Cogitare Enfermagem

ORIGINAL ARTICLE

CARE PRACTICES FOR PEOPLE WITH HUNTINGTON'S DISEASE FROM THE PERSPECTIVE OF FAMILY CAREGIVERS

HIGHLIGHTS

1. Lack of knowledge of rare diseases interferes with care practice.
2. Care requires adaptations to the environment and routine.
3. Difficulty in diagnosis leads to proactivity on the part of the family member.

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ABSTRACT

Objective: to learn about care practices for people with Huntington's disease. **Method**: a qualitative study using the Patient- and Family-Centered Care framework, carried out with 20 family caregivers of people with Huntington's disease. Data was collected through semi-structured interviews, in February and March 2022, via Google Meet, and after being transcribed in full, it was submitted to Content Analysis. **Results**: The practice of caring for people with Huntington's requires adapting to the environment, readjusting the routine, and improvising aids. The difficulty in obtaining a diagnosis makes family members proactive in their search for knowledge to improve care conditions. **Conclusion**: although this is a rare disease, the care actions mentioned are like those carried out for people with other chronic diseases. However, recognizing the care practices carried out by family members can help nurses plan their care.

KEYWORDS: Huntington Disease; Rare Diseases; Family; Health Care.

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INTRODUCTION

Rare diseases, 80% of which originate genetically, affect around 65 out of every 100,000 people in Brazil and constitute a public health problem. They are characterized by a disparity of signs and symptoms, which makes diagnosis difficult; they are progressive and degenerative; and they have a direct impact on quality of life[1-2].

To provide a legal and political framework for the care of people and families living with rare diseases, in 2014 Ministerial Order 199 was approved, establishing the National Policy for Comprehensive Care for People with Rare Diseases (PNAIPDR, in Portuguese). This aims to organize the Health Care Network (RAS, in Portuguese), provide access to prevention, reception, diagnosis, treatment, and support until resolution, follow-up, and rehabilitation; access to resources, diagnostics, and treatments; and access to information, care, and genetic counseling, when indicated[2].

A study carried out in Spain showed that families of people with rare diseases are strongly affected and face needs that are different from those associated with the most common chronic diseases[3]. Among the needs identified by families as priorities are access to health and social services, knowledge about rare diseases, specialized social support structures, acceptance, and social integration, and preservation of personal and family life[3].

Even more specific aspects arise when looking at one or other rare disease, such as Huntington's disease (HD). HD is a neurodegenerative, autosomal dominant hereditary disease characterized by psychiatric, cognitive, and motor symptoms. Its progression destroys the cerebral cortex, which leads to deterioration in cognitive ability and the onset of dementia[4].

The neurological manifestations of HD begin insidiously and include dysarthria, dysphagia, postural instability, dystonia, urinary incontinence, and choreics movements. As the disease progresses, psychiatric symptoms occur, characterized by personality and mood changes, agitation, depression, impulsivity, and, in some cases, aggression[5].

The incidence of HD varies from three to 10 individuals per 100,000 in Europe and North America. In Brazil, there are still no official statistics, but it is estimated that there are between 13,000 and 19,000 carriers of the gene[6].

In terms of the progression of the disease, people with HD lose their ability to carry out Instrumental Activities of Daily Living (IADLs), such as cooking, tidying the house, taking care of household finances; and carrying out Activities of Daily Living (ADLs), such as taking care of their hygiene. Therefore, the affected person becomes dependent on the family, causing changes in the family's routine due to the progressive physical and emotional strain caused by providing daily care for these individuals[7].

Given this context of care, the Patient and Family Centered Care (CCPF, in Portuguese) approach to care planning for people with chronic conditions is based on mutually beneficial partnerships between health professionals, patients, and families[8]. Thus, studies carried out with family members of people with HD that address their needs in the care process can contribute to building knowledge concerning identifying points of support and weaknesses in the care process.

This study aims to find out about care practices for people with Huntington's disease.

METHOD

This is a descriptive, exploratory, qualitative study based on the theoretical framework of Patient and Family Centered Care[8]. The construction of the study report followed the recommendations set out in the Consolidated Criteria for Reporting Qualitative Research (COREQ). The research was carried out at a national level, in partnership with the Brazilian Huntington's Association (ABH, in Portuguese)[9], which helped to publicize the research project to family members of people with HD registered with this organization in Brazil.

The inclusion criteria were family members of people with HD who provided direct care, were over 18 years old, and had contacted the main researcher expressing their interest in taking part in the study. In turn, family members who had also been diagnosed or screened for HD were excluded.

