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Experiences surrounding the diagnostic process and care among parents of children diagnosed with Phelan-Mcdermid Syndrome: A qualitative study in Spain.

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Keywords:	Phelan-Mcdermid Syndrome, Telomeric 22q13 Monosomy Syndrome, Rare Diseases, Parents, Qualitative research

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Running title: Parents and Phelan-Mcdermid Syndrome

Abstract

Aim

The Phelan-Mcdermid Syndrome (PMS) is a rare, underdiagnosed disease with no cure. The purpose of this study was to explore the experience of parents with children diagnosed with Phelan-Mcdermid Syndrome, regarding the diagnostic process, treatment, and medical care.

Method

A qualitative exploratory study was conducted. Participants were recruited using non-probabilistic purposeful sampling. In total, 32 parents with children with PMS were included. In-depth interviews and researchers' field notes were used. An inductive thematic analysis was performed.

Results

Five themes emerged: "The diagnostic process" described the diagnostic process and how it is communicated to the parents; "Treatment and expectations" described the expectations and hopes built on future treatment; "Family planning" described how parents deal with genetic counseling, planning to have more children after the diagnosis of PMS; "The world of disability" describes the parents' entry into the environment of dependency and disability after the diagnosis; "The family economy" showed the economic difficulties due to the high cost of therapies and daily care products.

Interpretation

Our results provide insight on how the diagnosis and their consequences are experienced by parents with children with PMD. These results can be used by health professionals to help and support parents.

Keywords (MeSH): Phelan-Mcdermid Syndrome; Telomeric 22q13 Monosomy Syndrome; Rare Diseases; Parents; Qualitative research

List of Abbreviations in alphabetical order

ASD: Autism Spectrum Disorder

CGH: Comparative genomic hybridization

DICE-APER: Primary Care for Rare Diseases

FISH: Fluorescence in situ hybridization

PMDS: Phelan-McDermid syndrome

RD: Rare diseases

What this paper adds

- The Phelan-Mcdermid Syndrome breaks parents´ expectations regarding diagnoses
- Parents experience diagnoses process and its communication as different process
- Disability and dependence appear abruptly in the parents´ life.
- Financial costs impact on family's structure and planification.

1. Aim

Phelan-McDermid syndrome (PMDS), or 22q13.3 deletion, consists of a genetic mutation of the SHANK3 gene. This chromosomal abnormality is of autosomal dominant inheritance or appears spontaneously ("de novo")[1,2]. Due to the great phenotypic variety, PMD is an underdiagnosed syndrome[3]. The differential diagnosis of PMDS includes Prader-Willi, Fragile X, Angelman, Smith-Magenis, tricorhinophalangeal and velocardiofacial syndromes, Autism Spectrum Disorder (ASD), cerebral palsy, developmental delays and syndromes associated with hypotonia[1,2,4]. Upon confirmation of the genetic diagnosis, parents are recommended to receive genetic counseling[5]. Genetic counseling involves education sessions and multiple genetic testing to determine inheritance, understand diagnosis, and the progression of PMDS[4]. In rare diseases (RD), parents highlight how the diagnosis causes a strong emotional impact[6,7].

At present, there is no specific pharmacological treatment for PMDS[8]. Most treatments are aimed at treating comorbidities such as anxiety, hyperactivity, and impulsive behaviors[9]. Due to the variability of the PMDS phenotype, shankopathies may present with neurodevelopmental impairment, neonatal hypotonia, intellectual disability, and/or ASD-like symptoms. Other neuropsychiatric manifestations include epilepsy, bipolar disorders, and schizophrenia[10,11]. PMDS is classified according to the involvement of the SHANK3 gene.

As with other pediatric-age rare diseases, parents have difficulty accessing medicines and public health services (diagnosis and therapies), having to bear the economic costs of private and specialized health care[12,13]. The experience of having children diagnosed with PMDS is highly individual, and qualitative research can provide a more holistic view that may be more meaningful to parents[12]. In contrast, there are no previous qualitative studies describing the experience of parents with children diagnosed with PMDS. The purpose of this study was to explore the experience of parents with children diagnosed with PMDS regarding the diagnostic process, treatment, medical care, and disability.

