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Desafios da maternagem de filhos com doenças raras

The challenges of mothering children with rare diseases

Los retos de ser madre de niños con enfermedades raras

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RESUMO

Objetivo: compreender a experiência de maternidade de mulheres cujos filhos enfrentam doenças raras e requerem cuidados contínuos. **Método**: abordagem qualitativa. Narrativa de Vida, juntamente com a técnica de Snowball para ampliar as entrevistas, foi conduzida uma pesquisa descritiva. As entrevistas abertas foram guiadas pela pergunta central: descreva a descoberta do diagnóstico da doença rara de seu filho e sua jornada desde então. A análise dos dados ocorreu através da leitura flutuante e agrupamento por similaridade. **Resultados**: as participantes possuem alta escolaridade, em sua maioria se autodeclaram brancas, têm a presença do pai da criança na maioria dos casos, e metade enfrenta desafios de emprego devido aos cuidados intensivos. **Conclusão:** o estudo atingiu seu objetivo, destacando a necessidade de cuidado psicossocial de enfermagem para mães cuidadoras, dada sua vulnerabilidade psicológica. Este trabalho pretende ser um ponto de partida para pesquisas subsequentes, enfatizando a importância da visibilidade da maternagem atípica.

Descritores: Enfermagem materno-infantil; Doenças raras; Criança hospitalizada; Enfermagem; Pediatria.

ABSTRACT

Objective: to comprehend the motherhood experience of women with children facing rare diseases and requiring continuous care. **Method:** a qualitative approach was employed, utilizing the Life Narrative method along with the Snowball technique to expand interviews in descriptive research. Open-ended interviews were guided by the central question: describe the discovery of your child's rare disease diagnosis and your journey since then. Data analysis involved floating reading and grouping by similarity. **Results:** participants exhibit high educational levels, mostly self-identifying as white. The child's father is present in most cases, and half face employment

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challenges due to intensive caregiving. **Conclusion:** the study achieved its objective, emphasizing the need for psychosocial nursing care for caregiving mothers given their psychological vulnerability. This research aims to serve as a starting point for subsequent investigations, emphasizing the importance of visibility for atypical motherhood.

Descriptors: Maternal-child nursing; Rare diseases; Child hospitalized; Nursing; Pediatrics.

RESUMEN

Objetivo: comprender la experiencia de la maternidad de mujeres con hijos que enfrentan enfermedades raras y requieren cuidados continuos. **Método:** se empleó un enfoque cualitativo, utilizando el método de Narrativa de Vida junto con la técnica de Snowball para ampliar las entrevistas en una investigación descriptiva. Las entrevistas abiertas fueron guiadas por la pregunta central: describe el descubrimiento del diagnóstico de la enfermedad rara de tu hijo y tu trayectoria desde entonces. El análisis de datos incluyó lectura flotante y agrupamiento por similitud. **Resultados:** los participantes tienen niveles educativos elevados, en su mayoría se autoidentifican como blancos. El padre del niño está presente en la mayoría de los casos, y la mitad enfrenta desafíos laborales debido al cuidado intensivo. **Conclusión:** el estudio logró su objetivo, enfatizando la necesidad de cuidado psicosocial de enfermería para madres cuidadoras dada su vulnerabilidad psicológica. Esta investigación pretende ser un punto de partida para investigaciones posteriores, destacando la importancia de la visibilidad de la maternidad atípica.

Descriptores: Enfermería maternoinfantil; Enfermedades raras; Niño hospitalizado Enfermería; Pediatría.

INTRODUCTION

Rare diseases (RD) pose significant health challenges due to their low prevalence within the general population. Although individually rare, these diseases affect a substantial number of people globally, making them an important topic of research and medical attention. With an impressive diversity of clinical manifestations, rare diseases often present late diagnosis and difficulties in accessing appropriate treatments.[1]

RD can cause a wide range of physical, psychological, and social symptoms. Physical symptoms can range from mild to severe and can affect any system in the body. Psychological symptoms can include anxiety, depression, and social isolation. Social symptoms can make it difficult to participate in school, work, and social life.[2]

Estimating the global prevalence of RD is challenging, primarily because of data diversity of the data, which is derived from a variety of disparate sources of information that are not standardized or are difficult to combine, including published case reports or systematic reviews, patient records, expert opinions and other anecdotal evidence. [3]

