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# When Diagnosis Comes Too Late: Psychological Impact of Rare Diseases in Children
# Newborn Screening for Rare Diseases in Children

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**KEYWORDS**
Rare diseases
Newborn screening
Diagnostic delay
Psychological impact

**ABSTRACT**

Introduction. Late diagnosis of rare diseases in childhood is a significant public health issue with important clinical and psychosocial consequences. The absence of early detection prolongs diagnostic uncertainty and can have a significant psychological impact on children and their families. Newborn screening is a key tool for early detection and may act as a protective factor against the emotional distress associated with diagnostic delay. The aim of this study was to analyse the psychological impact of late diagnosis of rare diseases in childhood and the protective role of newborn screening. Method. A systematic review was conducted following the PRISMA recommendations. Searches were carried out in MEDLINE, Psychology and Behavioral Sciences Collection, APA PsycInfo, Scopus and OpenAIRE. Empirical studies published in Spanish or English during the previous five years were included. Methodological quality was assessed using the AXIS and CRF-QS tools. Results. Twenty-four studies were included. Late diagnosis was associated with higher levels of stress, anxiety and parental emotional burden, as well as poorer family quality of life. Diagnostic uncertainty and difficulties in accessing healthcare and psychological resources were relevant factors. Conclusions. Late diagnosis has a significant psychological impact, whereas newborn screening acts as a protective factor by reducing uncertainty and facilitating family adaptation.

**PALABRAS CLAVE**
Enfermedades raras
Cribado neonatal
Retraso diagnóstico
Impacto psicológico

**RESUMEN**

Introducción. El diagnóstico tardío de enfermedades raras en la infancia constituye un problema relevante de salud pública con importantes repercusiones clínicas y psicosociales. La ausencia de detección precoz prolonga la incertidumbre diagnóstica y puede generar un impacto psicológico significativo en los menores y sus familias. El cribado neonatal representa una herramienta clave para la detección temprana y puede actuar como factor protector frente al malestar emocional asociado al retraso diagnóstico. El objetivo de este estudio fue analizar el impacto psicológico del diagnóstico tardío de enfermedades raras en la infancia y el papel protector del cribado neonatal. Método. Se realizó una revisión sistemática siguiendo las recomendaciones PRISMA. La búsqueda se llevó a cabo en MEDLINE, Psychology and Behavioral Sciences Collection, APA PsycInfo, Scopus y OpenAIRE. Se incluyeron estudios empíricos publicados en español o inglés durante los últimos cinco años. La calidad metodológica se evaluó mediante las herramientas AXIS y CRF-QS. Resultados. Se incluyeron 24 estudios. El diagnóstico tardío se asoció con mayores niveles de estrés, ansiedad y sobrecarga emocional parental, así como con menor calidad de vida familiar. La incertidumbre diagnóstica y las dificultades de acceso a recursos sanitarios y psicológicos fueron factores relevantes. Conclusiones. El diagnóstico tardío tiene un impacto psicológico significativo, mientras que el cribado neonatal actúa como factor protector al reducir la incertidumbre y facilitar la adaptación familiar.

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## 1. Introduction

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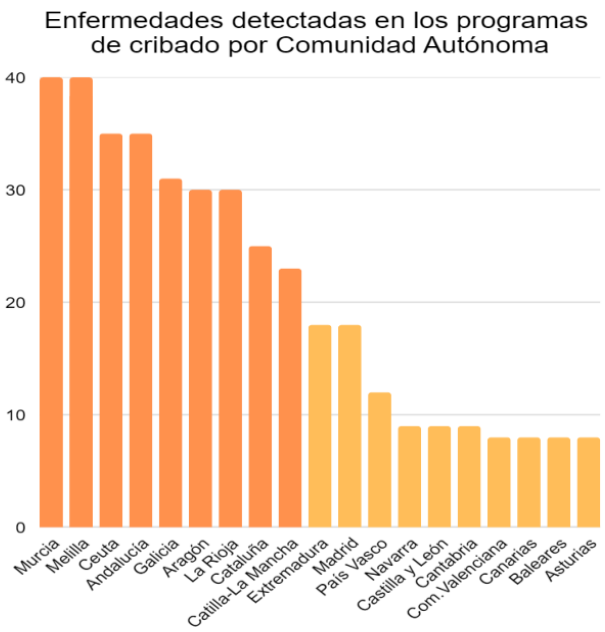
The birth of a child is one of the most significant events in family life, generally accompanied by positive expectations and plans for the future. For some families, however, this moment may mark the beginning of a complex and uncertain healthcare journey, particularly when a rare disease (RD) is present but is not diagnosed at an early stage. In this context, newborn screening programmes represent an essential public health tool, enabling the early detection of potentially serious congenital diseases and facilitating timely interventions that can significantly alter the subsequent prognosis (Ministerio de Sanidad, 2010; Organización Mundial de la Salud [OMS], 2024). Despite its importance, the implementation of newborn screening in Spain varies considerably across autonomous communities. While some regions screen for more than thirty conditions, others are limited to the diseases included in the Common Services Portfolio, meaning that opportunities for early diagnosis depend largely on place of birth (Observatorio Legislativo de Enfermedades Raras y Medicamentos Huérfanos, 2022). These territorial inequalities are reflected in the number of diseases detected through newborn screening (see Figure 1) and have particularly significant consequences for neurodegenerative or metabolic disorders, such as metachromatic leukodystrophy, in which early detection allows the initiation of treatments that can modify the clinical course, whereas late diagnosis entails irreversible neurological loss.

