/374 (56.1)
Not	164/374 (43.9)
Reasons for receiving psychological care	
Coping/adaptation to disease	243/384 (63.3)
Impact of diagnosis	76/384 (19.8)
Diagnostic uncertainty	63/384 (16.4)
Other psychological/contextual difficulties	146/384 (38.0)
Reported barriers among psychological care non-users	
No professional recommendation/referral	310/389 (79.7)
Financial barriers	54/389 (13.9)
Voluntary refusal	17/389 (4.4)
Scheduling incompatibility	13/389 (3.3)
Logistics/transport	12/389 (3.1)

Notes: n indicates the number of participants who selected each option; N indicates the number of participants with a valid response to the corresponding question. Data on access to psychological care were available for 792 participants. Of the 398 participants who had received psychological care, 374 answered the question on professional recommendation or referral (24 did not answer it) and 384 answered the question on the reasons for receiving this care (14 did not answer it). Of the 394 participants who had not received psychological care, 389 answered the question on barriers to access (5 did not answer it). Participants who did not answer each question were excluded only from the corresponding denominator.

 Multiple responses were allowed. Percentages were calculated among participants who answered each question, rather than among all options selected. Categories are not mutually exclusive, and percentages may sum to more than 100%.

3.3- Characteristics of psychological care

Among the 398 participants who reported receiving psychological care, intervention modalities, public funding, session costs and positive appraisals of care were examined. Percentages were calculated

using the valid responses to each question; denominators therefore varied across outcomes and modalities. Table 3 summarises these characteristics.

Intervention modality

Individual therapy was the most frequently reported modality, selected by 71.6% of respondents with valid data for this question (n = 265). Group therapy was reported by 23.5% (n = 79), and family therapy by 24.6% (n = 81). Participants could report more than one modality, so these categories were not mutually exclusive.

Waiting time

Waiting time from referral or prescription to initiation of psychological care was clinically relevant and ranged from 84 to 93 days across intervention modalities. These data suggest that, even when psychological care was recommended, initiation was commonly delayed by nearly three months.

Treatment adherence

Adherence differed substantially by intervention modality. Among participants receiving individual therapy, 70.1% (n = 169) reported completing all planned or recommended sessions. Completion rates were quite lower for group therapy, with only 45.3% (n = 34) reporting to have completed all sessions, and family therapy with 38.4% (n = 28) of users to complete all sessions.

Public funding and session costs

Among participants who received each modality and provided funding information, publicly funded care was reported by 33.7% of individual therapy recipients (n = 84), 31.6% of group therapy recipients (n = 24), and 24.3% of family therapy recipients (n = 18).

Mean reported session costs were €55.2 (SD 30.4; n = 123) for individual therapy, €42.2 (SD 51.6; n = 34) for group therapy, and €68.5 (SD 52.1; n = 39) for family therapy.

Positive appraisal of care

A positive appraisal, defined as selecting “Agree” or “Strongly agree” on the corresponding item, was reported by 85.8% of respondents receiving individual therapy (n = 205), 58.1% receiving group therapy (n = 43), and 64.4% receiving family therapy (n = 47).

Open-ended responses provided further insight into participants' experiences with psychological care. Participants described psychological support as helpful in accepting the diagnosis, adapting to the disease, managing emotional distress (including anxiety, sadness, fear, and frustration), feeling listened to and understood, improving family relationships, and fostering autonomy and self-awareness.

Although satisfaction with psychological care was generally high, only 33.8% (n = 106) reported that the intervention fully met their needs, whereas 66.2% (n = 206) reported unmet or only partially met needs.

The open-ended responses provided more information about these results. Participants pointed to several factors that hindered access to psychological care, including the insufficient availability of psychological care within the public healthcare system, long waiting lists, infrequent sessions, the high cost of private therapy, limited professional experience with rare diseases, and regional inequalities in access to care.