The nature of RD poses additional challenges due to the small number of cases, compounded by significant clinical heterogeneity. Some vary in frequency across the geographical area, due to population genetic diversity, environmental or social pressures or survival problems in different regions.[4]

The point prevalence of rare clinical presentations may be overestimated or underestimated due to their overlap with common comorbidities. A conservative estimate of the population prevalence of RD ranges from 3.5% to 5.9%, which equates to 263 to 446 million people affected globally at any given time. This figure is derived from data on 67.6% of prevalent rare diseases, using the European definition of 5 per 10000 and excluding rare cancers, infectious diseases and poisonings.[3-4]

It is noteworthy that RD definitions vary globally, and the lack of standardization in diagnostic and coding criteria can affect estimates. In addition, the field of rare diseases is constantly evolving with new genetic discoveries and advances in medical research, which can influence estimates over time.[4]

In Brazil, there are around 13 million Brazilians who have RD.[5] However, the causes and origins of these diseases are still unclear, but studies show that 80% of them are due to genetic factors and some may be due to environmental and infectious factors, among others. [6]

Studies report that 30% of patients affected by these diseases die before the age of five and in 75% of cases, they affect children.[7] According to the Statute of the Child and Adolescent (ECA), under the terms of Article 20 of Law 8.069/90, a child is considered to be a person up to 12 (twelve) years of age.[8] When affected by DR at the beginning of their development, the family's efforts will have to be redoubled, beyond what is expected for this stage, and these individuals will become dependent throughout their existence. However, it is known that the maternal figure excels in this care, almost always because the female figure has been constructed as the most suitable to welcome, feed and care for.[9]

Parenting children with RD introduces challenges that exceed those of typical parenting.. These conditions are often chronic, complex and may require specialized medical care, making the journey for parents, especially mothers, unique and challenging. Important aspects of parenting children with rare diseases, such as diagnosis and adaptation, can be time-consuming and emotionally draining for parents. Uncertainty and the search for answers often mark the first years of their child's life. Acceptance of the diagnosis is a crucial step, and mothers play a central role in adapting to the new reality.[10]

These mothers often face significant emotional challenges. Feelings of isolation, anxiety, sadness, and stress are common. Mental health management thus becomes an essential part of the maternal role, and they become active advocates in the search for resources, treatments, and support for their children. Advocacy is an integral part of mothering children with rare diseases, involving participating in support communities, raising awareness about the condition, and fighting for policies that benefit their specific needs.[11-12]

The cost of medical care and therapies can be substantial. Mothers often face significant financial challenges to ensure that their children have access to all the necessary resources. This can include treatments not covered by insurance, expenses for specialized medicines and costs associated with frequent appointments.

In summary, mothering children with RD is a complex journey that requires resilience, determination, and unconditional love. Mothers play a central role in their children's lives, facing extraordinary challenges as they seek to provide the best possible care amid the unique circumstances of these conditions, which is closely linked to the concept of primary maternal concern, where a mother's emotional fusion with her baby occurs. [13]

These mothers experience a sensitive state that gradually increases towards the end of pregnancy and continues for a few more weeks after the birth of their baby. In this state, the author says that the mother puts herself in her child's place, responding to their main needs due to her increased sensitivity, which can then be understood as preparation for the exercise of motherhood.[13]

Thus, due to this particularity, it becomes difficult for the *mother to* deal with her child's illness, because even if she is prepared to perform motherhood, her psychological state is not prepared to provide this assistance, because there is a difference between motherhood and maternity, the former is related to the bond of caring for and welcoming a child by a mother, while the latter refers to the consanguineous bond between mother and child[14] , refuting the idea that women are born with the instincts of motherhood and reinforcing that the maternal function is a process of construction.

The mother's role in adapting to her baby's needs is anchored in three main tasks: *holding* - which would be the representation of the act of holding the baby, referring to the security for the construction of this baby as a person, *handling* - which relates to the bodily care of the baby by its caregiver in processes such as bathing, changing diapers, feeding and among other things; and the presentation of objects - where the mother in the position of person who maintains contact with the social world, satisfies the function of presenting the world to the child and the child only world, being then, the good enough mother.[12-13]

Thus, the mother of a child with RBD would provide security to her child through *holding*, even during hospitalizations, characterized by a familiar face and voice capable of calming her child. In *handling*, she would take care of her child, being the main caregiver, giving her assistance, unlike the way she takes care of other children, such as performing bladder catheterization instead of just changing a diaper; bathing her in a hospital bed, where she could just be at home; watching her child being fed with the aid of bottles or feeding tubes, where she would like to be doing this task, maintaining breastfeeding contact.