**Figure 1.**

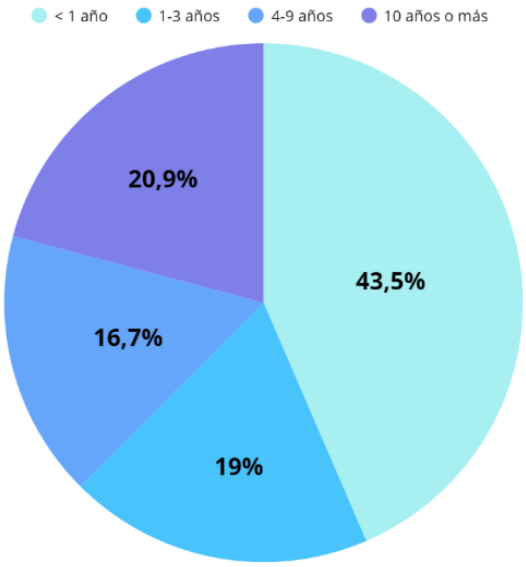



Diseases detected through screening programmes by autonomous community.
Note: Adapted from *Herramientas para el diagnóstico de las enfermedades raras: El cribado neonatal*, by Observatorio Legislativo de Enfermedades Raras y Medicamentos Huérfanos. (2022). https://centroestudiospoliticaspublicas.com/wp-content/uploads/2021/03/220630_Informe-cribados-neonatales.pdf

Newborn screening is both a diagnostic tool and a fundamental pillar of health equity. From a public health perspective, it helps prevent disability, reduce the burden on healthcare services and promote healthy child development (Ministerio de Sanidad, 2010; OMS, 2024). The benefits of early detection are also reflected in health-economic terms, through reductions in the burden of disease and increases in quality-adjusted life years (QALYs) (Ministerio de Sanidad, 2010). Nevertheless, the screening process can also generate uncertainty and emotional distress among families, particularly in the case of positive or uncertain results, highlighting the need for multidisciplinary care that includes psychological support and appropriate clinical communication (Domaradzki et al., 2025; Emerson et al., 2018). One of the main problems associated with RDs is diagnostic delay (DD), defined as the time elapsed between the onset of the first symptoms and diagnostic confirmation. DD is a common phenomenon affecting a significant proportion of patients

with RDs and is associated with important clinical and psychosocial consequences. In Spain, the average time to diagnosis is approximately six years, with more than half of patients waiting at least one year and around one fifth waiting more than ten years to obtain a definitive diagnosis (Benito-Lozano et al., 2022). The distribution of diagnostic times can be seen in Figure 2, which shows that, although a substantial proportion of diagnoses are made during the first year of life, a significant percentage of patients experience prolonged delays.

**Figure 2.**




Average time to diagnosis in Spain.

Note: Adapted from *Diagnostic delay in rare diseases: Data from the Spanish rare diseases patient registry*, by Benito-Lozano, J., López-Villalba, B., Arias-Merino, G., Posada de la Paz, M., & Alonso-Ferreira, V. (2022). https://doi.org/10.1186/s13023-022-02530-3

Although diagnostic times vary according to the condition, the general pattern indicates that diagnostic delays hinder early intervention and exacerbate clinical and functional consequences. Beyond their medical implications, diagnostic delays have a significant impact on the psychological and social well-being of families. Prolonged periods of uncertainty often involve multiple medical consultations, active information-seeking and difficulties in accessing specialised resources, generating high levels of stress and emotional exhaustion (Pelentsov et al., 2015). Several studies have shown that this situation is associated with higher levels of anxiety, frustration and psychological vulnerability among caregivers, as well as difficulties in family functioning, social isolation and repercussions for employment and financial stability (Atkins & Padgett, 2024; Rafferty & Beck, 2020).

The psychological impact of DD can be understood through the biopsychosocial model proposed by Engel (1977), which views illness as the result of interactions between biological, psychological and social factors. From this perspective, a delay in neonatal diagnosis affects not only the child's health status but also the family's emotional and relational balance, with effects extending into the educational and social spheres. Perinatal psychology has likewise highlighted the importance of emotional support during pregnancy and the postpartum period, particularly in the presence of complex medical conditions or uncertain diagnoses (Rodríguez-Muñoz et al., 2023). In this regard, early detection promotes informed decision-making, facilitates family reorganisation and reduces the emotional burden associated with diagnostic uncertainty (Pelentsov et al., 2015; Atkins & Padgett, 2024).

The emotional process experienced by families in the face of a late diagnosis bears similarities to the reactions described in classical models of grief. Kübler-Ross's (1969) model describes five common emotional responses to highly stressful situations —denial, anger, bargaining, depression and acceptance— which may also arise during the diagnostic process for a chronic or rare disease. These responses should not be understood as linear stages, but rather as a useful conceptual framework for understanding emotional adaptation to situations of considerable uncertainty. From this perspective, ongoing psychosocial support is a fundamental element in facilitating family adaptation during the diagnostic process and following confirmation of the disease (Rafferty & Beck, 2020). The availability of healthcare and social resources, access to early intervention programmes and perceived social support act as protective factors against the emotional impact of DD (Atkins & Padgett, 2024; Rafferty & Beck, 2020).

Accordingly, the overall aim of this study is to analyse, on the basis of the available scientific evidence, the psychological impact of late diagnosis of rare diseases in childhood, with particular attention to newborn screening as a protective factor and to its implications for health psychology practice. The specific objectives are as follows: (1) To analyse the role of newborn screening as a public health strategy in the prevention of rare diseases and in reducing diagnostic delay in childhood. (2) To examine the consequences of late diagnosis for child development, with particular regard to its psychological and developmental implications. (3) To analyse the impact of late diagnosis on the family, particularly in terms of parental stress, family reorganisation and psychological well-being. (4) To identify variables that moderate the psychological impact associated with diagnostic delay, such as social support, access to healthcare and psychosocial resources, and the availability of early intervention programmes. (5) To describe the role of the health psychologist in the diagnostic and care process, with particular attention to psychosocial intervention with the child and their family.