Table 3
Characteristics of psychological care according to intervention modality

Characteristic	**Individual therapy**	**Group therapy**	**Family therapy**
Use of psychological care	**n/N (%)**	**n/N (%)**	**n/N (%)**
Participants receiving this modality, n/N (%)	265/370 (71.6)	79/336 (23.5)	81/329 (24.6)
Mean waiting time (days)	84	93	86
Treatment adherence			
Completed all sessions, n (%)	169/241 (70.1%)	34/75 (45.3%)	28/73 (38.4%)
Funding pathways and public coverage			
Publicly funded, n/N (%)	84/249 (33.7)	24/76 (31.6)	18/74 (24.3)
Partially or fully funded by patient organisations, n/N (%)□	38/165 (23.0)	18/51 (35.3)	17/56 (30.4)
Mean cost per session (€)	55.23	42.24	68.49
Positive appraisal of care			
Positive evaluation, n/N (%)□	205/239 (85.8)	43/74 (58.1)	47/73 (64.4)

Notes: n indicates the number of participants reporting each characteristic; N denotes the number with a valid response to the corresponding question. Among the 398 psychological care users, modality data were available for 370 participants for individual therapy, 336 for group therapy and 329 for family therapy. Funding, session costs and positive evaluations were assessed among participants who received the corresponding modality and provided valid data for each item. Missing responses were excluded from the denominators. Participants could report more than one modality; categories are therefore not mutually exclusive. Waiting time refers to the interval from referral or prescription to the start of psychological care.

□ Among participants whose therapy was not publicly funded and who provided a valid response to the patient organisation funding question.

□ Positive evaluation was defined as selecting “Agree” or “Strongly agree” on the positive evaluation item.

3.4 Access to psychological care and public coverage according to patient characteristics

Exploratory analyses examined access to psychological care, defined as having received psychological care, and public coverage according to age, gender, diagnostic group and autonomous community of residence. Selected results are summarised in Table 4.

Regarding access to psychological care, adults received psychological care more frequently than paediatric patients. Specifically, 271 out of 510 adults (53.1%) received psychological care, compared with 127 out of 282 (45.0%) children and adolescents.

Of all participants identified as female, 53.3% (n = 286/535) received psychological intervention while 43.6% (n = 109/250) of male participant did. However, the proportion of female vs. male users was that of 71.9% (n = 286/398) female vs. 27.4% (n = 109/398) male users.

Public coverage also differed by gender in exploratory analyses. The proportion of psychological care that was publicly funded was higher among male patients than among female patients (34.55% (n = 101/293) vs. 18.5% (n = 24/130). This pattern should be interpreted carefully because coverage depends not only on patient gender but also on age, diagnosis, type of therapy, care pathway, place of treatment, and regional service organisation.

The territorial analyses also showed variability among the autonomous communities, both in terms of access and public coverage. Compared with Catalonia, participants living in Galicia were more likely to receive psychological care (OR 2.31, 95% CI 1.18–4.74), whereas those residing in the Principado de Asturias Community were less likely to receive publicly funded psychological care (OR 0.43, 95% CI 0.09–1.61). Regarding public coverage of the therapy (not comparing among types of interventions), Castilla-La Mancha (OR = 0.11; 95% CI 0.01–0.61) and Comunidad Valenciana (OR = 0.21; 95% CI 0.08–0.51) showed less probability for public coverage. Galicia (OR = 0.34; 95% CI 0.09–1.01) and Region of Murcia (OR = 2.70; 95% CI 0.96–8.38; p = 0,068) presented tendency to differences with respect to Catalonia, although there was no statistical significancy.

Analyses by diagnostic group suggested descriptive variation in both psychological care access and public coverage across disease categories. However, no statistically significant overall heterogeneity in psychological care access was observed across diagnostic groups.

These results should be interpreted with caution, as they are exploratory analyses. Sample size varied considerably among regions, some estimates were based on a small number of cases, the diagnostic-group variable was a multiple-response variable, and no correction for multiple comparisons was applied.

Table 4
Psychological care access and public coverage according to patient characteristics

Outcome	Characteristic or comparison	Result n/N (%)
Psychological care access		
Age	Adults	271/510 (53.1)
	Children and adolescents	127/282 (45.0)
Gender	Female	286/535 (53.3)
	Male	109/250 (43.6)
Region	Galicia vs Catalonia	OR 2.31 (95% CI 1.18–4.74)
Diagnostic group	Overall comparison across groups	No statistically significant overall heterogeneity observed
Gender distribution among psychological care users		
Gender	Female	286/398 (71.9)
	Male	109/398 (27.4)
Public coverage		
Gender	Male	101/293 (34.55)
	Female	24/130 (18.5)
Region	Principado de Asturias vs Catalonia	OR 0.43 (95% CI 0.09–1.61)
	Castilla-La Mancha vs Catalonia	OR 0.11 (95% CI 0.01–0.61)
	Comunidad Valenciana vs Catalonia	OR 0.21 (95% CI 0.08–0.51)
	Galicia vs Catalonia	OR 0.34 (95% CI 0.09–1.01)
	Region of Murcia vs Catalonia	OR 2.70 (95% CI 0.96–8.38); p = 0.068