In this way, this woman will fulfill her role in the baby's adaptation, so that even with the difficulty of mothering a child with an illness, she will be a good enough mother. [15]

Based on this thought, the act of caring implies losses in the life of the main caregiver, and these are related to changes in their social life due to the circumstances of the care demanded by their child's illness.[16] These changes include giving up their job, which ends up having a considerable financial impact, fluctuations in their marital relationship, as well as a lack of time to look after themselves and a reduction in their social relationships.

Caregivers are often overloaded with activities because they don't have family support and this makes it difficult for them to carry out their routine and care for their well-being.[17] Thus, with the mother caregiver in mind, it is very important to focus assistance not only on the children, but also on providing focused and humanized care to these mothers, especially when they discover the diagnosis of the disease and during the course of treatment.

Despite the challenges, mothering children with RD can also be a rewarding experience. Mothers of children with RD can develop unusual strength and determination. They can also learn to value life and appreciate the little things, so considering the worldwide emergence of RD, the aim of this study is to understand the mothering experience of women with children with RD who are dependent on continuous care.

The study is justified by the possibility of contributing to a better understanding of this unique and challenging experience, by seeking to understand the impacts that can be generated on a mother's health when caring for a child who requires special attention. The study can provide information about the challenges and rewards of mothering children with rare diseases, as well as the strategies mothers use to deal with these challenges.

METHOD

Design

This is a descriptive study, with a qualitative approach, based on the theoretical framework of social mobility and life stories[18], using the life narrative method, which addresses the conceptions of *les récits de vie,* or life stories, and which consists of an oral account of the journey lived, discarding external research and sociological explanations, thus aiming for sociological understanding. [19]

Location

The research was conducted using a web conference application, due to the fact that most of the participants lived in other states, and with only the researchers present, guaranteeing the mothers' privacy.

Population, eligibility criteria and sample

An open-ended interview was conducted with ten mothers of children with rare diseases who lived in Brazil between March and September 2022. The participants were selected using the *Snowball* technique, which consists of a non-probabilistic sample and is often used in social research, in which the first participants in the study constantly refer new participants until the expected goal is achieved.

This study included mothers of children with rare diseases, without specification, since there is a wide range of these diseases, and over the age of eighteen. On the other hand, the exclusion criteria were mothers who had a mental disorder that made it impossible for them to recount their lives.

The first interview was with a mother who had studied with the researchers and had abandoned her undergraduate nursing studies to move to another state in favor of better care for her son's rare disease. Finally, from the researchers' point of view, this triggered them to see the impact of rare diseases on motherhood and on the lives of these mothers, who take on the role of main caregiver. In this way, the first interviewee spread the word to other mothers who were experiencing the same reality, to generate a wider range of interviews.

The sample consisted of ten mothers of children with rare diseases, aged between 26 and 58, with the highest incidence of age in the 40s. The data collection instrument used was designed by the authors themselves based on the purpose of the study. It is a previously established form, made up of 29 questions to characterize the participants.

Ethical and legal aspects

This research was approved following the research guidelines CAAE - 51931421.8.0000.8157, in addition to following the ethical and legal aspects of Resolution No. 466/2012 and Resolution No. 510/2016 13.

The interviews were recorded with the consent of the participants, in a single meeting. They lasted between 30 minutes and 3 hours. The data collection period lasted seven months and ended when the saturation point was reached, when no new data was captured in the narratives. A single guiding question was used: tell us how you found out about your child's rare disease and your journey since then. The data was analyzed based on initial floating reading and grouping by similarity of the empirical material, allowing common themes to emerge.

RESULTS AND DISCUSSION

All the participants in this study have a high level of education, the most predominant self-declared color was white, most of the interviewees have the presence of the child's father and only half of them have an employment contract due to the routine of caring for their children.