Data collection took place between February and May 2022. It began with the survey being publicized on social media (Instagram, WhatsApp) and by the ABH to people registered with the association, using an open letter with information about the survey. Next, those interested in taking part in the survey were sent a link (https://docs.google.com/forms/d/e/1FAIpQLSeKt_2MIqkaCVsSfo767PB4p9ili9735bLDlDAp9cxfD3MOTg/viewform), which led to a survey form (Google forms) with questions about the inclusion and exclusion criteria. Once the inclusion criteria had been met, they were directed to the Informed Consent Form and informed that if they agreed to take part in the study, they should fill in their e-mail address and cell phone number, so that we could contact them and send them the ICF via e-mail, signed by the researchers.

Next, those who met the criteria and expressed an interest in taking part in the research were contacted via WhatsApp to schedule an interview, depending on the participant's availability. On the same occasion of contact via WhatsApp, a link was sent directing the participant to fill in a questionnaire via Google Forms, prepared by the main researchers, to characterize the family caregivers taking part. To this end, the following variables were used: gender, age, marital status, schooling, family income, history and degree of kinship with people with HD, time since diagnosis, and most frequent symptoms.

After the research was announced, 40 people contacted the main researcher by telephone or e-mail, expressing an interest in taking part in the study. However, after three attempts to call back, 12 did not answer, five were not available for an interview and three did not meet the inclusion criteria because they did not live with the family member with HD daily. The study therefore involved 20 family members of people diagnosed with HD.

The interview was conducted using the Google Meet platform or a video call via WhatsApp, according to the participant's preference, and included the following support question: "What care practices do family caregivers use for people with Huntington's Disease?". In addition, support questions were prepared which covered how the family caregiver prepares to carry out care actions daily, how these procedures are carried out, and what factors hinder and/or facilitate them. Participants were asked to open a camera to promote acceptance; however, the recording was audio only. They were conducted by an undergraduate nursing student who had trained in the interview technique with members of the research group. In addition, after the first interview, the student presented the transcript, which she read without any need for adjustments. The main researcher had no previous relationship with the interviewees.

The interviews lasted an average of 40 minutes and were then transcribed in full; the data was submitted to Content Analysis, which followed the stages of pre-analysis, exploration of the material, treatment of the data obtained, and inference[10]. The transcribed interviews were not presented to the participants for validation but were submitted for evaluation by the study supervisor to check for possible mistakes in the analysis, which were not verified. A total

of 73 codes were obtained which gave rise to 13 nuclei of meaning, which were grouped, considering the poles of attraction of the communication, and formed two categories.

The results were interpreted and discussed in the light of the central assumptions of the CCPF: dignity and respect; sharing information; family participation in care and decision-making; and collaboration between patients, families, health professionals, and managers in the development of public policies, professional education, and research, as well as in the provision of care[8].

The research followed all the ethical precepts laid down in current legislation and was approved by the Research Ethics Committee under opinion no. 5.247.124.

RESULTS

The interviewees' characteristics are shown in Chart 1. Concerning schooling, 12 reported having completed higher education, four incomplete, and four completed high school.

Chart 1 - Characterization of HD family participants. Campo Grande, MS, Brazil, 2023.

Interviewee	Gender	Degree of kinship	Age	Marital Status
E1	Female	Son	50	Divorced
E2	Female	Son	32	Single
E3	Female	Son	54	Stable union
E4	Female	Wife	28	Single
E5	Female	Uncle	32	Married
E6	Female	Wife	59	Stable union
E7	Female	Wife	43	Married
E8	Female	Son	49	Married
E9	Female	Son	46	Married
E10	Female	Son	29	Stable union
E11	Female	Mother	70	Single
E12	Female	Son	33	Single
E13	Female	Son	29	Single
E14	Female	Wife	42	Married
E15	Female	Son	31	Married
E16	Female	Son	43	Stable union
E17	Female	Son	41	Married
E18	Female	Son	41	Single
E19	Male	Son	19	Single
E20	Female	Wife	34	Married

Source: The authors (2023).

The states of residence of the participants were: Distrito Federal, Minas Gerais, Alagoas, Mato Grosso do Sul, Rio de Janeiro, Espírito Santo, Piauí, Rio Grande do Sul, Bahia, Goiás,

and São Paulo, where five of the 20 individuals lived. Most of the participants who had been diagnosed with HD were grandparents (n=nine; 40.9%), but they also had other family members such as mothers, fathers, children, and nephews with the same disease.