## 2. Methodology

Study Design

To address the stated objectives, a systematic literature review was conducted following the recommendations of the PRISMA statement (Preferred Reporting Items for Systematic Reviews and Meta-Analyses; Page et al., 2021). This methodological approach enabled a structured synthesis of the available scientific evidence on the psychological impact of late diagnosis of rare diseases in childhood and the role of newborn screening as a protective factor. Only primary empirical studies were included in order to avoid duplication of evidence derived from systematic reviews or meta-analyses and to ensure that the synthesis was based on original data. Studies published within the previous five years were also selected to ensure the currency and relevance of the evidence analysed.

Literature Search Strategy

The literature search was conducted in December 2025 in the following electronic databases: MEDLINE, Psychology and Behavioral Sciences Collection, APA PsycInfo, Scopus and OpenAIRE. A consistent procedure was applied across all databases.

The search strategy was based on combinations of terms related to the topic of the study using Boolean operators (AND/OR), following the general structure: ("rare diseases" OR "diagnostic delay" OR "delayed diagnosis") AND ("parental stress" OR "caregiver burden" OR "psychological impact" OR "psychosocial impact" OR "family functioning").

Inclusion and Exclusion Criteria

The inclusion and exclusion criteria were defined prior to the selection process in order to ensure the methodological consistency of the review. The PICOS framework (Population, Intervention, Comparison, Outcomes and Study design), widely used in systematic reviews to structure the key elements of a study, was employed to establish these criteria. Based on this framework, criteria were defined in relation to the population of interest, the type of exposure examined, possible comparisons, the outcomes considered relevant and the design of the included studies (see Table 1).

**Table 1**. *Study selection criteria according to PICOS.*

| | **Inclusion criteria** | **Exclusion criteria** |
|---|---|---|
| **Population (P)** | Studies focusing on children with RD and their families, particularly in the early stages of development. | Studies focusing exclusively on adults or older people; samples that do not include children or families. |
| **Intervention (I)** | Newborn screening, early diagnosis or diagnostic delay in RDs. | Studies that do not address newborn screening or the diagnostic process. |
| **Comparison (C)** | Comparison between early diagnosis versus diagnosis after the onset of symptoms, or limited screening versus expanded screening. | Studies that do not allow the timing of diagnosis to be differentiated or different types of screening to be compared. |
| **Outcomes (O)** | Psychological impact of late diagnosis, parental stress, caregiver burden, family functioning and well-being, age at the time of diagnosis, access to early intervention and consequences for child development. | Studies without an assessment of outcomes or with non-measurable outcomes. |
| **Study Design (S)** | Empirical studies published in academic journals. | Systematic reviews, meta-analyses, purely biomedical studies or non-academic documents. |

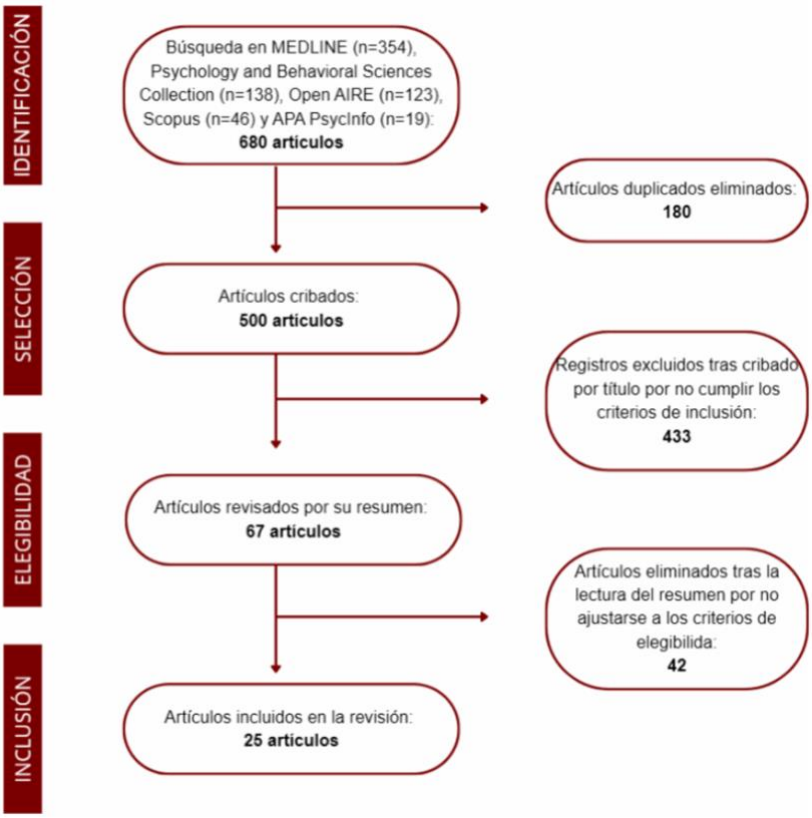
Source: Authors' own elaboration.

