''I was not an ordinary mother'': The difficulties of motherhood in Arthrogryposis Multiplex Congenita

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Ronaldo Antonio da Silva[1], Odaisna Maria da Silva Costa[2], Elizy Gabrielly da Costa Kaduch[3], Haustini Ruze Nonato Xavier[4] and Roseany Patrícia da Silva Rocha[5]

ABSTRACT

The chapter addresses Arthrogryposis Multiple Congenita, a rare disease, that is, which is infrequent in the population, which consists of the presence of multiple joint contractures that lead to limitation of limb movements. It is a disease that imposes many specificities on the life of the person and family, especially the mother who assumes the role of main caregiver. The main objective is to present the difficulties faced by the mother from the repercussions of Arthrogryposis Multiplex Congenita in the maternity ward. These are the results of a field research with a qualitative approach that had as a methodological reference the method of a single case study. The research was carried out with the mother of a child who had a clinical diagnosis of Arthrogryposis Multiplex Congenita, selected based on specific criteria. The fieldwork was carried out at the home of the participating mother and in-depth interviews and observation were used as techniques to collect empirical data. The narratives were submitted to thematic content analysis according to the stages proposed by Bardin. The research was submitted for consideration and approved by a Research Ethics Committee. The reading of the results will make it possible to know the main difficulties faced by a mother based on the repercussions of this disease in motherhood.

Keywords: Arthrogryposis, Multiple Abnormalities, Rare Diseases, Pediatric Nursing.

[1] Master of Science in Nursing
University of the State of Mato Grosso (UNEMAT)
[2] Bachelor of Science in Nursing
University of the State of Mato Grosso (UNEMAT)
[3] Bachelor of Science in Nursing
University of the State of Mato Grosso (UNEMAT)
[4] Bachelor of Science in Nursing
University of the State of Mato Grosso (UNEMAT)
[5] Master of Science in Nursing
University of the State of Mato Grosso (UNEMAT)

INTRODUCTION

In Brazil, the Ministry of Health (MH) considers as rare diseases those conditions that have a low prevalence in the population, that is, they have 65 cases per 100,000 people or 1.3 per 2,000 individuals (Brasil, 2014). However, despite this definition, the epidemiology of rare diseases in the country is unknown, due to the absence on the Ministry of Health's compulsory notification and health problems list (Silva et al., 2020a; Moro, 2020). It is estimated that there are about six to eight thousand different types of rare diseases in the world, in which for 95% of them, there is currently no effective treatment (Interfarma, 2013; Souza et al., 2019).

Among the rare diseases, Arthrogryposis Multiplex Congenita (AMC) will be highlighted in this chapter, a rare syndrome that consists of the presence of multiple joint contractures that lead to limited limb movements (Brasil, 2022). It was first described in 1841 by Adolf Wilhem Otto as congenital myodystrophy, and in 1923 it was titled AMC by Stern (Oliveira et al., 2021). Regarding epidemiology, a literature review highlighted that in every 10,000 births, three may be diagnosed with AMC, also indicating that in less developed countries this estimate may increase (Saccani, Umpierres and Basegio, 2008). In Brazil, there are still no epidemiological studies regarding the frequency of arthrogryposis in the population (Costa, 2021), although the condition is included in the list of priority congenital anomalies for birth surveillance in the country (Brasil, 2023). When analyzing anomalies that present limb defects, in which AMC is included, a recent epidemiological bulletin from the Ministry of Health highlighted that this group was more prevalent, with 25 individuals in every 10,000 live births (Brasil, 2023).

AMC is an uncommon syndrome of the musculoskeletal system, with multifactorial etiopathology, however, the exact cause is still unknown by science, which presupposes the relationship of genetic, parental, environmental factors and/or abnormalities during fetal development that affect the mother and the embryo, such as the influence of medications, infections, the condition of oligohydramnios or polyhydramnios, uterine alterations and chronic diseases and traumas (Oliveira et al., 2021).

The clinical diagnosis can be made soon after birth, as the child often presents stiffness and severe deformities in the joints, which can range from mild cases, with two or three affected joints, to severe cases, with deformities in important joints of all limbs, including the spine (Saccani, Umpierres and Basegio, 2008). In addition to the involvement in the joints of the limbs, the child may also present facial asymmetry, micrognathia, saddle nose, thin skin, muscle atrophy, scoliosis, degeneration in the nervous and urinary system, pulmonary hypoplasia, immature or short intestine, and osteoporosis of long bones (Oliveira et al., 2021).

