

Personal experiences in the health-disease process of Spinocerebellar Ataxia 7: a phenomenological study

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Abstract

Spinocerebellar ataxia type 7 (SCA7) is an autosomal dominant neurodegenerative disorder characterized by gait disturbances, dysarthria, and vision loss. In Mexico, a high incidence has been documented in the state of Veracruz, with many families having one or more affected members. This has negative repercussions on the economy, social, work, and family relationships, making it a public health problem. This study employs qualitative and interpretive methodologies derived from phenomenology based on people's everyday lives to understand their experiences. A person with the disease, a primary caregiver, and a person at risk describe the meaning and impact of the disease on their family and community. From a holistic perspective, the objective is to enable social work and the multidisciplinary team to understand how daily life is altered by the changes to which people must adapt due to the disease, their daily experiences in relation to their knowledge, how they feel upon receiving the diagnosis, the prognosis, the explanation, and the meaning they attribute to SCA7. This will enable the design of individualized, group, and community intervention strategies to mitigate the social effects of the disease and address the factors that disrupt their family and community relationships.

1. Introduction

Spinocerebellar ataxias (SCAs) are a group of neurodegenerative disorders that are heterogeneous from a pathological and phenotypic point of view, characterised mainly by loss of motor coordination. Specifically, spinocerebellar ataxia type 7 (SCA7) is characterised by gait disorders, speech disorders (dysarthria) and progressive vision loss (retinopathy) [1]. This condition has an autosomal dominant inheritance pattern. This means that every time an affected person has a child, this has a fifty percent chance of inheriting the causal gene and developing the disease at some point in their life. [2]. Furthermore, due to the inherently progressive nature of SCA7, individuals diagnosed with this condition will invariably be entirely dependent on another person to assist them in activities of daily living.

The disease presents two social problems: isolation and abandonment. The first results from the patient's progressive loss of independence. The second problem affects the patient's loved ones, who often become their caregivers. This caregiving can lead to fatigue, depression, anxiety, and other issues. The need to care for the patient exacerbates the caregiver's lack of free time for leisure activities, which can lead to abandonment. This occurs more frequently with women, who often return to their families of origin. In most cases, these women already have children, further complicating their families' economic situation [3]. As Daker-White's (2015) research indicates, ataxia has a substantial impact on patients' day-to-day lives, impacting aspects such as mobility and social interactions. A significant number of patients report a sense of compulsion to remain at home, often as a consequence of others' limited comprehension of their condition. The consequences of the disease, in terms of job loss and financial difficulties, are compounded by the rarity of the condition [4]. Daker-White (2013) have highlighted that patients encounter existential and clinical uncertainties due to the progressive nature of the disease. Furthermore, patients have expressed frustration at being confronted with an incurable disease [5].

Research conducted by the National Institute of Neurology and Neurosurgery in Mexico has indicated a higher prevalence of SCA7 in the central area of the State of Veracruz, Mexico [6]. Furthermore, corroborated the founder effect for the disease, meaning the affected families share a common ancestor who first transmitted the SCA7 causative genetic defect, which has been passed down through the generations [7]. Although SCA7 has been extensively studied, these studies have primarily focused on its biological and quantitative aspects, such as identification, description, and evolution. To date, no qualitative studies have examined the experiences and meanings of individuals coping with illness in their daily lives using a phenomenological approach. Thus, this study employs this approach to gain insight into how illness affects daily life. Socio-family analysis allows social workers to understand how primary and secondary support networks can be restructured and strengthened, enabling the health team to make decisions centered on the family and the real context, in addition to training for primary health care.

A comprehensive understanding of intersubjectivity among individuals with illnesses, their caregivers and relatives, and individuals at risk, as influenced by their values, opinions, experiences, and behaviours concerning illness, is key to grasping the problem deeply. One methodological approach that focuses on intersubjectivity and its relation to reality is phenomenology, which is the study of phenomena as they are experienced, lived, and perceived by each person. According to Husserl (2013, p. 154/74), phenomenology is a philosophical approach that establishes knowledge by allowing "things themselves" to become present in their essential content as they are immediately given. It involves approaching reality as a phenomenon of meaning. Grasping how things show themselves is an essential part of approaching them [8].