Study Selection Process

The study selection process was conducted in three phases, following the PRISMA framework (see Figure 3). (1) Identification of records: Following searches of the selected databases and application of the established filters, 680 records were identified. After duplicates were removed (n = 180), a total of 500 unique studies remained and proceeded to the initial screening stage. (2) Initial screening: In the second phase, titles were screened to exclude studies that did not meet the predefined inclusion criteria. As a result of this process, 67 studies were selected for abstract review. (3) Eligibility assessment: The abstracts were subsequently reviewed to confirm the thematic and methodological suitability of the studies. At this stage, studies focusing on adults with rare diseases (n = 15), studies with a predominantly biomedical focus (n = 12), studies for which the full text was unavailable (n = 5), systematic reviews (n = 4), studies focusing on instrument validation (n = 3) and studies unrelated to rare diseases (n = 2) were excluded. As a result of this process, 25 studies were selected for full-text review. Following assessment of methodological

quality, one study was excluded for failing to meet the established minimum criteria, resulting in a final total of 24 studies included in the main synthesis.

**Figure 3.**




PRISMA flow chart.

Methodological Quality Assessment

The methodological quality of the included studies was systematically assessed in order to evaluate methodological adequacy, reporting transparency and the internal consistency of the evidence analysed. Specific tools were used according to study design: (1) AXIS (Appraisal Tool for Cross-Sectional Studies) for quantitative cross-sectional studies (Downes et al., 2016). The AXIS tool assesses cross-sectional studies using 20 items related to study design, sampling and data analysis. Methodological quality was expressed as the percentage of criteria met, with studies classified as follows: High quality (≥ 80%), Moderate quality (50–79%), Low quality (< 50%). (2) CRF-QS (Critical Review Form – Qualitative Studies) for qualitative studies (Letts et al., 2007). The CRF-QS assesses qualitative studies using eight criteria comprising a total of 19 items. Methodological quality was classified as follows: Poor (≤ 11 points), Acceptable (12–13 points), Good (14–15 points), Very good (16–17 points), Excellent (≥ 18 points).
During this process, one study was excluded following full-text review after receiving a score of 9/19 on the CRF-QS, indicating insufficient methodological rigour and limiting the interpretation of its findings. In addition, one study with a single-group pre–post quasi-experimental design (Rovira-Moreno et al., 2020) fell outside the scope of the standardised assessment tools used. This study was included because of its thematic relevance to the objectives of the review, although its findings are interpreted with caution owing to the absence of assessment using standardised methodological tools.

## 3. Results

Results of the Methodological Quality Assessment

Of the 18 cross-sectional studies assessed using AXIS, 16 showed high methodological quality (≥ 80%), with clearly defined objectives, consistency between the study design and research question, use of validated instruments and appropriate statistical analyses. Only two studies were rated as being of moderate quality: Domaradzki and Walkowiak (2024b) and González et al. (2024), with scores of 75% and 70%, respectively, mainly due to limitations in sampling, lack of justification for sample size and insufficient control of bias; in the former case, ad hoc questionnaires without validation were also used.

Across the studies, the most frequent limitations concerned items 5 and 6, relating to the method of sample selection and justification of sample size. Convenience samples were commonly used, often recruited through patient associations or online platforms (Carbyn et al., 2025; Chiarotti et al., 2023; Koehly et al., 2023; Valcárcel-Nazco et al., 2022), and statistical power calculations were frequently absent, even in studies with large samples (Boettcher et al., 2021; Jobling et al., 2025; Wu et al., 2020). These limitations should be interpreted within the context of rare disease research, which is characterised by the low prevalence and clinical and diagnostic heterogeneity of the populations studied.

Specific limitations were also identified in the reporting of attrition and non-response (item 12) in Domaradzki and Walkowiak (2024a, 2024b), as well as weaknesses in the control of confounding factors (item 10) in studies based on bivariate analyses that did not allow simultaneous adjustment for other relevant variables (Boettcher et al., 2025; Lim et al., 2025; Maltseva et al., 2023), although in most cases these issues were acknowledged in the discussion.

Overall, the AXIS assessment indicates a high overall level of methodological quality among the quantitative studies included, although the findings should be interpreted and generalised with caution, taking into account the inherent challenges of research involving populations affected by rare diseases.

With regard to the qualitative studies, assessment using the CRF-QS showed overall methodological quality ranging from good to very good. Three studies achieved very good quality: Hager et al. (2024) (16/19; 84.2%), Hoffmann et al. (2025) (17/19; 89.5%) and Witt et al. (2023) (17/19; 89.5%), characterised by clearly formulated objectives, congruent qualitative designs and well-described analytical procedures. Smits et al. (2022) and Aldiss et al. (2021), meanwhile, were rated as good quality (15/19; 78.9%), mainly due to limitations in the justification of sample size and insufficiently explicit reflection on potential biases (see Table 2).

**Table 2.**

*Key findings of the methodological assessment using CRF-QS.*

| Study | Population | Quality | Main limitations |
|---|---|---|---|
| Hager et al. (2024) | 16/19 | Very good | Selection bias due to self-selected online recruitment and lack of power calculation; possible inclusion of items without independent validation. |
| Smits et al. (2022) | 15/19 | Good | Small, single-centre sample, with limited applicability. Lack of reflexivity and formal inter-rater reliability assessment. |
| Hoffmann et al. (2025) | 17/19 | Very good | Limited applicability due to a highly selected and homogeneous sample. |
| Witt et al. (2023) | 17/19 | Very good | The researcher's reflexivity is underdeveloped. Transferability is limited by diagnostic heterogeneity and the over-representation of mothers. |

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|---|---|---|---|
| Aldiss et al. (2021) | 15/19 | Good | Small, homogeneous sample (mothers only), with self-selection and limited reflexivity. |

Source: Authors' own elaboration.