With regard to treatment, in the case of congenital anomalies, such as AMC, it depends on early diagnosis, which is essential to provide the opportunity for individuals and their families to be

referred to access appropriate and timely treatments and interventions (Brasil, 2022). Specifically for AMC, the treatment has the perspective of improving joint function and, consequently, the person's quality of life, enabling autonomy in activities of daily living (Quintans, Barbosa and Lucena, 2017). For this, continuous monitoring with a multidisciplinary team is necessary, with indication of physiotherapy and orthopedic surgeries, in which the therapeutic plan is carried out on an individual basis (Quintans, Barbosa and Lucena, 2017).

Thus, considering the complexity of this syndrome, it is known that the diagnosis can directly affect the experience of motherhood, in view of the expectations that are built during the gestational period. A child diagnosed with a rare disease can cause changes in the family's daily life, with difficulties both with the community in which it is inserted and with health institutions that are often not prepared to serve them (Luz, Silva and Demontigny, 2016; Silva et al., 2020a).

In fact, studies show that these families face a lack of information about the disease, difficulty in accessing diagnosis and treatment, and also a lack of teams with qualified health professionals to meet their demands (Luz, Silva and Demontigny, 2016; Silva; Silva et al., 2020a; Silva et al., 2020b). Families may also be faced with the need to reorganize the roles of their members, who need to adhere to new responsibilities, in addition to those inherent to the daily needs of a child when they are born, they also need to seek social, health, financial and emotional services (Luz, Silva and Demontigny, 2016).

Thus, based on the problem presented and also the lack of research in the scientific literature with the perspective of approaching the experience of maternal care in rare diseases (Silva et al., 2020b), which can contribute with subsidies for the care provided by health professionals, including the nursing team, this chapter aims to present the difficulties faced by the mother from the repercussions of MCA in motherhood.

METHODOLOGY

This is a field research with a qualitative approach that has as a methodological reference the method of a single case study according to Yin's assumptions. The single case study, as a qualitative research method, refers to an experimental analysis, in which a current event is investigated, in depth and in its real-world situation, especially when the boundaries between the event and the set of circumstances cannot be clear and evident (Yin, 2015).

The present study was carried out with the mother of a child who had a clinical diagnosis of SMA and lived in a municipality in the interior of the state of Mato Grosso, Brazil. The case was identified through contact with the Municipal Health Department and selected considering compliance with the following inclusion criteria: being the mother of a child or adolescent with a clinical and/or genetic diagnosis of a disease that is classified as rare, that is, that affects 65 people in

every 100,000 inhabitants according to the Ministry of Health (Brazil, 2014); be eighteen years of age or older; reside in the state of Mato Grosso, to enable access by researchers during data collection; and being a user of the Unified Health System (SUS).

The fieldwork was carried out at the home of the participating mother and in-depth interviews and observation were used as techniques to collect empirical data. The open or in-depth interview is a conversation with intentionality that occurs freely, in which the researcher presents the purpose and meaning of the conversation and its enunciator pronounces freely on the topic addressed (Minayo, 2014).

Thus, two interviews were conducted with a total duration of 45 minutes and 64 seconds. The first was held at 09:00 am on February 9, 2023, lasting 20 minutes and 42 seconds; and the second at 08:50 am on February 14, 2023, lasting 25 minutes and 22 seconds. To apprehend the object investigated in this study, the following guiding question was used: 'what has it been like for you to experience motherhood in the face of AMC?'. However, in order to gradually deepen the memories of the participating mother, after the first interview, the following questions were elaborated: How was it for you to receive the news of the change in the exam in the 3rd month? What were the biggest difficulties faced? You get very emotional when you talk about the things he goes through at school. How did you prepare him for this moment? When he started studying, was there a conversation before? Did you talk about the *bullying* you might suffer? Did you receive guidance from health professionals about the things you would face in society? How is it cared for at home? Is his diet different from yours? What would you say to a mother who has just discovered this syndrome? What advice would you give? And at that moment, a cell phone was used to record the interview and enable the full transcription.

In addition, the observation technique is operationalized through the researcher's direct contact with the individual being interviewed, aiming to obtain more detailed information about the participant's reality (Minayo, 2014). And in order to complement the narratives obtained with the in-depth interview, the non-verbal communication of the participating mother and also the way she expressed herself when talking about the issues involving the child diagnosed with AMC were recorded.