2. Method

2.1. Study design

A qualitative exploratory study was conducted[14]. This study was conducted according to the Standards for Reporting Qualitative Research[15]. Ethical approval for this study was granted by the Local Ethical Committee of Universidad X (code: 0810202017820). All participants provided oral informed

consent prior to their inclusion[14]. Six researchers (one man) participated in this study, including one research nurse (DPC), one physical therapist (SGB) and four occupational therapists. All researchers had experience in research in health sciences.

2.2. Participants, and sampling strategies

The study included parents with Children diagnosed with PMDS by a medical specialist[16] and who were attending the Asociación Síndrome Phelan McDermid (https://22q13.org.es/), Spain. The inclusion criteria were: a) Parents who, at the time of the study had children diagnosed with PMDS (with genetic diagnosis), and/or legal guardian; b) the diagnosis of PMDS was made by the pediatrician and/or the neurologist, c) children could present any variation of PMDS (deletion or mutation), and d) signing the informed consent. Exclusion criteria: a) a diagnosis not confirmed by the pediatrician and/or neurologist, and b) not signing the informed consent.

Purposive sampling was used, based on relevance to the research question (not clinical representativeness)[14]. In in the current study, the sample size was determined following the Turner-Bowker et al proposal[17]. These authors[17] reported that 99.3% of concepts, themes, and contents emerged with around 30 interviews. With this proposal, a greater capacity to identify codes, categories, and topics is achieved. In addition, the current proposal also helps researchers to know when to stop data collection and participant recruitment.

2.3. Data collection

Data were collected over a four-month period between November 2020 and February 2021. A semi-structured question guide was used (**Table 1**). In addition, researcher field notes were kept during the interviews[18]. During the interviews, researchers used prompts to encourage the participant to provide further details.

Due to the lockdown situation established by the Spanish Government in response to the COVID-19 pandemic[19], interviews were conducted via a private video chat room using the Microsoft Teams platform (https://www.microsoft.com/es-es/microsoft-teams/log-in). Each participant received a private and personalized email with an invitation. All interviews were conducted by three authors (CGB, RMMP and DPC). With participant oral permission, all interviews (n= 32) were audio- and video-recorded, recording a total of 3,205 min of interviews overall (average of 100.16 ± 18.2 min each interview).

Additionally, 32 field notes were collected by the researchers during the semi-structured interviews, since field notes provide a rich source of information[18].

2.4. Data analysis

An inductive thematic analysis was performed[14]. The analysis consisted of identifying the most descriptive content in order to obtain codes, and subsequently reduce and identify the most common meaningful groups (categories). In this manner, groups of meaningful units were formed (i.e., similar points or content that allowed the emergence of the topics that described the study participants' experience)[14,18]. This process was performed separately on the interviews and the researchers' field notes. Also, double, and independent coding was performed by two investigators (CGB, DPC). After they then met to discuss, compare, and for a round of refinement. Subsequently, joint meetings were held to combine the results of the analysis, to represent the parents' experiences[14,18]. The Excel program was used to organize and share the coding process.

2.5. Rigor

The techniques performed to control trustworthiness are described in **Table 2**[14,20].

3. Results

Thirty-two parents were recruited (23 women). The mean age of parents was 43 years and 4 months (SD ±6.85). The mean age of children with PMDS was 11 years and 7 months (SD ±9.79) and the age at the children´s diagnosis was 7 years and 1 months (SD ± 10.04). The clinical and demographic features of participants are shown in **Table 3**.

Five specific themes emerged: a) The diagnostic process; b) Treatment and expectations; c) Family planning; d) The world of disability; and e) Family economy. We included some of the parents' narratives taken directly from the interviews in relation to the emerging themes (See **Supplementary file 1)**.

Theme 1: The diagnostic process

The parents described the diagnostic process as a search. In that search they felt alone and that a lot of time is lost. Time that is essential for their children's treatment. In addition, they described a constant struggle to get the public health system to request specific genetic tests for diagnosis:

"They requested it [the test], but as always, it was a streggle. We don't ask for much, it's for his health and when you claim or ask for more specific tests for your child, they keep closing the door on you." (P27).

Our participating mothers pointed out how their criteria or assessments of their child and his development were questioned, causing them very unpleasant moments. Some parents highlighted the moment when they were informed of the diagnosis as the most shocking news of their lives. They were so "shocked" that they stopped listening, unable to hear anything else.