Notes: Results summarise the comparisons reported in the accompanying text. Analyses were exploratory; sample sizes varied considerably across regions, some estimates were based on small numbers of cases, and no correction for multiple comparisons was applied. The diagnostic-group variable allowed multiple responses. Catalonia was the reference category for regional comparisons. OR, odds ratio; CI, confidence interval.

4-DISCUSSION

This study examined access to psychological care among people affected by RDs in Spain and identified several relevant findings. First, access to psychological care remained limited, with nearly half of participants reporting that they had not received psychological support during the previous year; the most frequently reported barrier was the lack of professional recommendation or referral. Second, among those who received psychological care, individual therapy was the most common modality and showed the highest levels of adherence and satisfaction, although many participants reported unmet needs despite having received support. Third, public funding was limited across all intervention modalities, and patient organisations played a key role in facilitating access to care and covering associated costs. Finally, exploratory analyses suggested potential inequalities in access and public coverage according to patient age, gender, autonomous community of residence, and diagnostic group.

Rare diseases (RDs) are associated with significant psychological and psychosocial challenges for both patients and their families. Previous research has consistently reported elevated levels of anxiety, depression, uncertainty, caregiver burden, and reduced quality of life among people affected by RDs [7, 8, 11, 12]. However, while the emotional burden and psychosocial needs associated with RDs have been widely documented, considerably less attention has been paid to actual access to psychological care and to the factors that facilitate or hinder such access.

Recent European data reinforce this concern. In the 2025 Rare Barometer survey of 9,897 respondents across 45 European countries, 68% reported at least one mental health difficulty and, notably, while 73% reported needing professional psychological support during the previous six months, only 49% had received it [18]. Although based on a self-selected sample, these findings provide recent European evidence of a substantial gap between psychological support needs and access to professional care in the rare disease community.

In this context, one of the most relevant findings of the present study is the substantial gap in access to psychological care, with nearly one in two participants reporting that they had not received psychological support during the previous year. Particularly noteworthy was the finding that the absence of professional recommendation or referral emerged as the main reported barrier to access rather than voluntary refusal of care. This suggests that unmet psychological needs may be related not only to limitations in the availability of psychological services, but also to insufficient identification of psychosocial difficulties and the absence of systematic referral pathways within routine healthcare. As a result, access to psychological care may depend on factors unrelated to clinical need, creating potential inequalities in care.

This interpretation is consistent with previous studies showing that psychological distress among people living with RDs and their families often remains under-recognised despite its well-established association with poorer quality of life, lower treatment adherence, less favourable health behaviours, and worse physical health outcomes [17, 19]. In this context, several authors have advocated the systematic integration of psychosocial assessment, routine psychological screening, and structured referral pathways within multidisciplinary models of rare disease care.

Such approaches appear particularly relevant during periods of heightened psychological vulnerability, including diagnostic suspicion, diagnosis disclosure, and the early stages of adaptation to the disease [10, 13].

Another important finding relates to the funding and organisation of psychological care. Public funding was limited across all intervention modalities, meaning that access to psychological care frequently depended on private payment or financial support provided by patient organisations. These findings are consistent with recent evidence from Catalonia. In a 2026 study of 291 people affected by RDs and their families, Calmaestra-Carrillo et al. identified substantial unmet healthcare, psychological and social support needs, with participants perceiving existing public resources as insufficient to meet their needs [20]. This result is particularly relevant in the context of RDs, where patients and families often face substantial economic and social burdens, including healthcare-related expenses, travel costs, reduced employment opportunities, and the long-term demands associated with caregiving [12, 21]. Consequently, the need to self-finance psychological care may constitute an additional barrier to accessing appropriate psychological support.

Patient organisations emerged as key stakeholders, providing not only information and peer support but also facilitating access to psychological care and contributing to its funding. This finding is consistent with previous studies highlighting their essential role in addressing unmet psychosocial needs among people living with RDs [19]. Nevertheless, while patient organisations represent a valuable component of the RD ecosystem, access to psychological care should not depend on their availability, geographical reach, or financial capacity. Such reliance on external resources raises important concerns regarding equity in access to psychological care and the long-term sustainability of psychosocial care provision.