For the organization of the empirical data collected, the material from the two interviews and the observation records were entered into a word file, which constituted the *corpus* of analysis of the research. This file was typed in *Times New Roman* font, size 12, spacing 1.5 cm and had 16 pages. This material was submitted to content analysis of Bardin's thematic type, following the stages of organization of analysis, coding, categorization and inference (Bardin, 2016). This process gave rise to themes, which were grouped together and resulted in two thematic categories, as highlighted in chart 1.

Table 1. Operationalization of thematic content analysis based on Bardin's assumptions, Mato Grosso, Brazil, 2023.

Grouped topics	Parent topic	Categories	Chapter
* Complications after birth *Facing the diagnosis *Support group *Uniqueness of the disease *Difficulty *Dealing with care *Treatment *Specialized professionals	Difficulties faced after birth and diagnosis of AMC	Daily care in the particularities of the rare disease	"I was not an ordinary mother": the difficulties of motherhood in Arthrogryposis Multiplex Congenita
*Bullying *School *Social judgment *Resignify the wheelchair *In the future * Be questioned	Fight against social judgment	Facing Social Judgment and Prospects for the Future	

Source: Prepared by the authors.

The research that originated this book chapter is linked to the matrix project entitled 'Rare mothers and narratives about motherhood', which was submitted for consideration by the Research Ethics Committee of the State University of Mato Grosso (CEP/UNEMAT) respecting the ethical precepts of research with human beings established in resolution No. 466/2012 of the National Health Council (Brazil, 2012). The aforementioned research project received a favorable opinion from the CEP/UNEMAT on May 18, 2022, under CAAE number: 57621722.3.0000.5166 and opinion 5.417.066.

The ethical commitment in this study was mediated through the signature by the participant and the coordinating researcher of the Informed Consent Form (ICF) in two equal copies, one copy remaining with the participating mother and the other with the coordinating researcher.

RESULTS

The case selected for investigation in this study refers to that of a 42-year-old mother, married, dental office assistant, graduated in Biological Sciences, post-graduated in the lato sensu modality in Health and Environment, municipal civil servant since 2005, living in her own home. She is the mother of three children, one miscarriage and two alive, in which one of the children, six years old, a regular student in public school, had the clinical diagnosis of the rare disease AMC.

From the analysis of this mother's narratives, the themes that emerged were grouped and gave rise to two thematic categories, namely: I. Daily care in the particularities of the rare disease, in which the interviewed mother reported the main difficulties she had to face to take care of her child; and II. Facing social judgment and perspectives for the future, which highlighted the coexistence with judgments, *bullying* and also the concern with the adolescence and adult life of the child.

DAILY CARE IN THE PARTICULARITIES OF RARE DISEASES

In the care experience of the interviewed mother, the reports highlighted that there was knowledge about the bone malformation that the child presented during pregnancy. And for this reason, the mother's expectation already considered the birth of the child with such a complication. However, receiving the diagnosis of the rare disease was a difficult moment, enhanced by the lack of knowledge about AMC, as she presents in the narrative: *"It was difficult. We received the diagnosis, it was a very complicated phase for us. Because we don't know what to expect, we don't know what will happen. And when he was born, I was prepared to receive a child with crooked feet and crooked hands, but that's all, you know? only this to solve (Atypical Mother)".*

In the participant's mother's report, some points were highlighted in relation to the complications that the child presented after birth. Fractures, a common complication in AMC, are directly related to the particularities of this rare disease, whose main characteristic is bone fragility. And so, immediately after birth, the child had to be admitted to the Intensive Care Unit (ICU) as a result of the fractures, as the mother recalled: *"The hardest thing to face was that he broke his arm, broke his leg and went to the ICU. So, that was the phase I think was the most difficult to face, to see him [...] with that cast that caught on his waist (Atypical Mother)".*

Another particularity of the rare disease that was highlighted involved muscle strength and was perceived and recognized by the atypical mother over time. In the interview, for example, she narrated that at school, food needs to be cut into smaller sizes, to enable the child to chew properly: *"The only difference I talk about at school is that he can't chew hard things, he doesn't have as much strength in his mouth. He doesn't open his mouth very big, so it's the chewing part. You put everything we eat, but in small pieces, so he doesn't suffer so much to chew (Atypical Mother)".*