On the other hand, part of the category of the world of life refers to the experience of common sense and everyday life in which "man presupposes the bodily experience of his fellow men, his conscious life, the possibility of intercommunication and the historical character of social organization and culture". According to Schütz, quoted by Salas (2006, pp. 171–172), "people give a prior meaning to the illness and their experiences in everyday life" [9].

Therefore, the objective of this study was to understand the daily life experiences of three persons related with SCA7: a person with the disease, a primary caregiver and a person at risk, all of them from the center of Veracruz, Mexico. In addition, describe the meaning and significance that SCA7 gives to their daily lives, their relationship with their family and community, and identify the dimensions of daily life based on the experiences of each selected person.

2. Method and Research Design

2.1. *Theoretical Framework*

Qualitative research with a descriptive phenomenological design based on Edmund Husserl's theory, which allows us to understand the health-illness process from a subjective perspective and describe the

essence based on the experience felt, understood, and lived of people related to SCA7, respecting how it manifests itself in their consciousness and daily life, contributing to more sensitive care for patients and their families, which is not addressed in quantitative biomedical studies.

An attempt is made to make sense of the phenomenon based on the description of the experience as it is lived, through the exercise of phenomenological reduction, epoché [10], which made it possible to give meaning to the phenomenon of consciousness, as experienced and understood by the people interviewed.

2.2. Ethics

This study was approved by the clinical and research ethics committees (Protocol 159/24); participants gave their informed consent; the confidentiality of their personal information was guaranteed.

2.3 Access to the phenomenon and context

The reconstruction of reality was based on the experiences of the participants themselves; the information was obtained from semi-structured interviews conducted via the Zoom platform (due to the pandemic), which allowed for in-depth, detailed, and concrete descriptions of the experience. Free narration of the experience was privileged.

As previously outlined, given the high incidence of SCA7 in Mexico, many families are believed to originate from the same geographical region, specifically the central mountainous area, which is situated in the central portion of the state of Veracruz. Its location near the Sierra Madre Oriental mountain range makes the terrain very mountainous. There are many geographical depressions, such as the Tuzamapan (in Coatepec) and Limones (in Cosautlán de Carvajal) ravines (Places of origin of participants). With agriculture as one of the main sources of employment and economy in the region.

2.4. *Participants and Procedures*

For this research, the people invited were those related to SCA7: a person with the disease, a person at risk, and a caregiver. However, at the time of the interviews, two more family members joined in, as they expressed their desire to share their experiences and were therefore included in the interview: the son of the person at risk, who also served as a caregiver for two sick people (maternal aunts), and the husband and caregiver of the person with the disease, who, due to the importance of their experiences and discursive testimony, were included in the analysis of the interviews. To ensure the confidentiality and anonymity of each participant and, at the same time, to recognize what each person said, each participant was assigned an identification code.

Information Collection Strategy

Due to the geographical distance between the participants' place of residence and the research centre, the interviews were conducted remotely via Zoom, with the date and time agreed upon in advance, and

were stored on a personal computer to which only the researchers had access. They were subsequently transcribed for analysis using only audio.

Information Collection Instruments

For this research and given the objectives considered, narrative interviews were chosen, which, due to their characteristics, facilitate spontaneity on the part of the interviewees. Three semi-structured interview guides were created, consisting of open-ended questions related to the research objectives.

2.5 Analysis Methodology.

The analysis was carried out using eidetic reduction, that is, the internal 'meaning' of consciousness and how essences are revealed were sought. This was done in four stages:

1. 1. Holistic reading of the interpretations carried out by a multidisciplinary team (psychologist, bioethicist, social worker, medical doctor), maintaining a phenomenological attitude, with suspension of prior judgements.
2. 2. The texts were segmented manually and concentrated in a matrix, respecting the language of the participants, without theorizing or reinterpreting, which is known as units of meaning. Those obtained were: 1) Experiences of their daily life in relation to the disease, 2) Feelings when they learn the diagnosis and prognosis, 3) Explanation they give to the disease (origin and meaning), 4) Changes in interpersonal relationships, both family and community, 5) Economic repercussions for treatment and care, 6) Perception of discrimination or stigma among family members, the community where they live, or those close to them.
3. 3. The information was reformulated in phenomenological language, remaining faithful to the experience described, without introducing causal explanations or external theoretical frameworks.
4. 4. Construction of shared meaning structures. Common structures were identified to understand the socially constructed phenomenon situated in everyday life, which allowed for eidetic understanding (identification of the invariant elements that constitute the essence of the lived experience).