The reasons for referral identified in this study also provide important insights into the timing and focus of psychological care. Adaptation to the disease, the emotional impact of diagnosis, and the management of uncertainty were the most frequently reported reasons for seeking psychological support. These results are broadly consistent with previous research identifying diagnostic uncertainty, prolonged diagnostic journeys, and diagnosis disclosure as key periods of heightened psychological vulnerability for both patients and their families [3, 6, 10]. Furthermore, individuals who experience longer diagnostic delays report greater psychosocial impact than those who receive an earlier diagnosis [6]. Together, these findings support the integration of psychological assessment and support from the earliest stages of the diagnostic process, rather than waiting until psychological difficulties become clinically established.

These findings reinforce the view that adaptation to a rare disease is not a single event, but a dynamic process that requires continuous psychological adjustment over time. Consequently, psychological care should not be limited to periods of acute distress or crisis but should be integrated as a routine component of multidisciplinary rare disease care pathways. Systematic psychosocial assessment and longitudinal monitoring may be particularly important during key stages of the disease trajectory, including diagnostic suspicion, diagnosis disclosure, disease progression, healthcare transitions, and

periods of increased family burden. Furthermore, uncertainty may persist or recur throughout the course of the disease and represent a substantial source of distress for patients and their families.

Identifying emotional difficulties early and providing timely psychological support may facilitate adaptation, strengthen coping resources, and help reduce the long-term psychosocial impact of living with a rare disease.

Among participants who received psychological care, individual therapy was the most frequently used intervention modality and showed the highest levels of adherence and satisfaction. However, an important discrepancy emerged between participants' positive evaluation of psychological care and their perception that their psychological needs had been fully met. Although individual therapy was generally well rated, only a minority of respondents considered that the support they received adequately addressed their needs. These findings suggest that satisfaction with psychological care does not necessarily reflect the adequacy of its intensity, frequency, duration, or degree of specialisation.

In addition, mean waiting times of approximately three months across intervention modalities indicate that delays may persist even after referral to psychological care. Qualitative responses further highlighted limited availability of services, low session frequency, financial barriers, and limited professional expertise in rare diseases as persistent obstacles to receiving appropriate psychological support. Similar findings have been reported in studies, where patients and families described ongoing difficulties in accessing specialised psychological services despite the considerable emotional burden associated with RDs [19, 21, 22]. Together, these findings suggest that improving access requires not only increasing referral to psychological services but also ensuring timely, sufficiently intensive, and rare disease-informed psychological care integrated within multidisciplinary care pathways.

Exploratory analyses also suggested potential inequalities in access to and public coverage of psychological care according to age, gender, autonomous community of residence, and diagnostic groups. Although these findings should be interpreted cautiously, they suggest that access to psychological care may be influenced not only by patients' psychological needs but also by differences in help-seeking behaviour, professional detection, referral practices, clinical pathways, and local service organisation. The higher use of psychological care observed among adults and female patients is consistent with broader evidence showing greater use of mental health services among these groups. In contrast, the higher levels of public coverage observed among male patients should be interpreted with caution, as they may reflect differences in diagnosis, treatment modality, referral pathways, or regional service organisation rather than gender itself.

The observed territorial variation is particularly relevant in the context of Spain's decentralised healthcare system, where access to psychological care may vary according to local service availability, funding arrangements, professional awareness, and the extent to which psychosocial support is integrated into RD care pathways. These findings are consistent with previous studies describing unmet psychosocial needs and variability in access to specialised services among people living with RDs [17, 19]. Together, they reinforce recent calls for integrated, multidisciplinary, and family-centred models of RD care in

which psychosocial assessment and psychological support are embedded as core components of routine clinical care [13].

The findings have several implications for clinical practice and the organisation of services: psychological care should be integrated as a routine component of multidisciplinary care for rare diseases, rather than being reserved for periods of acute psychological distress; systematic psychosocial assessment and longitudinal follow-up can facilitate the early identification of emotional difficulties, especially during key stages of the disease course, maintaining a form of active “monitoring” throughout the patient’s journey; at the same time, clear referral pathways and streamlined coordination systems must be established between rare disease services and psychological care to ensure timely access, reduce variability in referral practices, and minimize reliance on the individual initiative of patients, families, or healthcare professionals.