"The day of the diagnosis was the worst day of my life, hands down. I've never been closer to insanity ever, feeling like my brain was going to go ploff [popping gesture]." (P9).

However, other parents were relieved by the diagnosis because they found an explanation for their children's behavior.

"I felt relief. It upsets you, and it hurts, but it helped because after a long wait, we finally had an answer." (P11).

Most of the parents described how the communication of the diagnosis by the health professional was devastating, due to lack of training and knowledge of PMDS.

"It's unacceptable that the person who is giving you the diagnosis doesn't even know what it is...it's devastating." (P23).

Theme 2: Treatment and your expectations

Parents described how, in the absence of specific pharmacological treatment, therapies such as physiotherapy, occupational therapy or speech therapy are their only tool and hope to fight PMDS. The participants described how these therapies should be carried out by trained professionals, because no matter how much the professionals teach parents to perform certain techniques at home, they perceive a clear difference when the children go to a professional center.

"I talk to other parents, and they tell me that they have learned to become therapists and physical therapists, but it's not true. We can't get so much out of the kids as they do. Taking her to therapy is the most important thing I can do for her." (P23).

A moment of great uncertainty and complexity for parents is to assess when a medication or therapy is causing harm to their children and to decide when to stop.

"... there comes a time when you ask yourself, what are we waiting for to keep taking this medication? Because you start to see other alterations that force you to rethink everything again, and you may harm your daughter." (P3).

Finally, all study participants emphasized that they had no hope for future treatment due to the genetic complexity of the study.

"It is very sad, but we have no hope. We aren't going to fool ourselves; our son isn't going to get better. We are aware that regression will come, we don't know when, but it will come. There is no hope at all." (P1).

Theme 3: Family planning

This theme describes how parents consider having another child after the diagnosis of PMDS by seeking genetic counseling, together with the couple's disagreements on the subject and the feeling of guilt that sometimes ensues.

The decision to have another child is accompanied by great uncertainty and fear among couples. After the diagnosis of PMDS, some participants considered genetic counseling as a tool to consider having children again. Moreover, they recounted how receiving genetic counseling should always be accompanied by psychological support, as it is a stressful and distressing situation.

"We went to genetic counseling, and they tried to convince me that the probability of having a child with the syndrome again was very low... but don't talk to me about probability because I have had a child Phelan McDermid Syndrome. Not everything is statistics... Besides, where is the mental health service? We were never given that." (P9).

For some couples, considering a new pregnancy is an opportunity to recover all those experiences that they have not been able to live, and to have a more complete experience of motherhood/parenthood. Meanwhile, for other participants, the decision for another pregnancy caused conflicts between the couple. Some of the participants reported that they felt guilty about deciding to have another child, because taking care of the sibling with PMDS could be a burden for their sibling in the future.

Theme 4: The world of disability

The parents interviewed described how they are immersed in a world they never knew existed and had never been interested in. All our parents pointed out that no one has prepared them to accept that their children "have a disability" and are labeled as "disabled".

"It's hard to have a child and acknowledge that they are a person with a disability...because you've already put the label on them, a disabled person...I felt a huge grief..." (P24).

Paradoxically, parents described how they had to use "identifiers" on their clothes to highlight that their child had a disability and to be able to tolerate the child's behavior on the street. This type of marking was used during the confinement period of the COVID-19 pandemic, where only children with special or health conditions were allowed to go out on the street.

"We had to put a blue armband on them so we wouldn't have problems with people. To have them marked with a bracelet as if they were animals so that people would understand. People aren't prepared to understand disability, much less our children who are not physically noticeable." (P8).

Some parents described how the assessment of disability by public agencies is erratic, takes a long time, and the staff that performs it is unqualified and do not know what PMDS is.

Theme 5: The family economy

Parents must make a considerable financial effort to cover the daily care needs of their children (adapted car and wheelchair, diapers) with PMDS. Another problem reported was the difficulty of combining work schedules with the schedules of therapies and medical appointments for their children. This situation is intensified when both parents work to obtain financial resources to meet all the costs of therapy and daily care.

"On a work level it is complex having to combine all the schedules, but you can't stop working because you have to pay for a lot of things." (P11).

Eventually, one of the parents must give up work, due to the high demands of caring for a child with PMDS. In our study, it was common for the mother to reduce her working hours or quit her job.