To deal with the child's care on a daily basis and to face the particularities that the diagnosed rare disease had, the mother reinforced the importance of having been inserted in a support group, formed specifically by families who shared the experiences of care at AMC. According to the narratives, this group helped to clarify several doubts: *"It helped a lot, it certainly helped. All the doubts we had about surgeries, with the clubfoot, with the boots we had to wear, the surgeries, everything there we answered the questions, we always ask "did your son have this? Did you have that?" I was exchanging ideas and it made it a lot easier. So today we help others who are arriving, who have a lot of cases, some new ones are arriving and now we are helping (Atypical mother)".*

Among the difficulties that the mother had to face to take care of her son, there was also a concern with the social treatment he would receive, in view of the particularities of the disease. In this experience, the mother understood that the unusual would not be treated as common: *"At first it was very difficult like that, because I would like them to treat it as common. But, how are they going to treat common when it is not common. So, the crooked hand draws a lot of attention, and people*

often say: "ah, but you don't have a little ortho, right? to tidy it up". As if we had never done anything, you know? And orthosis is what he has! Physiotherapy is what he has had all his life! But the lack of musculature leads the hand to become crooked, it just can't have deformities, but the little hand is crooked (Atypical mother)".

One of the daily care that the child with AMC needs to perform includes physiotherapy sessions. In the mother's experience, the narratives presented as a difficulty the situation of witnessing the child's suffering during the physiotherapy sessions, because the child often felt pain: *"He needs to face the pain because otherwise things get even uglier. So, he faces it, there are days when he is cool, there are days when he cries a lot, there are days when he questions. There were days when he talked to me with the physiotherapist there with a swollen eye asking why he suffers so much. And at this moment, for the mother it is difficult, the mother cries together, right? and then I welcome (Atypical Mother)".*

Due to the clinical evolution of AMC, there are influences on the growth and development of some body systems and thus, the child may need therapeutic interventions in the future. The mother reported some alternatives that were presented to her son, and that can be performed later: *There are some cases in Arthrogryposis that the doctor can transfer the muscles, for example, if he finds a muscle here [showing] in the pectoralis major, he can put this muscle in his little arm then he bends his arm. So, there are also orthopedists who work with this. [...] Today there is also bone stretching that the orthopedist can do, because of the stature. Because they are usually very short. They can increase the child's stature to be a little taller, but everything is in the future. The hand that is turned, right? the orthopedist can also tidy up his hand, when the child decides, that's the child's decision (atypical mother)".*

In addition to these confrontations presented, the mother also had to locate specialized professionals who were committed to the rare situation experienced by the family. Thus, the atypical mother highlighted the attention she had when considering the physiotherapist who would perform the physiotherapy sessions: *"He doesn't need physiotherapy more or less, because there is a professional who gives you physiotherapy more or less. You have to have a committed professional who looks at your child, who is committed to his life and does his best (Atypical mother)".*

FACING SOCIAL JUDGMENT AND PROSPECTS FOR THE FUTURE

When experiencing the child's illness due to a rare disease, which presents changes in the physical body and that are easily perceived, dealing with people's judgment from social life was one of the issues that the mother had to face in motherhood. Because it is a child, memories about the manifestations of social judgment were frequently experienced at school. In this regard, the interviewed mother reported a situation experienced in this social space that involved *bullying*: *"Last*

year the teacher said to me like this: "ah! Did you see what happened? That the little boy is imitating him?". He was walking down school and there was a boy imitating him. Then she said that she had already called the child, talked about how he should behave, that he needs to respect the other, and then I told the principal: "what you did was very beautiful, it must be done so that the children learn to respect each other. But it's just the beginning! Unfortunately, he will go through this, it's just the beginning! We are not used to what is different, none of us" (Atypical mother)".

Also in this regard, when narrating the difficulties that her son faced daily at school, the mother was very moved. According to her, in addition to *bullying*, the child also needs to deal with the repercussions of the particularities that the rare disease imposes, which influence, for example, recreational activities: *"But like, it's a challenge! School I think [that] will always be a challenge. Because he can't like, if he's at recess, the other kids are there running, playing, right? And he's looking. Because he needs someone to go with him. And there at that time, there is no one. He doesn't have a mother, he doesn't have a father, but he goes with the teacher, when he can eat faster he goes [gets emotional]. But he can't always eat fast, because the muscles in his mouth are also more fragile [crying]. But then, the school (Atypical Mother) is challenging".*