After analyzing the data, the nuclei of meaning were identified and grouped into two categories: ***"Care practices carried out by family caregivers of people with HD",*** *and* ***"Lack of knowledge about HD and its implications for care practices".***

Care practices carried out by family caregivers of people with HD

According to the reports of the family participants, the care practices and strategies adopted were in line with the demands presented and influenced by the progression of the disease.

During lunch, I started to notice that when she ate the beans, she choked a lot and we decided to grind them up and make bean porridge. (E17)

She started having more falls. In all these years, my mother has broken her knee, kneecap, plateau, pelvis, ribs, shoulder, and collarbone. They were bad falls. She also fell on her head. (E9)

She dresses herself! Sometimes she puts on an upturned blouse, but she does it her way. In the shower, it's harder for her to wash her hair, it's harder to sanitize, and instead of cleaning herself, she gets dirtier. (E10)

Faced with this, family caregivers have taken certain actions, such as preventing falls, helping with feeding and bathing, handling invasive devices, dealing with sudden mood swings, and providing leisure activities.

We learn to put shoes on our feet, so they don't come off, always closed. You learn to put them on the bed, on the side, here we don't have a hospital bed, but we adapted it, we put a mattress on the side of the wall, so you don't hit your knee on the wall. 'To avoid drilling into the wall in the house, in the bathroom, use the "double suction cup", it attaches to the wall and can hold up to 60 kg, you can buy it in a building store.' My husband came up with the idea. You can stand up and do the exercises, and you don't have to spend a lot of money. You go for a walk, and you can take it with you". (E3)

And I only dress her when she's sitting up in bed because I don't dare to change her while she's standing up, because any carelessness is fatal, so I put on panties, diapers, pajamas, and clothes while sitting up! There's no way I'm going to put them on standing up!". (E6)

The overload due to the high demands of caring for people with HD meant that family caregivers had to seek psychological support. This contributed to building a space for talking/listening and promoting feelings of support.

In 2021, I found out about the Brazilian Huntington's Association and signed up to start therapy, and I started talking more. That's when I saw that there were thousands and thousands of people going through Huntington's too. Then I didn't feel so alone." (E5)

In 2020, I freaked out, it got really bad. It seemed that nobody spoke to me, nobody came to talk to me, nobody tried to find out or help. So, then I got really bad, and I sent a message to the ABH email and asked for help, I told them and explained that I was an only child and then they got in touch with me to start the therapies. Think of the therapy that

helped me, because I used to think about suicide and I'd always grown up with the idea, 'Whoever kills themselves is beyond salvation!' (E12)

Family caregivers pointed out the need to exercise patience in the face of mood swings in the family member with HD and to use coping strategies and daily reflections to live with the disease:

It's very difficult, but sometimes we must understand that things need to be at their time and not ours. Otherwise, they'll get angry, and you'll get angry with them, with the situation itself. And understand that it's a person, that they're in front of you and you must do your best for them now. And don't feel guilty when you lose patience or don't have the strength or the energy. There will be good days and bad days. (E4)

Go for a walk! Go for a walk in the square, enjoy the moment, do different things, and don't get stuck in your illness. You must live! We have a life beyond Huntington's, don't limit yourself. Whatever you can do, do it. (E8)

However, faced with the routine they lived; family members highlighted gaps in their preparation to carry out these care actions:

We do something every day and learn the hard way, but there's nowhere to turn, and look if there was, it would be great!" (E13)

[...] Many things are simple. People who don't know don't apply it because of that. For example, one day I arrived at the hospital to accompany my husband to surgery and the nurses were changing my decubitus position, and I said, 'My God, that's what my mother needs', before buying a mattress, that's what she needs. (E16)

I'm already able to bathe her, I just don't know if it's right [laughs]. But I think I could use some tips on how to bathe her if the way I change her diaper is right. For example, the medicine, is it right to put it in her hand or would it be better to put it straight in her mouth? These are doubts we have, you know?". (E9)

Lack of knowledge of HD and its implications for care practice

The analysis of the reports indicated the influence of the lack of knowledge about HD, both by professionals and family members, on the practice of care.