Of the ten interviewees, four lived in Rio de Janeiro, two in Rio Grande do Sul, three in São Paulo and one in Curitiba. It is worth noting that four mothers had lost their children to the disease.

However, as mothers take on the role of main caregiver, they start to give new meaning to their self-care, given that only half of the participants routinely monitored their health. All the interviewees showed an attachment to spirituality to cope with and adapt to their children's condition. In addition, of the ten, three considered psychological support to be necessary.

There are also common themes in these women's accounts of their lives, such as the mourning of the perfect child after the diagnosis, added to the onset of depression, the abandonment by the father, friends and family, and the change of state for the sake of their child's treatment.

Among the rare pathologies of the children of the ten mothers interviewed were Metachromatic Leukodystrophy, Addison's Disease, Mucopolysaccharidosis, Angelman Syndrome, Edwards Syndrome, Congenital Adrenal Hyperplasia, Hereditary Spastic Paraparesis and Adrenoleukodystrophy. In this study, it was observed that most of these illnesses interfere with the children's activities of daily living, i.e., carrying out their basic living needs, requiring a caregiver.

As far as the caregiver is concerned, it is known that they play a fundamental role in the therapeutic itineraries of patients with rare diseases, since the illnesses leave those affected less autonomous and more dependent, requiring the caregiver constantly and it is the caregiver who will take them to the health services, evaluate their therapeutic preferences and take care of them on a daily basis, resulting in many cases in changes to their life plans in order to care for the affected person.[9]

In this study, most of the interviewees reported having given up their jobs and professional careers as soon as they took on the role of caring for their child, according to mother 1:

> "I'm the only one, my husband can't feed her, I'm the only one who does, she's totally dependent [...] I suffered a lot to quit my job, to this day it's my biggest dilemma. I really wanted to find another job, but the problem is finding someone to look after Juliana." [mother 1]

It is known that the majority of caregivers are women, because the responsibility of care is tied to the female figure due to sociocultural construction and when the child has a special need, the mother's responsibility is even greater.[20] Given this, it can be seen that mothers who have children with special needs become full-time caregivers, an often exhausting responsibility.[21]

Thus, when the responsibility for the care routine is focused directly on this woman, it can lead to numerous health problems, since it is an extremely tiring routine, and can even lead to a decline in her mental health, according to mother 10:

> "Looking after them is extremely tiring, people. Imagine having a child who has swallowing difficulties to feed breakfast, lunch, snack, dinner, all day long. Lorenzo only eats with me, you know? And someone must hold him down for me to be able to feed him, because he eats everything, he eats hamburgers, he eats pizza, he eats everything. He eats freely, he chews, he swallows, but he is not like a five-year-old who has nothing, right? He

> falls over, he does not have the motor skills to chew, and the food falls over a lot and he makes a mess, you know, it is difficult. So, my mother tells me to give it to him, so, psychologically, there is no way, apart from having it, you know? Having a child diagnosed, you still go through it, you know? It is like this, you become a caregiver and nurse for your own child twenty-four hours a day. So, I ended up, you know, having depression because of exhaustion, you know? Lorenzo is me all the time, to sleep, to eat, to go out, for everything, you know? So, I ended up having it, you know, and now it was depression, these pains, this fatigue, this excessive sleepiness, you know?"[mother 10].

In this sense, studies corroborate the results of this research, where the mother is the most involved in the process of caring for children with chronic illnesses, and her responsibilities increase, because in addition to domestic care, this woman starts to perform complex activities resulting from this assistance to her child and this generates an overload in her health, which can cause physical and mental illnesses.[22]

The care provided by the mother is indispensable, but there needs to be a socio-cultural deconstruction of the female figure as the one best suited to caring, through the establishment of a family support network, where care is delegated to other family members, such as the father, avoiding the overload of this woman.[23]

This imposition by society for women to be the main caregivers can be seen in the words of mother 3:

> "A child with special needs demands a lot of time, it's not just spending, it's time and the caregivers are us, the mothers, because this society that we live in today, it's... designates that the woman, she has to be the one who demands care for her children, that's not equality [...] Nobody sees this side, it's very difficult for society to see this side, then when the woman freaks out, when the woman wants to prioritize a little time for herself, which is to take care of herself, society still turns around and condemns her, you know?"[mother 3].