From the mother's narratives, it was possible to verify that the social judgment experienced permeated the school space: *"Then it is difficult to look at them. It was difficult for people to understand that he has a normal mind, that he has to be treated like a normal person. Because he wants it. And it was hard for me. There are times when tears have fallen. To this day, but it's a day-to-day challenge (Atypical Mother)".* One of the concerns expressed by the mother was that her son would need to use a wheelchair, in view of the potential that this object would have in the social judgment that her son was already experiencing: *"For me it was one of the things that I did not accept, because I believe that all people have that schism like this. You don't even sit in a wheelchair, have you seen that? Don't even sit down, for God's sake! Because this is an unacceptable thing in our life, right? As if it were an inadmissible thing, no one could be there (atypical mother)".*

In addition, another confrontation pointed out by the mother was concerns about the child's future. In the following narrative, the atypical mother described her concerns about her son's adolescence and also the way she dealt with this issue: *"Today he [is] very full of joy. But he's a child, he doesn't understand much yet. So, [as] a teenager, we don't know how he's going to deal with it. But it's living one day at a time! [...] I think so, the biggest difficulty is this, living one day at a time, because we think a lot ahead, what will my son go through in the future? How will it be? So living one day at a time is the most extraordinary thing there is (Atypical Mother)."*

DISCUSSION

The results described the difficulties faced by the mother when experiencing motherhood in the face of a rare disease. In fact, the scientific literature corroborates the difficulties faced, as it points out that the birth of a child with a rare disease in the family context can lead to changes in the family's daily life, giving rise to unique feelings and also the need to (re)organize behaviors to face the demands arising from the new condition that presents itself (Luz, Silva and Demontigny, 2016; Silva et al., 2020a).

In the case of rare diseases, reaching the diagnosis can be one of the main difficulties to be faced by the family (Silva et al., 2020a). Generally, children can take many years to receive the correct diagnosis, whether clinical or genetic. This reality is influenced by the lack of knowledge of health professionals about rare diseases and their peculiarities, the large number of rare diseases cataloged in the world and the variety of clinical manifestations, the existence of few reference services for rare diseases in the country, the high cost of genetic tests necessary to reach the diagnosis accurately and also, the reduced number of physicians specializing in genetics (Luz, Silva and Demontigny, 2015; Luz, Silva and Demontigny, 2016; Silva et al., 2020a).

However, in the experience of the mother investigated in this study, the clinical diagnosis was made immediately after birth. This is a situation that results from the characteristics that the disease presented in the child's physical body. In AMC, joint contractures are often manifested in the lower or upper limbs, and this is noticeable and contributes to the clinical evaluation of doctors and the moose of diagnosis (Brasil, 2022). However, a study pointed out that this does not always happen, because even the child with Epidermolysis Bullosa being born with cutaneous aplasia, a rare dermatological disease, health professionals had difficulties in diagnosing it, according to the mother's memories and narratives (Silva et al., 2020a).

Another confrontation is related to the clinical evolution of the diagnosed rare disease and the respective complications manifested, which will directly influence the family's confrontations. In the experience of the interviewed mother, the ICU admission soon after birth and the decreased muscle strength were related to the particularities of the clinical evolution of the disease. In the case of AMC, due to the absence of muscle mass, strength is limited in children who receive this diagnosis and this causes the functionality of the lower limbs to be impaired, especially in relation to gait (Rozane, Carvalho and Ruzzon, 2012). In addition, another characteristic of this condition is joint stiffness, which has an extra-articular influence as a result of the shortening of the muscles and the retraction of the joint capsules (Carvalho and Santos, 2008).

According to the above, another particularity of the AMC is in relation to the musculature, which is rigid, including the facial muscles, causing the individual to have difficulty in feeding (Saccani, Umpierres and Basegio, 2008). Thus, in SMA, the person may manifest difficulty

swallowing (dysphagia), with narrowing of the temporomandibular joint, which often presents itself as an aggravating factor. When associated, these oropharyngeal pathologies can make it impossible to carry out safe oral feeding of the newborn, due to the risk of aspiration (Oliveira et al., 2021).

Thus, in view of the diagnosis of a rare disease and the repercussions imposed by the complications manifested in the child, it is important to integrate the family into a support group, formed mainly by families that share the same anxieties and concerns. These groups are recognized as supply strategies for the family caregiver, offering emotional and psychological support, as well as providing information and guidance in relation to aspects of the diagnosed rare disease (Rodrigues et al., 2021). For the group to be more effective, it is necessary to create an integral space, with the purpose of establishing reciprocal exchanges between the participants, about the distressing and happy experiences of daily life, as well as expressing and sharing ways to alleviate them (Rodrigues et al., 2021).