Across the qualitative studies, the most frequent limitation was the possible presence of bias (criterion 4), associated with the use of purposive and highly selected samples (Aldiss et al., 2021; Hager et al., 2024; Hoffmann et al., 2025; Smits et al., 2022; Witt et al., 2023). Recurrent weaknesses were also observed in the justification of sample size (criterion 6) (Aldiss et al., 2021; Hager et al., 2024; Smits et al., 2022; Witt et al., 2023) and, in some studies, in the description of the methodological validity of the interview guides (criterion 9), owing to the absence of specific discussion of their development, piloting or appropriateness (Aldiss et al., 2021; Hager et al., 2024; Smits et al., 2022).

Lastly, the study by Rovira-Moreno et al. (2020), which used a single-group pre–post quasi-experimental design, could not be assessed using AXIS or the CRF-QS. Its methodological quality was therefore evaluated through a narrative critical appraisal. The main limitations identified included the absence of a control group, a small sample size and limited generalisability. Nevertheless, the study also showed important strengths —a clearly defined objective, a detailed description of the intervention and the use of validated instruments— and was therefore retained in the synthesis because of its high thematic relevance, with its findings interpreted cautiously.

Results of the Study Selection

Based on the selected studies (see Table 3), the study characteristics were described according to the following areas: the psychological impact of diagnosis; uncertainty and lack of information during the diagnostic process; parental role overload and family tensions in the context of rare diseases; and relationships with the healthcare system, social support and peer networks.

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**Table 3.**

*Summary of objectives, methodology and main conclusions of the included studies.*

| Title and year of publication | Objectives | Participants | Research design | Key findings |
|---|---|---|---|---|
| Aldiss et al. (2021) | To explore the needs and experiences of parents caring for a child with an undiagnosed genetic condition. | 14 mothers of children with an undiagnosed genetic condition from the UK. | Exploratory qualitative study. | High levels of stress and uncertainty due to the lack of a diagnosis; lack of coordination of care and unmet emotional needs. |
| Benito-Lozano et al. (2023) | To analyse the psychological impact of a rare disease diagnosis in relation to diagnostic delay. | 524 people from the Spanish Rare Disease Register. | Quantitative, retrospective, cross-sectional observational study. | Diagnostic delay increases the psychosocial impact and the need for psychological support. |
| Boettcher et al. (2021) | To examine the factors influencing the mental health of mothers and fathers of children with rare congenital surgical conditions. | 109 mothers and 101 fathers of children with rare congenital anomalies in Germany. | Quantitative cross-sectional observational study. | There are gender differences in coping: the Double ABCX model, mediated by resources and stress, is confirmed among mothers. |
| Boettcher et al. (2025) | To analyse the factors associated with anxiety and depression in parents caring for children with rare congenital conditions, taking gender differences into account. | 675 families of children diagnosed with rare congenital anomalies, from Germany. | Quantitative cross-sectional observational study. | Mothers experience a greater burden and poorer mental health. Interventions should be interdisciplinary and gender-sensitive. |
| Carbyn et al. (2025) | To analyse how social support influences the relationship between the carer's depression and the emotional problems of the child with a rare disorder. | 90 informal carers of children with 22q11DS. Sample from the US, Canada and Europe. | Quantitative cross-sectional observational study. | The more severe the carer's depressive symptoms, the more severe the child's symptoms (internalising/externalising) and the lower the perceived social support. Social support mediates the |

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| | | | | relationship between carer depression and children's internalising symptoms. |
| Chiarotti et al. (2023) | To compare the burden on carers and quality of life between male and female carers of people with RDs. | 210 informal carers from Italy. | Quantitative cross-sectional observational study. | Mothers experience a greater burden and poorer quality of life than fathers. |
| Domaradzki & Walkowiak (2024a) | To examine the relationship between religiosity and caregiving experiences in families of children with RD. | 925 informal carers of children with RD in Poland. | Quantitative cross-sectional observational study. | Religiosity and spirituality act as protective factors for families and patients. |
| Domaradzki & Walkowiak (2024b) | To explore the experiences of carers of children with RD with health and social services. | 925 informal carers of children with RD in Poland. | Quantitative cross-sectional observational study. | RD and fragmented care increase the financial and emotional burden, highlighting the need for early psychological support and comprehensive family care. |
| Domaradzki et al. (2025) | To examine the relationship between the burden of care, quality of life and economic situation among carers of people with RD. | 942 parents of children with RD in Poland. | Quantitative cross-sectional observational study. | The importance of integrating psychological, social and financial support; moving beyond the biomedical model; the diagnostic odyssey is a common experience. |
| González et al. (2024) | To assess anxiety, depression, the burden of care and quality of life among carers of children with nephropathic cystinosis. | 9 mothers (carers) of children with nephropathic cystinosis from Barcelona, Spain. | Quantitative cross-sectional observational study. | Carers have a poorer quality of life than the general population; high prevalence of anxiety and depression. Need for screening and psychosocial support. |
| Hager et al. (2024) | To compare barriers to accessing mental health services between carers of children with rare diseases and carers of children with common medical or mental health conditions. | 305 informal carers of children with common illnesses or rare diseases in the US. | Descriptive qualitative study. | Barriers: limited access to specialists, financial burden, insufficient coverage. Need for comprehensive care, psychological support, and social and healthcare coordination. |
| Hoffmann et al. (2025) | To explore how parents cope with grief and uncertainty whilst caring for a child with a rare | 24 parents of children with rare diseases or undiagnosed conditions in the UK experiencing significant | Descriptive qualitative study. | Diagnostic uncertainty leads to ongoing grief, which requires processes of reframing, a search for meaning and psychological support. |