4. Discussion

This study has highlighted the experiences of parents of children with PMDS. Overall, they experience a difficult and lengthy diagnostic process, waking up in the world of disability with the shock that this entails, which comes with a high financial burden. In addition, the parents spoke of the need to incorporate multiple non-pharmacological therapies to help their children and one of the issues that was emphasized was the need to seek genetic counseling should they decide to have another child.

Previous studies in RD describe the diagnosis as a long process, where many professionals have poor knowledge of the disease, making it difficult to request appropriate tests[6,21,22]. Ivarsson et al.[6] described how parents must continually complain to get a diagnosis. Participants in the present study noted the constant struggle to get the diagnostic tests that would serve to identify PMDS. Moreover, our participants described the anguish and uncertainty they felt in finding the cause of the disease to be able to give "a name" to what was happening to their child. In agreement with our results, several studies describe the "journey from doubt and hope to diagnosis" experienced by parents of children with RD, which helps them to find explanations for their child's behaviors[6,23,24]. During the search for a diagnosis, children with PMDS, previously go through several diagnoses. Previous studies show that children with RD receive several diagnoses before reaching a definitive diagnosis, which is often delayed in time[21,23].

The communication of the diagnosis by the health professional is a very delicate moment for the parents. In our study, receiving the diagnosis from a professional who has no knowledge of the disease is a devastating process. Previous studies describe the moment of communicating the diagnosis of RD to the parents as a key element for understanding and coping with the disease, as well as for psychological well-being and proactive participation in the different treatments[7,25,26]. In addition, they distinguish between poor and effective communication, based on the involvement and interest of the health professional[7,25,26].

Currently, there is no pharmacological treatment for PMDS and medical treatments are aimed at treating comorbidities[8,9]. The lack of treatment for RD derives in feelings of frustration and distress among parents[21]. In addition, previous studies in RD highlight the difficulty parents have in adhering to treatments that attempt to reduce symptoms[21,27]. Parents must decide which therapies and treatments to continue based on the presence of benefits and harms (side effects) for their children[21,27]. In other studies, this decision is influenced by disease status and progression[27,28]. SHANK3 gene involvement can lead to the development of behavioral symptoms consistent with ASD. Recent studies affirm that ASD-targeted therapies, are a good option for children with PMDS with SHANK3 involvement[29]. In addition, parents of children with

RD rate therapies such as physiotherapy, occupational therapy and/or speech therapy as a fundamental pillar in the daily struggle against disability and symptoms[18,30].

Genetic counseling is a multidisciplinary resource that includes diagnosis, information about the disease, psychological support, and consultation about the possibilities of having a child with an anomaly or other detectable genetic problem[31]. This would explain the importance given by parents to genetic counseling when planning to have another child. However, along with genetic counseling, psychological and emotional support for parents should be incorporated[22-24].

Regarding having another child after the diagnosis of PMDS, our results coincide with previous studies[24,32] where some parents wish to have another child after receiving the diagnosis, whereas others choose to not have any more children because of the high daily care demands of the child with PMDS and the associated financial burden.

Haegele et al.[32], in their study on children with chronic metabolic disease, describe how after the diagnosis, parents experience moments of frustration and dread when they think that their child is "different". Our participants also spoke of encountering and discovering the world of disability and the impact they experienced when "labeling" their child as disabled. Along these lines, previous studies also describe the psychological burden and difficulty of assimilating disability among parents of children with RD[32-34]. This can be explained by the stigmatization and lack of understanding of their social environment, including their own family[32-34]. For parents "being different" is a problem they face in their daily lives, feeling judged and misunderstood by other adults[32]. Our participants also spoke of the feeling of incomprehension of others towards their children's disability and disruptive behaviors.

Our results show how parents assume a high financial burden to cover therapies and daily care. A significant financial investment is common in children with RD, where families cover all expenses that are not covered by insurance or public health care[20,26]. Orthopedic aids, autonomy devices and individualized therapies are some of the most identified costs[22,32]. Paradoxically, in addition to these costs, the high burden of care and childcare forces some parents to reduce their working hours or leave work to care for their children, reducing their income[22,33].