However, in addition to these confrontations related to the clinical aspects of the rare disease, there are others that involve social issues. The results showed that the mother was concerned with social life, in which the child would be exposed to social judgment, which could permeate different spaces and be enhanced by the use of wheelchairs in the future. In this sense, it is understood that rare diseases have social consequences for patients and their families, with psychological and economic involvement, affecting all members (Catana, 2013). Negative social effects for these families can include isolation of the person diagnosed with the rare disease, who is subject to stigmatization, discrimination, and reduced educational and employment opportunities (Catana, 2013).

Another aspect that stood out in the narratives of the mother participating in the study is related to coping *with bullying* and recreational activities at school. The term *bullying* has English origins and designates actions of mistreatment against another person, whether physical, psychological, or both (Silva, 2017). It is a theme that is often present in debates held in school environments, a space for building personal relationships and training (Ribeiro, 2023). Due to the medium and long-term consequences, it is important to provide spaces for discussion, especially on ways to cope with *bullying* in schools, recognized as a space that is also favorable for the occurrence of conflicts (Ribeiro, 2023).

The family and the school function as two essential organizations to motivate the child's development processes, operating as stimulators or intimidators of their intellectual, affective, and social growth (Freitas, Freitas, and Cavalcante, 2021). Both share functions associated with education, social skills, and political thinking and behavior (Freitas, Freitas, and Cavalcante, 2021). Therefore, they help and influence the child's education, being responsible for the transmission of values and knowledge (Lazzaretti and Freitas, 2016). Thus, family and school can work together with

children on the inclusion of people with disabilities, to foster the feeling of belonging to the environment in which the child who has a disability is inserted (Lazzaretti and Freitas, 2016).

In addition to this, the mother needs to frequently witness the emotional suffering of the child. In the experience shared in this study, during the physiotherapy sessions, the mother witnessed her child's pain. Specifically in the case of AMC, pain is strongly related to the need to perform physical therapy to improve the quality of life of the person diagnosed with the syndrome (Rozane, Carvalho and Ruzzon, 2012). This result highlights the discussion about the feeling of helplessness that mothers may feel, as occurred in a study that showed that, when witnessing the pain of the infant daughter during the dressings, necessary due to the diagnosis of a rare dermatological disease, the mother's feeling was that of not being able to do anything to relieve it (Silva et al., 2020a).

Still in this treatment scenario, the mother faces concern about the therapeutic interventions that the child will be subjected to in the future and also about the adolescence phase. It is known that over time, health-related technologies evolve more and more and as this advance happens, new procedures and therapeutic alternatives emerge, including for AMC (Costa, 2021). For example, malformations of the limbs that limit functions are common, and are often treated with surgery. Treatment on the feet is the most necessary, followed by procedures on the knees and hips. Surgeries in the upper limbs are suggested to correct fixed joint contractures, the elbows are the most approached in the upper limbs. The long-term purpose is to increase joint mobility and muscle strength and provide adequacy for gait acquisition and improvement in the quality of life of people living with AMC (Costa, 2021).

Finally, when considering the difficulties faced in daily life, the results highlighted the mother's search for professionals committed to the particularities that AMC requires. It is noteworthy that in the case of illness due to a rare disease, the family often makes the move to seek qualified professionals to complement the care (Luz, Silva and Demontigny, 2015). In the case of AMC, treatment needs to be monitored by a multidisciplinary team on a regular and cautious basis, due to the various complications that the syndrome can present, the multiple joints that can be affected, and the particularities of recurrences over the years, as contractures and stiffness of periarticular tissues make it impossible to perform decisive osteoarticular remodeling (Oliveira et al., 2021).

In fact, the literature points to difficulties that families face in their relationship with health services and professionals. This occurs due to the lack of knowledge of health professionals about the specificities of rare diseases (Luz, Silva and Demontigny, 2016; Silva et al., 2020b). However, it is with health services and professionals that families who experience illness in the face of rarity will inevitably be linked for a long period (Luz, Silva and Demontigny, 2016; Silva et al., 2020b). However, in these spaces, families are seen unequally, not precisely because of prejudice (Luz, Silva and Demontigny, 2016). It is emphasized that the rights related to access to quality health services,

equality, problem-solving capacity and comprehensiveness in conduct are not always respected, either because the services do not have technological resources to attend to a rare condition or even because health professionals are not adequately prepared for this reality (Luz, Silva and Demontigny, 2016; Moro, 2020).