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|---|---|---|---|---|
| | disease or an undiagnosed condition. | psychological distress due to their role as carers. | | |
| Jobling et al. (2025) | To analyse the impact of having a child with a rare disease on family functioning. | 462 families with children with RD in Australia. | Quantitative cross-sectional observational study. | The impact on the family is significant and depends on the child's health; it is exacerbated by financial difficulties, delays in care and an increased family burden, highlighting the need for psychosocial support. |
| Koehly et al. (2023) | To analyse how relationships within the carer's social environment influence involvement in the care of children with rare or undiagnosed conditions. | 166 primary informal carers of children with rare diseases or undiagnosed conditions. | Quantitative cross-sectional observational study. | Social support and family dynamics are key to alleviating the carer's stress. |
| Lim et al. (2025) | To assess how the child's functioning and the family context influence the quality of life of parents of children with Barth syndrome. | 64 parents: 33 of children with BTHS and 31 of unaffected children, from the USA. | Quantitative cross-sectional observational study. | The child's functional ability influences the parents' quality of life. There is a need for comprehensive, multidisciplinary support. |
| Manalel et al. (2024) | To examine the relationship between carer stress and positive aspects of caregiving, as well as the role of interpersonal coping strategies. | Survey of carers of: children with inherited metabolic disorders (n=167), undiagnosed children (n=23) and typically developing children (n=74). | Quantitative cross-sectional observational study. | There are high levels of parental stress; emotional support mitigates stress; support-based interventions show promise. |
| Maltseva et al. (2023) | To analyse the sleep quality of carers of children with Dravet syndrome and its relationship with mental health. | 108 carers / patients with Dravet syndrome in Germany. | Quantitative cross-sectional observational study. | Poor sleep quality and high levels of anxiety/depression among carers; relationship with patient symptoms; need for a comprehensive therapeutic approach. |
| Nematollahi et al. (2025) | To describe the quality of life of children with congenital multiple arthrogryposis and analyse its association with clinical and psychosocial factors. | 192 children and young people (aged 8–21) with arthrogryposis multiplex congenita (AMC) from the USA. | Quantitative cross-sectional observational study. | Caregivers' quality of life deteriorates as the need for care increases. There is a need for ongoing psychosocial support and interdisciplinary interventions. |

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|---|---|---|---|---|
| Rovira-Moreno et al. (2020) | To assess the impact of an educational programme on rare diseases on the knowledge, psychological well-being and satisfaction of patients and their families. | 37 participants (patients, family members and members of rare disease associations) from Catalonia, Spain. | A quasi-experimental study with a single-group pre-post design. | Significant improvement in knowledge and psychological adjustment following the programme. |
| Smits et al. (2022) | To identify the main support and care needs of parents of children with RDs. | 12 participants with children with RD who had received follow-up or treatment for their child less than a year previously in the Netherlands. | Descriptive qualitative study. | Lack of coordination of care, lack of emotional and financial support, and social isolation. |
| Valcárcel-Nazco et al. (2022) | To examine the relationship between the burden of care and quality of life among carers of people with RA. | 825 informal carers of patients with RDs in France, Germany, Italy, Spain, Sweden and the United Kingdom. | Quantitative cross-sectional observational study. | Care burden predicts anxiety/depression. Need for psychological, social and respite support. |
| Walkowiak & Domaradzki (2025) | To compare the psychosocial burden and quality of life among mothers of children with different neurological rare diseases. | 302 mothers of children with 22q11.2 DS, AS, DS or WS; recruited from patient support groups on Facebook in Poland. | Quantitative cross-sectional observational study. | High burden and exhaustion among mothers; late diagnosis and financial difficulties exacerbate the impact. |
| Witt et al. (2023) | To identify the psychosocial experiences and needs of children with developmental disorders and their families in order to improve care. | 74 informal carers and 15 children/adolescents with RDs from Germany. | Exploratory qualitative study. | Living with a rare disease affects the entire family system and requires specific support, easier access to services, trained professionals and integrated psychosocial care. |
| Wu et al. (2020) | To analyse the impact of rare paediatric genetic conditions on parents' health. | 207 parents of children with rare diseases in Australia. | Quantitative cross-sectional observational study. | Significant indirect effects on parents' health, which must be taken into account when assessing the diagnosis and care. |

Source: Authors' own elaboration.

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Psychological and Emotional Impact of Diagnosis

Consistently, caring for a child with a rare disease was associated with sustained emotional distress among parents, including high levels of stress, anxiety, anguish and perceived burden, particularly in contexts of clinical uncertainty and insufficient psychological support (Aldiss et al., 2021; Hager et al., 2024; Smits et al., 2022). This impact tended to intensify during the pre-diagnostic period and at times of complex decision-making, leading parents to systematically prioritise their child's needs and delay seeking psychological support for themselves (Hoffmann et al., 2025; Smits et al., 2022).

Several quantitative studies found that clinically significant symptoms of anxiety and depression were associated with poorer family emotional adjustment and a significant reduction in health-related quality of life, particularly in situations involving a high caregiving burden and accumulated stress (Boettcher et al., 2025; Carbyn et al., 2025; González et al., 2024; Jobling et al., 2025; Maltseva et al., 2023; Valcárcel-Nazco et al., 2022; Wu et al., 2020). In addition, several studies identified greater psychological vulnerability among mothers who were caregivers, associated with greater involvement in caregiving, greater disruption to employment and heightened concern about their child's future, increasing the risk of anxiety, depression and emotional burden (Boettcher et al., 2021; Chiarotti et al., 2023; Walkowiak & Domaradzki, 2025). More specifically, diagnostic delay was associated with greater parental psychological distress, expressed through frustration, fear, irritability, difficulties in decision-making and poorer quality of life (Benito-Lozano et al., 2023; Walkowiak & Domaradzki, 2025). At the same time, qualitative evidence described parental grief as an ongoing and fluctuating process that coexists with parenting and is shaped by clinical uncertainty, disease progression and the support available, without a clearly defined endpoint (Hoffmann et al., 2025).