He has had symptoms for 20 years but was treated as something else. The neurologist hadn't done any tests. When we got to the new neurologist, she said, 'Look, I don't think your mother has restless legs syndrome. We're going to do a genetic mapping test', and she even asked about family history, but we didn't know [...]. So, she was the first to be diagnosed in the family". (E1)

At first, we didn't know anything, it took almost ten years for them to find out. They tested for Parkinson's, and Alzheimer's and nothing came up! And we went to lots of doctors until a doctor in Pará diagnosed it. (E3)

But before she was diagnosed with "chorea", which is the first symptom of HD, they didn't say anything more. She even went to a psychiatrist, who said she had bipolar disorder, but that was because she already had mood swings and then nothing else came of it. Only a neurologist, who by chance told us to do a genetic test. (E4)

Understanding the behavior of individuals at the time of care:

It seems that many people are not yet prepared to deal with this illness. Once I went to the hospital and the doctor and nurse said to her 'Stay calm' and I said, 'She's not nervous', and they kept saying 'Stop moving for just a minute so we can put the tube in', and I was so desperate! I couldn't stand it and said, 'She doesn't stop because she doesn't want to, but because she can't'. You may even know, 'You know it's genetic, degenerative, such and such. But I don't think there's any practical knowledge of how the patient behaves. (E1)

[...] It's very frightening to go to a place and say 'My husband has HD. And the person goes and looks it up on Google, it's very scary. It's something that makes you wonder, 'What do you mean? Everywhere I went 100% of the time, the only person who knew what HD the neurologist was who examined him, unfortunately [...] The issue of HD is a 'seven-headed puzzle', with lots of unanswered questions. (E5)

Because when I talk about Huntington's, people have never heard of it in their lives, and I say, 'You're thinking of Alzheimer's mixed with Parkinson's without memory loss', and they say, 'Is that it? That's it! And a lot of people don't know, they don't know what's going to happen. (E7)

Some family members mentioned events that happened in hospital environments that contributed to their insecurity about the treatment:

And what I find frustrating in the aspect of exams, is to keep asking for an exam of that every six months and it's not going to change anything, it's degenerative disease. There's no other way out, it won't get better. And that makes me wonder, sometimes, what works and what doesn't, even the medication she's taking: 'If I take them all away, what will happen? And I keep thinking, 'Is it worth it? (E10)

And there are still some [professionals] who look at us a bit strangely. And it's really bad, you feel a bit abandoned, imagine you're in the hospital, you're supposed to feel supported, you're supposed to know what's going on, and I know that a lot of people go through this. (E16)

As a result, the guidance offered by professionals to families is compromised.

There's the neurologist who accompanies the psychiatrist, the psychologist. But there was no time for guidance on how to look after her. (E10)

It's hard to find a professional who gives advice. I haven't had much preparation; we just get into the routine. (E16)

It's very difficult to talk about a specific moment, because it was more in the day-to-day running around that we learned, and it was that I ended up guiding the doctors, and the carers, rather than the other way around. (E20)

Family members also highlighted the influence of ignorance about the disease on access to issues related to citizens' rights, such as retirement and leave of absence from the INSS:

There was a time when we stopped giving her medication because she was going to be examined by the INSS, and as HD is a more behavioral and subtle disease, at first we were afraid that the INSS wouldn't accept her retirement or sickness benefit. This was suggested by the doctor himself. (E17)

When she retired it was very sad, they don't allow companions during this process, and I asked the doctor and with a lot of insistence, he let me in. Then he started asking, 'What's your name?' She looked at me and I said, 'Tell me your name, Mom', and she said

it in her little way, with difficulty. Then he asked, 'What do you feel?' She looked at me and remained silent.... Then he said, 'Tell me, so-and-so...' what your mother has, and I went on to explain, 'It's this, this, this, and crying...'. When I'd finished, he said, 'Well, don't worry, I'm going to ask for her pension'. (E18)

Sometimes, faced with a lack of guidance, family members look for resources to direct their care actions:

I went looking for information, I read articles, I went to the ABH website, and I registered. I'm always looking for something that matches what she has to offer. (E1)

But I looked for testimonials from people who had groups, and associations. 'Now I'm recent, I follow Instagram, I try to find out about the subject. (E2)

On Google [laughs]. Everything that comes out I read a little bit. And on a day-to-day basis, you pick it up". (E12)

DISCUSSION

Care practices for rare disease patients are built on the experiences of individuals and their families in the face of the circumstances arising from the disease, such as personal hygiene practices, food, clothing, and leisure. These conditions will last a lifetime and can evolve to different levels of complexity, which causes changes in care needs and the actions provided[11].

In the case of HD, care actions follow the individual's stage of the disease and aim to maintain physical and mental integrity, prevent complications, and even minimize the risk of death[12], which generates the need for a wide variety of care approaches and initiatives[13].

In this context, the role of the family caregiver is evident and the principles of the CCPF reiterate the magnitude of the family bond. In addition, it proposes that the care provided by health professionals should focus on personalized, coordinated, empowering care and be supported by what is important to the person receiving care[8].