5. Conclusions

Our results show how parents experience the diagnosis of PMDS and its impact on parents. These results can be used by healthcare professionals to help and support parents during diagnosis and treatment and/or therapy. There is a need to develop programs that integrate health and social interventions that help parents of children with PMDS to care for and minimize the impact of the disease on the family.

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Supporting information

Supplementary Material 1, reports some of the patients' narratives taken directly from the interviews and personal letters regarding the emerging themes.

Supplementary Material 2, online table of contents of manuscript.

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Table 1. Semi-structured question guide

Areas of research	Questions
Disease	What is it like to live with Phelan McDermid Syndrome? What is most relevant for you? How does it affect your daily life? What are your expectations about the disease and its evolution?
Diagnosis	What was the process leading up to the diagnosis? What was the most relevant aspect of this process? How was the diagnosis communicated to you?
Treatment and care	What treatment is your child currently receiving? What is most relevant to you? How does it affect or limit your daily life? Does it have any side effects? What are your treatment expectations? What barriers and facilitators do you think influence following through with the recommended treatment? Do you use any strategies to cope with the symptoms of the disease? How do you apply caregiving to your child? What barriers and/or facilitators do you have when caring for your child?
Impact on parents	How do you experience your child's illness? What is most relevant? How has it influenced the family's day-to-day organization and planning? At what times/situations have you needed to seek help? What type of help was it?

Table 2. Trustworthiness criteria.

Criteria	Techniques Performed and Application Procedures
Credibility	Investigator triangulation: each interview was analyzed by two researchers. Team meetings were performed in which the analyses were compared and categories and themes were identified. Triangulation of methods of data collection: semistructured interviews were conducted and researcher field notes were kept. Participant validation (member-checking): asking the participants to confirm the data obtained at the stages of data collection. All participants were offered the opportunity to review the audio and/or video records to confirm their experience. None of the participants made additional comments.
Transferability	In-depth descriptions of the study performed, providing details of the characteristics of researchers, participants, contexts, sampling strategies, and the data collection and analysis procedures.
Dependability	Audit by an external researcher: an external researcher assessed the research protocol, focusing on aspects concerning the methods applied and study design. An external researcher specifically checked the description of the coding tree, the major themes, participants' quotations, quotations' identification, and themes' descriptions.
Confirmability	Investigator triangulation, member-checking, and data collection triangulation. Researcher reflexivity was encouraged via the performance of reflexive reports and by describing the rationale behind the study.

Table 3. Demographic and clinical features

Participants	n=32
Sex	Male: 9 Female: 23
Age	Mean age: 43 years & 3.8 months SD: 6.85
Civil status	Married: 25 Divorced: 7
Number of children	Mean: 1,94 SD: 0,72
Age of children with PMS	Mean: 11 years & 6.6 months SD: 9.79
Child's age at diagnosis	Mean: 7 years & 0.4 months SD: 10.04
Diagnoses	Deletion: 30 SHANK3 gene mutation: 2

Supplementary information

Supplementary file 1. Narratives from Themes

Narratives from Theme: The diagnostic process
Search for the diagnosis: *"And as parents, you are always saying: 'for sure, this test will be nothing and will remain an anecdote', and of course, you don't know that there are rare diseases or anything, it is a world that is totally distant from you until you get to know it."* (P24). *"You feel like everything is a waste of time. You feel alone because you go places and no one knows, no one understands, and no one wants to know. It wastes precious time for my son*." (P1).
Fighting for the diagnosis: *"We insisted a lot to the pediatrician to send us to the neuropediatrician and after a lot of struggle, they sent us the tests. We talked about this and it took more than a year for the test to be carried out…"* (P2).
Feeling crazy: *"The feeling I have is that they saw me as a crazy person... a crazy mother who saw things where there were none... it is a very unpleasant feeling…"* (P8). *"They labeled me as being a very crazy mother. They doubted what I told them, what I saw in my son. They told me I was seeing ghosts, that I was imagining things."* (P11).
Communication of the diagnosis: *"They give you the biggest news of your life in the crappiest way. We left the office without knowing what I had. We were shocked"* (P25). *" When they tell you the name you don't hear anything else, it's like they drop the bomb and then you just hear a ringing sound, like the one at the end of a movie. It doesn't matter what they tell you because you don't hear anything anymore."* (P24).
Receiving the diagnosis: *"Even though you know something is going on, you actually break down because you always have a hope that it's nothing. Naming it gave me a lot of peace, even though you are told that it's for life."* (P14). *"I felt relieved because I had been assuming it for a long time, I just wanted to be told clearly what it was."* (P11). *"when you get the diagnosis you hear nothing but the name of the syndrome, no matter what they tell you."* (P19).
Communication of the diagnosis by health care providers: *"Imagine, my world came crashing down when I didn't even know what a neurologist was...".* (P20).
Narratives from Theme: Treatment and expectations
Therapy: *"It's the only thing we can do as parents, take him to therapies. There are times when you think,*