In relation to depression, the study points out that depressive symptoms are common even when the real baby corresponds to most of the mother's expectations, given that all the interviewees reported suffering on discovering the diagnosis, and the grieving process becomes more difficult when they are faced with the loss of the baby they kept alive inside.[24] As such, depressive symptoms are quite common following the discovery of the diagnosis. Having said this, it can be seen that the grieving process is necessary even when the real baby corresponds to the expectations set by the mother, according to the study described above.

Thus, the discovery of the diagnosis leads to the mourning of the idealized child and, as a consequence, depression, given that this happened to four of the ten participants in this study, according to mother 1:

> "I went into depression, like this... I just cried, so I started taking... sleeping pills and two more tranquilizers during the day, anxiety medication and another tranquilizer."[mother 1]

It should be emphasized that upon discovering the diagnosis, the interviewees claimed that they were born a new mother, forgetting their role as a woman, resulting in their annulment, as described by mothers 8 and 9, respectively:

> "So, this is... this phase in which you really need to be reborn, you're reborn because of the discoveries, you're reborn... because you become someone else, you become. It's the first phase where you forget, you start to forget there, all the changes start there, there you start to do so much for your child, there you start to... no more... then you used to do your nails every fortnight, you don't even remember that anymore, because you're searching, searching and when you're not searching so much, sometimes you don't do it either, because you find other things, you know?" [mother 8].
>
> "I was promoted from mother to caregiver mother, so this is already a first, a first change, right, so it's like you leave everything, you forget everything, you stay in this routine, which for me was years in the hospital." [mother 9]. [mother 9]

In this new phase, the mother rediscovers herself, since she is overburdened by the care demanded by children with special health needs (CRIANES), losing her leisure time, with her psychosocial care suffering numerous health risks due to less participation in society, resulting in her isolation due to the care of her child, bringing a great risk of social withdrawal.[25]

In one study, a high number of caregivers developed anxiety and depression as a result of this care routine.[22] The accumulation of activities, professional and family helplessness, social prejudice, the aggravations of the disease and neglect of health cause caregivers to become mentally ill, as well as reducing their quality of life. [26]

With regard to the interviewees' self-care, their resignification was noticeable, given that of the ten interviewees, only half of the mothers reported carrying out routine examinations every year. It is believed that living in an intense way, in order to accept the reality of their child's rare disease, results in the need to live intensely in the present, without thinking about what might happen in the near future, both in relation to themselves and to their child's prognosis, according to mother 1:

> "Once you've been diagnosed, you learn to live one day at a time, not to think about what's going to happen tomorrow. Living today, that is what brought me back to reality."[mother 1]

With regard to psychological counseling, only three of the ten mothers showed an interest in doing so, while the other seven reported not feeling the need, even though their vulnerabilities were implicit, since they carry scars from their child's illness. In this context, it was clear that all the mothers felt relieved and grateful to have been listened to when sharing their experiences with their children's rare diseases, and this only shows that if they had assistance focused on psychosocial care, their afflictions would be alleviated.

It is clear that in atypical motherhood, paternal, family and social abandonment is very common, due to the tireless routine of the mother who gives of herself day after day in treatment and care, which is painful for the mother, since she feels even more alone in this struggle, since they are her only forms of emotional support, also impacting on her mental health, as mothers 3 and 8 report:

> But it is very difficult for a father to understand that his wife must dedicate herself, especially since she's usually not the only child in the family, if she has another child, if she has siblings, even if they're healthy, that also requires care. So, she must... wear herself out with treatment and at the same time have to look after the house, she ends up not looking after herself, not prioritizing herself and living tired and they end up leaving. So, they do not abandon just because of a disabled child, they abandon because they feel abandoned, because that woman does not have any more time, not even for herself... (she pauses for a long time), you know? [mother 3]

> "You know when people come to my house? When I tell them tomorrow is their birthday and I am going to bake them a cake. Do you know when people come to my house? When, at the end of the year, they want to go to the beach house, they start to get closer... you do not have to come (shakes head as he speaks). I used to beg for it, but not today. So, come on, we're building our own network, it shouldn't be like this, it shouldn't, but you bring in other people who are experiencing the same things and there they see each other (smiling as she speaks), they start to love each other...as they're experiencing this too, they want this love, this affection."[mother 8]