Recent evidence from Catalonia similarly highlights the relevance of generating coordinated, person- and family-centered models that address health, psychological, social, educational, and employment needs throughout the rare disease journey [20]. Overall, our findings support the integration of psychological care as a fundamental component of rare disease care. Ensuring timely, specialized, and equitable access to this care is essential for advancing toward more person-centered healthcare systems.

5-CONCLUSIONS

Despite growing recognition of the psychological impact of RDs, access to psychological care remains insufficiently integrated into routine rare disease services. This study, one of the largest national investigations conducted to date on this topic, identifies substantial gaps in access to care, with nearly half of participants reporting that they had not received psychological support during the previous year. The absence of professional recommendation or referral emerged as the principal barrier to access, while limited public coverage and reliance on patient organisations highlighted important challenges related to equity and sustainability.

Beyond documenting unmet needs, these findings provide novel evidence regarding access pathways, funding mechanisms, and the perceived adequacy of psychological care among people affected by rare diseases. Notably, although satisfaction with psychological care was generally high, many participants reported that the support they received did not fully meet their needs, underscoring the importance of ensuring not only access to care but also its adequacy, timeliness, and responsiveness to the specific psychosocial challenges associated with RDs. Taken together, the results support the integration of systematic psychological assessment, psychosocial monitoring, structured referral pathways, and equitable access to specialised psychological care within comprehensive RD services.

Addressing these gaps may contribute not only to improved psychological well-being, but also to better long-term adaptation and quality of life for patients and families living with rare diseases.

Declarations

Declarations

Ethics approval and consent to participate

Consent for publication

Not applicable.

Competing interest

The authors of this article declare that they have no conflicts of interest that could affect the results or interpretations presented in this study.

Funding

This study was supported by the Patient Engagement in Research Area at Institut de Recerca Sant Joan de Déu.

Author Contribution

LRG was responsible for the conception and design of the paper. All authors were involved in the acquisition, analysis, and/or interpretation of the data captured in the survey. LRG drafted the paper and subsequent re-drafts. All authors reviewed the manuscript and approved the final version.

Acknowledgement

The authors would like to express their sincere gratitude to the patient organisations that supported the dissemination of the survey and to all the patients who generously contributed their data. Their contributions were essential to the successful conduct of this research.

Data Availability

The datasets used and analysed during the current study are available from the corresponding author on reasonable request.

References

1. Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020;28(2):165–73. https://doi.org/10.1038/s41431-019-0508-0.
2. European Commission. Rare diseases. Available from: https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/rare-diseases_en
3. Faye F, Crocione C, Anido de Peña R, Bellagambi S, Escati Peñaloza L, Hunter A, et al. Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. Eur J Hum Genet. 2024;32(9):1116–26. https://doi.org/10.1038/s41431-024-01611-8.
4. Benito-Lozano J, López-Villalba B, Arias-Merino G, de la Posada M, Alonso-Ferreira V. Diagnostic delay in rare diseases: data from the Spanish Rare Diseases Patient Registry. Orphanet J Rare Dis. 2022;17:418. https://doi.org/10.1186/s13023-022-02594-6.
5. Ancochea A, Aparicio A, Ruiz B, Arias P, Albacete L, Tomé M, et al. Estudio sobre situación de Necesidades Sociosanitarias de las Personas con Enfermedades Raras en España. Estudio ENSERio. Madrid: FEDER; 2017.
6. Benito-Lozano J, Arias-Merino G, Gómez-Martínez M, Arconada-López B, Ruiz-García B, de la Posada M, et al. Psychosocial impact at the time of a rare disease diagnosis. PLoS ONE. 2023;18(7):e0288875. https://doi.org/10.1371/journal.pone.0288875.
7. Uhlenbusch N, Löwe B, Härter M, Schramm C, Weiler-Normann C, Depping MK. Depression and anxiety in patients with different rare chronic diseases: a cross-sectional study. PLoS ONE. 2019;14(2):e0211343. https://doi.org/10.1371/journal.pone.0211343.
8. Uhlenbusch N, Swaydan J, Höller A, Löwe B, Depping MK. Affective and anxiety disorders in patients with different rare chronic diseases: a systematic review and meta-analysis. Psychol Med. 2021;51(16):2731–41. https://doi.org/10.1017/S0033291721003792.
9. Courbier S, Berjonneau E. Juggling care and daily life: the balancing act of the rare disease community. Paris: EURORDIS-Rare Diseases Europe; 2017. Available from: https://www.eurordis.org/publications/juggling-care-and-daily-life-the-balancing-act-of-the-rare-disease-community/. Accessed 4 Jul 2024.
10. Kenny T, Stone J. Psychological support at diagnosis of a rare disease: a review of the literature. Dublin: Rare Disease Research Partners; 2022. Available from:https://www.infocoponline.es/pdf/220202-Literature-Review-Report.pdf
11. Atkins JC, Padgett CR. Living with a rare disease: psychosocial impacts for parents and family members-a systematic review. J Child Fam Stud. 2024;33(2):617–36. https://doi.org/10.1007/s10826-024-02790-6.
12. Černe T, Kragelj LZ, Turk E, Rotar Pavlič D. Experiences of quality of life and access to health services among rare disease caregivers: a scoping review. Orphanet J Rare Dis. 2024;19(1):319. https://doi.org/10.1186/s13023-024-03327-2.