will you be giving him enough therapies, or too many... will they be adequate or would he need others instead..." (P2).
" In the end, instead of taking him to soccer, you take him to therapies... you have to look at it like that." (P6).
Importance of therapies:
"Therapies are basic. As much as we want to, we can't give him everything he needs, sometimes we don't have the energy or we don't know how to approach it. Parents, no matter how much we say we are all-inclusive, we have different approaches." (P24).
"...having a daughter with such great difficulties makes you feel helpless. The only thing you can do is take her to other therapies because there is no pharmacological treatment..." (P9).
Medication:
"In the end you have to put everything in a balance. The benefits and what is bad for him due to the medication... if he doesn't die of his liver, he will die of something else... and at least he is better". (P32).
Expectations for future treatment:
"They are doing tests and things, but to fix it they would have to change the proteins he has in his whole body, I have no hope for that. It seems very complicated to me and the thought of it makes me very anxious." (P14).
Narratives from Theme: Family planning
Genetic counseling:
"genetic counseling is essential to be able to have a child with certain guarantees and peace of mind." (P3).
"the moment they call you to give you the results, it makes your heart sink." (P6).
Thinking about having another child:
"we were a little scared, but it didn't have to go wrong this time." (P4).
"We deserved to have a full parenting experience...enjoy ourselves in a way we hadn't with her." (P17).
Disagreements:
"We had a conflict as a couple because I would have liked to have another child. Besides, I thought it was beneficial for her, but my husband saw it as stealing time from her, stealing dedication from her and...and no." (P3).
"We went through a complicated period because I wanted another child, I thought it would help us in this madness, but my wife didn't want it and I didn't understand it." (P30).
Feeling guilty:
"You get to feel guilty when she thinks about it because he's a child who could have a better life and he's already conditioned. In the future, he's going to be a burden on his sister." (P31).
Narratives from Theme: The World of Disability
Being unprepared:

"It's something they never prepare you for. And when the diagnosis comes, your world falls apart." (P22).
Entering the world of disability: *"Suddenly, you find yourself in a world that you didn't know existed, you didn't know there were rare diseases or anything else and you didn't think about being part of it... it's a very big blow."* (P28). *"Suddenly you're in a parallel world that you hadn't stopped to look at and you don't discover it until you're inside."* (P20).
People don't understand disability: *"...everyone knows so much and tells you how you have to do it and...then, you spend all day crying because everyone thinks they know and they don't have a clue..."* (P5).
Disability assessment: *"The disability assessment process is cumbersome because you have to go there a thousand times, to be assessed... when they know that it"s a rare disease, which unfortunately for us, is not going to improve, and if it does, it will only improve slightly..."* (P6).
Narratives from Theme: The family economy
Financial effort: *"These children have a lot of expenses. Therapies, diapers, wheelchairs... and all of this is private because there is almost no help."* (P14). *"My son had a very strong regression and I had to quit my job, because I couldn't take care of him, take him to therapies and work. He needed me so much that I had to quit my job..."* (P8). *"Diana needs an adapted car that costs 3000€ to adapt. How does a family cope with that? It's a total ruin. And there are more expenses like diapers, which are daily."* (P9).
The need to work: *"...we can't afford a reduction in the working day because we have to keep up with all his therapies, aids, pills..."* (P4).
Having to stop working: *"When I was working, it was like drowning and, in the end, you look at the benefit and it's not that much. We decided to give up other things and be more comfortable taking care of the child".* (P14).

Developmental Medicine & Child Neurology

The present manuscript *"Experiences surrounding the diagnostic process and care among parents of children diagnosed with Phelan-Mcdermid Syndrome: A qualitative study in Spain*." was conducted following the Standards for Reporting Qualitative Research-SRQR (https://www.equator-network.org/).