A comprehensive study carried out with the families of people with rare diseases showed how families experience the process of becoming ill, and how their actions practically imply a search for support11. When it comes to HD, this search results in the strategies established by the families, such as adapting to the environment, using tools that facilitate a certain activity, carrying out the daily control of care through a notebook, changing the diet, dressing the individual sitting down to reduce the risk of falling, being attentive to signs of choking, adapting mattresses and other techniques[14].

It should be noted that the experience of care leads the family nucleus to identify possible complications, which generates the need for actions to prevent problems. This process becomes constant, since living with a degenerative disease demands frequent adjustments from those who are committed to the individual's daily care. In this context, the care process must be based on compassion and respect, and on preserving dignity[14].

To this end, the bond established between the health professional and the family caregiver needs to be strengthened, so that care planning and actions consider the individual aspects and needs of the person and their family8. As with other chronic conditions that generate dependency on care, valuing the family as the nucleus of care becomes fundamental for carrying out day-to-day functions[15].

Furthermore, knowing how care is organized and how the team interacts with patients and family caregivers contributes to the quality of the care offered and is in line with the evidence-based approach of the CCPF[8].

It should be noted that the particularities of a genetic disease such as HD, due to the complications arising from the evolution of the disease, require the expansion of care actions and generate uncertainties about the benefits of therapeutic treatments[15]. Among rare diseases, three events are identified, represented by the acronym "DOR", which is "scientific ignorance" (D), which involves the inability of professionals to differentiate between diseases. "Institutional omission" (O) is based on the scarcity of institutions that support diagnosed people and their families, which goes hand in hand with "non-existent social representation" (R), which contributes to barriers in guaranteeing access to constitutional rights[16].

Thus, the road to diagnosis is a long one, because there are many genetic diseases, variations in signs and symptoms, and no cure[17], which creates the need to plan care based on promoting quality of life[18]. Care planning includes the principle that governs its enabling property, dedicated to promoting independence[8]. It was observed that although knowledge appropriation, qualified guidance, and the presence of some professional figures were scarce, the caregivers demonstrated autonomy by seeking out the necessary information by their means.

Professionals' lack of knowledge about HD is widespread and influences the management and care of families and people with this disease[19]. Thus, it contributes to the feeling of insecurity and abandonment for family members, as they don't have enough information to quell their anguish and anxiety about the perception of the situations they may experience during the disease and the benefits of treatment[20]. Situations in which the professional is overwhelmed by uncertainties about how to conduct care can lead to unsafe care, which reflects on the quality of care[21], and the premises of the CCPF are not fully considered.

It is, therefore, necessary for care institutions and health services, as well as regulatory policies, to use care philosophies such as the CCPF, which provides, within its four principles, for the sharing of adequate and complete information that contributes to the care offered and decision-making. Likewise, mutual collaboration in the development of knowledge, health management, research, professional education, and existing public policies and programs[8] are valuable strategies.

In the interviews with family members, the neurologist, and professionals with specific training in mental health were mentioned as references in the care process. The lack of mention of nurses should be considered, especially as they are professionals who are in frequent contact with individuals and their families, and one of their duties is to promote health by offering guidance on self-care and care management[22-23].

A limitation of the study was that the interviews were conducted remotely since not all the individuals involved had access to the Internet. All the participants had their cameras on for the duration of the interview; however, due to connection problems at times, the calls had to be restarted, which may have interfered with maintaining the line of reasoning established at the time.

CONCLUSION

The family's knowledge and information about Huntington's disease guide the strategies used for care. Although it is a rare condition, the demands for care during the disease are like those of other chronic illnesses, such as: help with bathing, eating, oral hygiene, and care with invasive devices.

In this context, the role of nursing in providing guidance to family members/caregivers of people with rare conditions and monitoring their daily care practices stands out. The results of this study provide support for the planning of care actions by nurses, which include aspects experienced by family members of rare disease patients and can trigger reflections on the operationalization of public policies that address rare conditions.

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Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work - **Pleutim NI, Arruda BCCG, Arruda GO de, Palasson RR, Marcon SS, Teston EF.** Drafting the work or revising it critically for important intellectual content - **Pleutim NI, Arruda BCCG, Arruda GO de, Palasson RR, Marcon SS, Teston EF.** Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved - **Pleutim NI, Arruda BCCG, Arruda GO de, Palasson RR, Marcon SS, Teston EF**. All authors approved the final version of the text.

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