13. Smits RM, Vissers E, te Pas R, Roebbers N, Feitz WFJ, van Rooij IALM, et al. Common needs in uncommon conditions: a qualitative study to explore the need for care in paediatric patients with rare diseases. Orphanet J Rare Dis. 2022;17:153. https://doi.org/10.1186/s13023-022-02305-w.
14. Smits RM, Aerts A, Angelov T, Bentele M, de Blaauw I, Dellenmark-Blom M, et al. Addressing psychosocial vulnerability in rare diseases: a call to action from a European expert consensus study. Orphanet J Rare Dis. 2025;20(1):543. https://doi.org/10.1186/s13023-025-04017-3.
15. United Nations General Assembly. Addressing the challenges of persons living with a rare disease and their families. Resolution adopted by the General Assembly on 16 December 2021. A/RES/76/132. New York: United Nations. 2021. Available from: https://documents.un.org/doc/undoc/gen/n21/403/90/pdf/n2140390.pdf. Accessed 4 Jul 2024.
16. Hebestreit H, Lapstich AM, Brandstetter L, Krauth C, Deckert J, Haas K, et al. Effect of the addition of a mental health specialist for evaluation of undiagnosed patients in centres for rare diseases (ZSE-DUO): a prospective, controlled trial with a two-phase cohort design. EClinicalMedicine. 2023;65:102260. https://doi.org/10.1016/j.eclinm.2023.102260.
17. Gimenez-Lozano C, Páramo-Rodríguez L, Cavero-Carbonell C, Corpas-Burgos F, López-Maside A, Guardiola-Vilarroig S, Zurriaga O. Rare diseases: needs and impact for patients and families: a cross-sectional study in the Valencian Region, Spain. Int J Environ Res Public Health. 2022;19(16):10366. https://doi.org/10.3390/ijerph191610366.
18. Dubief J, Faye F. Improve Our Mental Health! Key findings from a Rare Barometer survey on the mental health of people with rare diseases. EURORDIS-Rare Diseases Europe; 2026. 10.70790/MHFS2026.
19. Spencer-Tansley R, Meade N, Ali F, Hunter A, et al. Mental health care for rare disease in the UK: recommendations from a quantitative survey and multi-stakeholder workshop. BMC Health Serv Res. 2022;22:759. https://doi.org/10.1186/s12913-022-08060-9.
20. Calmaestra-Carrillo E, Vernet R, Torrent-Farnell J, Arbiol-Rodríguez I, Cardona Cardona À, Grau-Sánchez J, et al. Health care, social, educational, and employment needs of individuals affected by rare diseases and their families in Catalonia, Spain. Orphanet J Rare Dis. 2026;21:132. 10.1186/s13023-026-04281-x.
21. Domaradzki J, Jabkowski P, Walkowiak D. Rare disease, common struggles: quality of life, caregiver burden and financial wellbeing of family caregivers in Poland. Sci Rep. 2025;15:22678. https://doi.org/10.1038/s41598-025-08866-7.
22. Nerney D, O'Malley E, Kenny C, Ward A, Sweeney G, McGrath V, Egan L, Treacy E. Psychological supports for people living with a rare disease in Ireland: an online survey-based study. Ir J Med Sci. 2025;194(2):421–30. https://doi.org/10.1007/s11845-025-03902-x.