Check list from **Standards for Reporting Qualitative Research (SRQR): 21-item checklist.**

Reference: O'Brien BC, Harris IB, Beckman TJ, Reed DA, Cook DA. Standards for reporting qualitative research: a synthesis of recommendations. Acad Med. 2014;89(9):1245-1251.

No	Item	Guide questions/description	Yes/no
	Title and abstract		
S1	Title	Concise description of the nature and topic of the study Identifying the study as qualitative or indicating the approach (e.g., ethnography, grounded theory) or data collection methods (e.g., interview, focus group) is recommended	Yes, page 1
S2	Abstract	Summary of key elements of the study using the abstract format of the intended publication; typically includes background, purpose, methods, results, and conclusions	Yes, page 1-2
	Background		
S3	Problem formulation	Description and significance of the problem/phenomenon studied; review of relevant theory and empirical work; problem statement	Yes, page 3
S4	Purpose or research question	Purpose of the study and specific objectives or questions	Yes, page 3
	Methods		
S5	Qualitative approach and research paradigm	Qualitative approach (e.g., ethnography, grounded theory, case study, phenomenology, narrative research) and guiding theory if appropriate; identifying	Yes, page 3-4

		the research paradigm (e.g., postpositivist, constructivist/interpretivist) is also recommended; rationale	
S6	Researcher characteristics and reflexivity	Researchers' characteristics that may influence the research, including personal attributes, qualifications/experience, relationship with participants, assumptions, and/or presuppositions; potential or actual interaction between researchers' characteristics and the research questions, approach, methods, results, and/or transferability	Yes, page 4
S7	Context	Setting/site and salient contextual factors; rationale	Yes, page 4
S8	Sampling strategy	How and why research participants, documents, or events were selected; criteria for deciding when no further sampling was necessary (e.g., sampling saturation); rationale	Yes, page 4
S9	Ethical issues pertaining to human subjects	Documentation of approval by an appropriate ethics review board and participant consent, or explanation for lack thereof; other confidentiality and data security issues	Yes, page 4
S10	Data collection methods	Types of data collected; details of data collection procedures including (as appropriate) start and stop dates of data collection and analysis, iterative process, triangulation of sources/methods, and modification of procedures in response to evolving study findings; rationale	Yes, page 4-5, table 1
S11	Data collection instruments and technologies	Description of instruments (e.g., interview guides, questionnaires) and devices (e.g., audio recorders) used for data collection; if/how the instrument(s) changed over the course of the study	Yes, page 4-5, table 1
S12	Units of study	Number and relevant characteristics of participants, documents, or events included in the study; level of participation (could be reported in results)	Yes, page 4-8-, table 3
S13	Data processing	Methods for processing data prior to and during analysis, including transcription, data entry, data management and security, verification of data integrity, data coding, and anonymization/deidentification of excerpts	Yes, page 4-5
S14	Data analysis	Process by which inferences, themes, etc., were identified and developed, including the researchers involved in data analysis; usually references a specific paradigm or approach; rationale	Yes, page 5
S15	Techniques to enhance trustworthiness	Techniques to enhance trustworthiness and credibility of data analysis (e.g., member checking, audit trail, triangulation); rationale	Yes, page 4-5, table 2

	Results/Findings		
S16	Synthesis and interpretation	Main findings (e.g., interpretations, inferences, and themes); might include development of a theory or model, or integration with prior research or theory	Yes, page 5-8
S17	Links to empirical data	Evidence (e.g., quotes, field notes, text excerpts, photographs) to substantiate analytic findings	Yes, page 5-8
	Discusssion		
S18	Integration with prior work, implications, transferability, and contribution(s) to the field	Short summary of main findings; explanation of how findings and conclusions connect to, support, elaborate on, or challenge conclusions of earlier scholarship; discussion of scope of application/ generalizability; identification of unique contribution(s) to scholarship in a discipline or field	Yes, page 9-10
S19	Limitations	Trustworthiness and limitations of findings	Yes, page 10
	Other		
S20	Conflicts of interest	Potential sources of influence or perceived influence on study conduct and conclusions; how these were managed	Yes, described at manuscript
S21	Funding	Sources of funding and other support; role of funders in data collection, interpretation, and reporting	Yes, described at manuscript