FINAL CONSIDERATIONS

The results of the study allowed us to approach the difficulties experienced in the maternity ward of an atypical mother. When she had to deal with the unusual in the maternity ward, this mother was faced with different difficulties, which were related to the process of diagnosing the rare disease, the complications and treatments to which the child was submitted, the search for health professionals committed to the peculiarities of the MCA, the repercussions of the disease on social life and also, concerns with future therapies and the adolescence phase.

Thus, the difficulties experienced in the daily life of this mother present relevant issues that need to be considered by political agents who work at the federal, state and municipal levels, as well as the managers of different health services and society. It is necessary to invest in scientific research and discussions on different rare diseases, offer training for health professionals, aiming to prepare them to diagnose early and/or refer children to specialized services, increasing the potential of therapeutic treatment and reducing sequelae, which have a negative impact on the quality of life of the person, family and also, imposes an increase in costs for health care.

Despite the relevant results, this study has limitations regarding the investigation of the reality experienced by a mother about the specific rare disease AMC. Thus, in order to mitigate this, new studies are encouraged, which should consider the daily lives of other atypical mothers, when experiencing illness and care for different rare diseases and in similar or different geographical contexts, corroborating the endorsement of the scientific literature on this theme.

REFERENCES

1. Bardin, L. (2016). *Análise de conteúdo: edição revista e ampliada* (3ª reimp. da 1ª ed.). São Paulo: Edições 70.

2. Brasil. Conselho Nacional de Saúde. (2012). Resolução no 466, de 12 de dezembro de 2012 - diretrizes e normas regulamentadoras de pesquisas envolvendo seres humanos. Disponível em: http://conselho.saude.gov.br/resolucoes/2012/Reso466.pdf. Acessado em: 2 abr. 2023.

3. Brasil. Ministério da Saúde. (2014). Portaria nº 199, de 30 de janeiro de 2014 - Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras. Ministério da Saúde. Disponível em: http://bvsms.saude.gov.br/bvs/saudelegis/gm/2014/prt0199_30_01_2014.html. Acesso em: 13 jun. 2023.

4. Brasil. Ministério da Saúde. Secretaria de vigilância em saúde e ambiente. (2023). Boletim epidemiológico. Vol. 54, nº 3, fev. 2023. Disponível em: https://www.gov.br/saude/pt-br/centrais-de-conteudo/publicacoes/boletins/epidemiologicos/edicoes/2023/boletim-epidemiologico-volume-54-no-03. Acesso em: 26 out. 2023.

5. Brasil. Ministério da Saúde. Secretaria de Vigilância em Saúde. Departamento de Análise Epidemiológica e Vigilância de Doenças não Transmissíveis. (2022). *Guia prático: diagnóstico de anomalias congênitas no pré-natal e ao nascimento*. Disponível em: https://www.gov.br/saude/pt-br/assuntos/saude-de-a-a-z/a/anomalias-congenitas/publicacoes/guia-diagnostico-de-anomalias-congenitas-no-pre-natal-e-ao-nascimento. Acessado em: 24 ago. 2023.

6. Carvalho, R. L., & Santos, C. E. (2008). Efeito da imersão associada à cinesioterapia na artrogripose. *Revista científica UNIFAE*, 2(1), 10-14. Disponível em: https://silo.tips/download/efeito-da-imersao-associada-a-cinesioterapia-na-artrogripose. Acessado em: 05 mai. 2023.

7. Catana, F. L. S. R. (2013). *A Doença Rara na Família e na Escola: a perspectiva parental*. Dissertação de mestrado, Universidade Católica Portuguesa, Viseu, Portugal.

8. Costa, M. (2021). *Caracterização clínica, morfológica e molecular de pacientes com artrogripose múltipla congênita*. Tese de doutorado, Universidade Federal da Bahia (UFBA), Salvador, Bahia, Brasil.

9. Freitas, M. C., Freitas, B. M., & Cavalcante, G. F. (2021). A importância da escola para crianças em contexto familiar monoparental. *Ensino em Perspectivas*, 2(1), 1-13. Disponível em: https://revistas.uece.br/index.php/ensinoemperspectivas/article/view/4536. Acessado em: 20 abr. 2023.

10. Interfarma. Associação da Indústria Farmacêutica de Pesquisa. (2013). *Doenças Raras - Contribuições para uma Política Nacional*. Interfarma. Disponível em: http://www.sbmf.org.br/pdf/biblioteca/doencas_raras_2013.pdf. Acesso em: 8 maio. 2023.