Figures

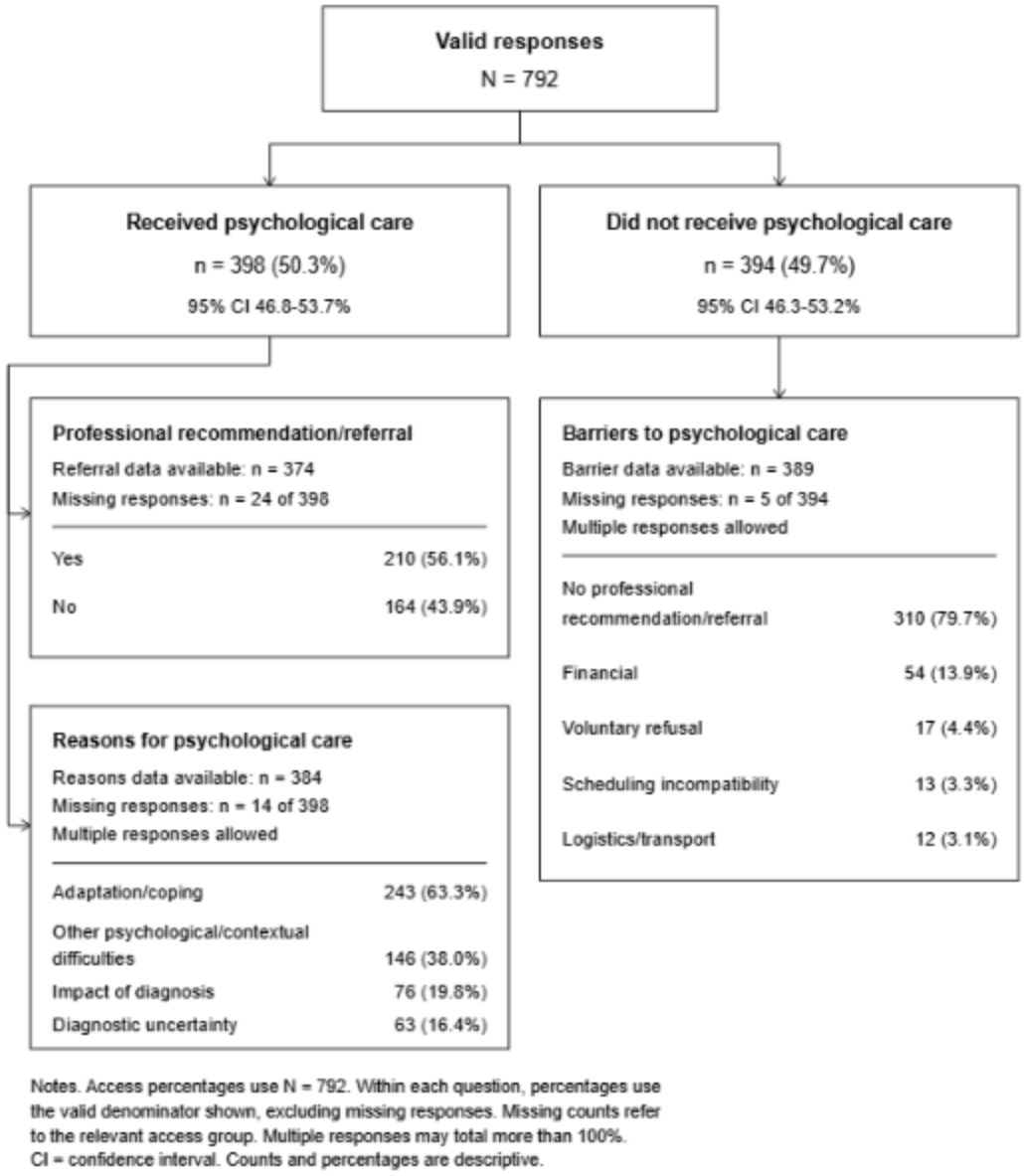

Figure 1

Flow diagram of psychological care access among people affected by RDs in Spain. The diagram shows the total number of valid responses, the proportion of participants who did and did not receive psychological care during the previous year, the main intervention modalities among users, and the main barriers reported by those who did not receive care.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- Supplementarymaterials.docx