Uncertainty and Lack of Information During the Diagnostic Process

Uncertainty regarding diagnosis, treatment, prognosis and the child's future emerged as a recurring feature of the family experience and was associated with significantly greater parental anxiety, emotional distress and a sense of vulnerability, particularly when the information provided by professionals was perceived as insufficient, unclear or inconsistent (Lim et al., 2025; Smits et al., 2022). This situation was exacerbated by a lack of specific knowledge about rare diseases and the absence of explanations regarding clinical procedures, forcing many families to assume an "expert" role within the care process and increasing persistent feelings of frustration and insecurity (Smits et al., 2022).

In this context, diagnostic delay —characterised by multiple consultations, prolonged delays and misdiagnoses— was associated with the so-called diagnostic odyssey, linked to greater psychological, social and functional impact, as well as a greater need for psychological care during the search for a diagnosis (Benito-Lozano et al., 2023; Domaradzki & Walkowiak, 2024b). The absence of a clear diagnosis and prognosis intensified fear, frustration and a sense of loss, hindered the construction of a coherent family narrative and increased anxiety among both parents and the children themselves (Aldiss et al., 2021; Hoffmann et al., 2025).

From the parental perspective, diagnostic delays were perceived as particularly harmful to the child's health because of their association with unnecessary hospitalisations, inappropriate treatments and repeated exposure to invasive procedures, thereby increasing the stress and emotional burden associated with caregiving (Walkowiak & Domaradzki, 2025).

Parental Role Overload and Family Tensions in the Context of Rare Diseases

Parents predominantly assumed responsibility for caring for the child as an almost exclusively personal obligation, involving a high physical and emotional burden and difficulties in delegating or seeking external support, even when they recognised the need for it (Koehly et al., 2023; Smits et al., 2022). Taking on this caregiving role was associated with tensions within the family and the couple relationship, particularly when decisions were made through trial and error and disagreements arose regarding the child's care (Koehly et al., 2023).

Rare disease affected the family system as a whole, frequently becoming central to everyday life and requiring constant reorganisation, with effects on the couple relationship and on attention to siblings, whose emotional needs could be relegated to a secondary position (Smits et al., 2022;

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Witt et al., 2023). Family functioning was particularly compromised when the child's health condition was severe, financial difficulties coexisted or more than one child was affected. In these cases, the time devoted to caregiving was associated with greater burden, poorer family functioning and reductions in or withdrawal from paid employment, particularly among women caregivers (Aldiss et al., 2021; Chiarotti et al., 2023; Jobling et al., 2025; Walkowiak & Domaradzki, 2025).

Consequently, caregiving burden was consistently associated with poorer health-related quality of life across multiple domains, highlighting the interdependence between caregiver well-being and that of the child (Domaradzki et al., 2025; Nematollahi et al., 2024; Valcárcel-Nazco et al., 2022; Wu et al., 2020).

Relationship with the Healthcare System, Social Support and Peer Networks

Interaction with the healthcare system was frequently described in terms of difficulties in empathic communication, experiences of not being listened to, limited specific knowledge of rare diseases and restricted involvement in clinical decision-making, all of which increased stress and undermined trust in professionals (Koehly et al., 2023; Smits et al., 2022). This was compounded by restricted access to mental health services due to waiting lists, shortages of specialised professionals and logistical barriers arising from the child's clinical condition, delaying or preventing psychological care even when a need for it was perceived (Hager et al., 2024).

Overall, the diagnostic process was perceived as particularly stressful because of fragmented care, poor coordination between professionals and repeated testing. Nevertheless, greater satisfaction with healthcare was associated with fewer symptoms of anxiety and depression among caregivers, suggesting a potentially protective role for perceived quality of care and clinical communication (Domaradzki & Walkowiak, 2024a, 2024b; Smits et al., 2022).

## 4. Discussion

The aim of this systematic review was to analyse the psychological impact of late diagnosis of rare diseases in childhood, with particular attention to the role of newborn screening as a protective factor. The findings indicate that newborn screening is not only an essential public health tool, but also a key element of health equity, since its absence or uneven implementation has medical and functional consequences and, particularly importantly, psychological and emotional consequences for families.

The evidence reviewed suggests that early detection helps reduce diagnostic uncertainty, parental anxiety and family stress, facilitating earlier and more structured processes of adaptation to the diagnosis (Noritz & Murphy, 2013; Pelentsov et al., 2016). In this regard, newborn screening may be understood as a protective factor not only from a clinical perspective, but also from a psychological one, as it can shorten the period spent seeking a diagnosis and facilitate the planning of care and support from the early stages of development. However, these benefits are not distributed evenly across Spain, where the implementation of newborn screening continues to vary between autonomous communities, making access to early diagnosis dependent on place of birth (Observatorio Legislativo de Enfermedades Raras y Medicamentos Huérfanos, 2022). This territorial variability raises a significant issue of health equity and highlights the need to move towards more standardised models of early detection.

From a psychological perspective, the findings show that late diagnosis cannot be understood solely as a biomedical problem, but rather as a prolonged stressor that significantly affects the family system. The so-called diagnostic odyssey involves prolonged periods of uncertainty, multiple medical consultations and difficulties in accessing adequate information and resources, creating a context of chronic stress that contributes to emotional exhaustion among caregivers (Aldiss et al., 2021; Jobling et al., 2025; Smits et al., 2022; Witt et al., 2023). This context also constitutes the child's developmental environment, suggesting potential indirect repercussions for their psychological well-being and socio-emotional development.