11. Lazzaretti, B., & Freitas, A. S. (2016). Família e escola: contribuindo para o processo de inclusão escolar de crianças com deficiências. *Caderno Intersaberes*, 5(6). Disponível em: https://www.cadernosuninter.com/index.php/intersaberes/article/view/376. Acessado em: 11 jul. 2023.

12. Luz, G. S., Silva, M. R. S., & Demontigny, F. (2016). Necessidades prioritárias referidas pelas famílias de pessoas com doenças raras. *Texto & Contexto Enfermagem*, 25(4), 1-9. https://doi.org/10.1590/0104-07072016000590015

13. Luz, G. S., Silva, M. R. S., & Demontigny, F. (2015). Doenças raras: itinerário diagnóstico e terapêutico das famílias de pessoas afetadas. *Acta Paul Enferm*, 28(5), 395-400. https://doi.org/10.1590/19820194201500067

14. Minayo, M. C. S. (2014). *O desafio do conhecimento: Pesquisa qualitativa em saúde* (14ª ed.). São Paulo: Hucitec.

15. Moro, R. W. (2020). *Doenças raras e políticas públicas: entender, acolher e atender*. São Paulo: Matrix.

16. Oliveira, D. K., Fernandes, B., Anjos, A. A., et al. (2021). Artrogripose Múltipla Congênita: relato de dois casos. *Medicina (Ribeirão Preto)*, 54(2). https://doi.org/10.11606/issn.2176-7262.rmrp.2021.173816

17. Quintans, M. D. S., Barbosa, P. R., & Lucena, B. (2017). Artrogripose congênita múltipla. *Rev. Ped. SOPERJ*, 17(3), 23-27. Disponível em: http://www.revistadepediatriasoperj.org.br/detalhe_artigo.asp?id=1025. Acessado em: 20 ago. 2023.

18. Ribeiro, C. A. U. (2023). *Bullying na escola: uma análise sobre os documentos de bases nacionais*. Trabalho de Conclusão de Curso, Universidade Federal de Goiás (UFG), Goiânia, Goiás, Brasil.

19. Rodrigues, B. D., Martins, L. F., Barbosa, L. C., & Xavier, M. G. S., et al. (2021). A importância da rede de apoio ao familiar de pessoas com deficiência, doenças crônicas e raras. *ANALECTA-Centro Universitário Academia*, 7(2). Disponível em: https://seer.uniacademia.edu.br/index.php/ANL/article/view/3122. Acessado em: 12 ago. 2023.

20. Rozane, J. M. S. G., Carvalho, R. P., & Ruzzon, D. V. L. (2012). Fortalecimento muscular na artrogripose múltipla congênita. *Fisioterapia Brasil*, 13(3), 226-230. Disponível em: https://pesquisa.bvsalud.org/portal/resource/pt/lil-764355. Acessado em: 14 out. 2023.

21. Saccani, R., Umpierres, C. S., & Basegio, C. (2008). Artrogripose Múltipla Congênita: um relato de caso. *Revdig. Buenos Aires*, 116. Disponível em: https://efdeportes.com/efd116/artrogripose-multipla-congenita.htm. Acessado em: 15 out. 2023.

22. Silva, B. A. G. (2017). *Bullying: a violência nas escolas de ensino fundamental I educando para a paz*. Trabalho de Conclusão de Curso, Faculdades Atibaia (FAAT), Atibaia, São Paulo, Brasil.

23. Silva, R. A., Santos, R. E. S., & Alencastro, L. C. S., et al. (2020a). A vivência do cuidado materno a uma lactante com Epidermólise Bolhosa. *Revista de Enfermagem do Centro-Oeste Mineiro*, 10, e4133. https://doi.org/10.19175/recom.v10i0.4133

24. Silva, R. A., Souza, S. P. S., Bernardino, F. B. S., & Alencastro, L. C. S. (2020b). Cuidado familiar à criança e ao adolescente com Epidermólise Bolhosa: uma revisão integrativa da literatura. *Rev Baiana Enferm*, 34, 1-13. https://doi.org/10.18471/rbe.v34.35781

25. Souza, I. P., Androlage, J. S., Bellato, R., & Barsaglini, R. A. (2019). Doenças genéticas raras com abordagem qualitativa: revisão integrativa da literatura nacional e internacional. *Ciência & Saúde Coletiva*, 24, 3683-3700. https://doi.org/10.1590/1413-812320182410.17822019

26. Yin, R. K. (2015). *Estudo de Caso: Planejamento e métodos* (5ª ed.). Porto Alegre: Bookman.