Friends also tend to move away from the mother after the pathology is diagnosed, according to mother 1:

> "And whether they want to or not... friends end up distancing themselves (sad look, lowers head)... I think, I do not know if they are afraid of taking it out on us, us taking the problems out on them, right? Because whether they like it or not, it is an extra burden."[mother 1]

However, it is known that the greater the parents' support network, through family, friends, organizations, and financial help, they tend to cope better with the affliction of having an atypical child.[27] It should be noted that kinship and friendship play an important role in the lives of caring mothers. Relatives generally play a role of mutual, financial, and psychological help.[25]

Of the ten women interviewed, three had to leave their home state, leaving their homes, friends, family and, above all, their support network, which in most cases were their mothers, to get specific treatment for their child's condition. A study shows that when those affected by the disease and their families live in small towns, they must leave their hometown due to the lack of a referral center, and then move to larger cities.[28] In this way, the PNAIPDR still does not ensure effective care in the most varied states of Brazil. Therefore, care cannot be restricted only to those affected by rare diseases, since the family needs to be taken care of due to its restructuring to monitor treatment.[29]

The accounts of mothers who have had to move to another state further reinforce how alone these women feel in this struggle, without support or assistance aimed at their psychosocial state, as seen in Mother 1's account:

> "I think it's like this, here... I am very lonely, and my friends and family are all in Minas and they work all day and there's no one to talk to, I don't even come home at night, it's me and her. My husband comes home a lot at night, my husband comes home tired from work, and it is... I miss having someone to talk to, to talk to and I end up being alone, I end up thinking about the same thing all day long, with no one to talk to, it is really like that, isn't it? And talking on WhatsApp does not seem to be the same thing, there is a lack of that, I think, someone around to vent to, to talk to."[mother 1]

The attachment to spirituality to cope with and adapt to the condition of their child with a rare disease was visible in this study. In this way, faith has a positive and favorable impact on the mental health of caregivers, as it helps them reframe and cope with adversity, providing comfort and support.[30]

According to the interviewees, faith helps them to remain in the care process, as mother 2 says:

> "So, I think that any religion gives you a lot of support, you know? I think that believing in something greater gives you the security you need, right? Because it is a very fine line between life and death as a whole [...]"[mother 2]

In this study, of the ten interviewees, four went through the mourning of losing their child to the disease, reporting that it was one of the most difficult moments in their lives, according to mothers 3 and 5, respectively:

> "Ahh (sighs heavily when talking about the loss) yeah, I don't think any mother is prepared to lose a child and... I saw him in the last two years of his life wither away and... he couldn't take it anymore, his body couldn't take it anymore and I had to allow my son to leave, accept it, but after he leaves, after you see that they're gone and you'll never see that little eye looking at you again, that smile, smell that smell, you freak out [...]"[mother 3].
>
> "The death of a child is an amputation, you have to learn to live without that arm, that leg, you're never the same person."[mother 5]

Facing the death of a child is a challenge with psychoanalytical and social implications to consider, where each mother is psychologically shaken in some way, even when years have passed.[27-30] Mourning can be characterized as a slow and painful process, involving deep sadness, withdrawal from society and any activity that is not linked to thoughts about the child, leading to a loss of interest in the outside world. [27]

Finally, we can see how much these women give of themselves to their children, and they need to be cared for so that they can care for their children affected by the rare disease. It is up to nursing to take on the role of caring for the family with actions aimed at helping them to discover their potential, to facilitate the mother's care for herself and for the affected child, given that nursing has caring as its principle and can therefore suggest, recommend, and advise. It should be emphasized that this care should not be prescribed, which differs from other therapies, since a way of caring is not prescribed.[28]

CONCLUSION

The study showed that the objective was achieved, given that the interviews made it possible to understand and highlight the life narratives of mothers with children suffering from rare diseases, who are dependent on continuous care. In this way, the findings of this research show that the maternal figure is more involved in caring for atypical children, and thus tends to be vulnerable to psychological illness. Furthermore, through the interviewees' accounts, it was possible to identify their singularities and their real health needs through their feelings. Psychosocial nursing care for caregiver mothers is essential, as are public policies aimed at reducing the impact on these women's health, as well as ensuring the visibility of atypical motherhood.

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