From this perspective, the psychological impact of late diagnosis can be conceptualised as a systemic and relational phenomenon affecting the family unit as a whole rather than only the primary caregiver. The constant reorganisation of everyday life, caregiving burden and uncertainty regarding the course of the disease generate cumulative emotional strain that affects family functioning, quality of life and socioeconomic stability. These findings are consistent with

previous research indicating that parental well-being is closely linked to the child's psychological adjustment and overall family functioning.

The findings reinforce the need to develop early and ongoing interventions to support families throughout the entire diagnostic process. In this context, the health psychologist plays a central role in identifying emotional needs, reducing caregiver burden and developing adaptive coping strategies (FEDER, 2009; Ministerio de Sanidad, 2014; Rafferty & Beck, 2020). However, the evidence reviewed shows that psychological support is not always systematically integrated into healthcare provision, with difficulties in access, limited referrals and an absence of emotional support even in contexts of substantial psychological need (Aldiss et al., 2021; Domaradzki & Walkowiak, 2024; Hager et al., 2024). This situation highlights the need to strengthen the presence of psychology professionals within healthcare services related to rare diseases.

Similarly, the findings suggest that the healthcare system should incorporate planned psychosocial support mechanisms alongside medical care. Peer support networks are particularly relevant, as they have shown positive effects in reducing social isolation and improving emotional coping. The promotion of these networks should not depend exclusively on family initiative, but should be integrated into coordinated strategies involving healthcare services, social services and patient associations, as has been proposed in the Spanish context (FEDER, 2014; Gaintza et al., 2024).

The evidence reviewed also shows that the financial burden associated with caregiving is a particularly relevant moderating variable in the psychological impact experienced by families. Healthcare costs, loss of income, difficulties in reconciling employment and caregiving responsibilities, and the absence of structural support intensify emotional stress and psychological distress among families, with possible indirect repercussions for the child's well-being (Domaradzki et al., 2025; Walkowiak & Domaradzki, 2025). These findings reinforce the need to address the impact of late diagnosis from a structural perspective that combines healthcare provision with social and economic measures aimed at reducing the overall burden of caregiving.

Overall, the findings of this review suggest that newborn screening and the early detection of rare diseases should be understood as essential components of comprehensive healthcare. Beyond their diagnostic function, these measures can contribute to improving the psychological well-being of families and reducing the emotional impact associated with the diagnostic process. From this perspective, preventing diagnostic delay constitutes not only a clinical objective, but also an important goal in promoting mental health in childhood and within family contexts.

Limitations

With regard to the limitations of this review, the scarcity of studies conducted in Spanish populations should first be noted. Although the literature search was initially geographically restricted to Spain and to the previous five years, only two studies meeting these criteria were identified, limiting the possibility of drawing specific conclusions applicable to the national context. This lack of evidence highlights an important gap in current research and hinders the design of interventions tailored to the characteristics of the Spanish healthcare system and the actual needs of families. The review also includes studies focusing on highly heterogeneous conditions, with differing levels of severity, clinical course and care needs. This diagnostic heterogeneity, together with the diversity of psychological variables assessed and measurement instruments used, makes direct comparisons between studies difficult and precludes integrated quantitative analyses, such as meta-analyses, thereby limiting the possibility of establishing overall estimates of psychological impact. Another limitation concerns the methodological characteristics of the included studies. As reflected in the AXIS and CRF-QS assessments, many studies used convenience samples, frequently recruited through patient associations or online platforms, which may introduce selection bias and limit sample representativeness. In addition, several studies did not justify sample size and showed limitations in controlling for confounding variables, aspects that should be taken into account when considering the generalisability of the findings.

Despite these limitations, the consistency of the findings across quantitative and qualitative studies lends robustness to the conclusions and makes it possible to identify clear patterns regarding the psychological impact of late diagnosis and the protective role of early detection.

## 5. Conclusions

The findings show that late diagnosis constitutes a significant risk factor for family psychological well-being, reinforcing the need to systematically incorporate psychological support into healthcare services for children with rare diseases. In this regard, further research in the Spanish context should be prioritised, particularly studies specifically examining the psychological impact of late diagnosis, assessing families' emotional and social needs, and enabling the design of evidence-based psychological interventions. The generation of applied knowledge in this field would facilitate the development of care models better suited to the characteristics of the Spanish healthcare system.

From an applied perspective, the findings of this review suggest the need to develop organisational and healthcare measures aimed at reducing the psychological impact of the diagnostic process by explicitly integrating psychological care into rare disease care pathways. The main operational recommendations include the following: (1) Establish automatic referral protocols for psychological care following the communication of positive newborn screening results or confirmation of a rare disease diagnosis. (2) Define coordinated care pathways involving paediatrics, clinical genetics, paediatric neurology and health psychology, enabling continuous support throughout the diagnostic process. (3) Develop structured diagnostic communication protocols that include guidance on conveying medical information and the early identification of families' emotional needs. (4) Integrate psychosocial support services and peer support networks as an institutional component of healthcare, in coordination with patient associations and organisations such as FEDER, thereby preventing access to these resources from depending exclusively on family initiative. (5) Facilitate early access to early intervention programmes and family support, particularly in situations of prolonged diagnostic uncertainty. (6) Promote specific training for healthcare professionals in rare diseases and clinical communication in order to reduce perceived diagnostic uncertainty and improve families' experience of care. These findings reinforce the need to move towards genuinely comprehensive healthcare models in which the health psychologist plays a central role in supporting children with rare diseases and their families.

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