3. Results

To present and discuss the results, a description of the participants and their family tree are first presented (see Fig. 1). The results are then presented according to the proposed analysis categories. Within each category, the most relevant subcategories identified during the coding process were included. Each category is supported by quotes from the original transcripts.

Patient: MMPE a 56-year-old woman who has a degree in accounting. She is married and does not have children. She has a maternal inheritance of the disease. She resides in Coatepec, Veracruz. She experienced the onset of the disease at age 41. Initial symptoms included visual disturbances and difficulties with gait and coordination. During the interview, she identified visual impairment as her primary disability. Currently, she depends on her husband for daily activities.

Subject At Risk: MSPR a 68-year-old, divorced, and retired teacher. She has a married son and lives alone in Xalapa, although her family of origin is from Tlaltetela and Cosautlán. Two of her sisters were affected, and the inheritance was paternal. The MSPR's son, MAPC, participated in the interview. He is married and lives independently. He helped care for his two maternal aunts from his teenage years until their deaths.

Caregiver (Daughter and Niece): MKPC a 30-year-old single woman with a degree in criminology and criminalistics. She is a police officer living in Xalapa, Veracruz. She lives with her disabled mother and older brother. During the interview, she mentioned that she cares for her mother, who is completely dependent due to her inability to walk or see, as well as her uncle, who requires full-time care. MKPC cared for them both for three years until she decided to place her uncle in an elderly care home because caring for them both was too much of a burden.

Once the notes were taken and after analysing the phenomenon and the lived experience, the essential content of the experiences became evident. According to Husserl (2013), coinciding meanings were identified among the participants [8].

3.1. Everyday life experiences in relation to disease.

When a person has a chronic degenerative disease, such as SCA7, their daily life and that of their family members is often significantly altered.

There are people who accept the suffering of their relatives. For the person who develops the disease, as in the case of MMPE for whom ataxia means 'not being sick because she 'does not feel sick', regardless of her lack of vision or inability to perform everyday relates feeling or not the disease with the possibility of not being completely dependent on the caregiver. This situation generates profound discomfort for people who are bedridden and dependent on outside help for care. This discomfort often manifests as a reluctance to accept care and a desire to end life. One alternative is to accept the prevailing norms and expectations (see Table 1).

Table 1
Experiences in daily life in relation to the disease

Excerpt information	Verbatim	
Excerpt 1: woman, person at risk of inheriting the disease MSPR	<i>"My mother was exceptional, that is, from her I learned never to complain, she never cursed what had happened to her, she always found a way to move forward".</i>	Accept the suffering of their relatives
Excerpt 2: woman, patient MMPE	<i>"At least I was able to live, dance, and work"</i>	Accept the prevailing norms and expectations
Excerpt 3: man, carer HACP	<i>"We had to make changes in our routine, in our life, in our home, in order to be able to burden and cope with the disease, that adaptive process was not the same for others, it is very difficult to lead a normal life because your whole world changes, when you become responsible and give moral, physical and economic support, many do not want to assume it...they withdraw...they talked to me and told me to come because your aunt has gone crazy, you had to go because there was no one else who could help".</i>	Feelings of abandonment, as well as an overload of work
Excerpt 4: woman, person at risk of inheriting the disease MSPR:	<i>When they told me let's pray for IPEH or for MPEH I said, 'Don't pray for them to live, pray for them to leave, I never prayed, 'Please help them...' I told my mum: "When they enter their final stages, no going to the hospital, no need to give them IV fluids, do this and that; we have to be aware that they have to go, they can't continue with this suffering".</i>	Thoughts and/or wishes for the patient to die

3.2 The feelings when they know the diagnosis and prognosis.

Individual responses to the disease vary significantly, and this variation often correlates with the individual's knowledge of the disease.

For individuals experiencing early symptoms, it can be reassuring to determine the cause of their discomfort. However, these feelings of relief may change as symptoms worsen, sadness, anger and helplessness may appear, among others.

This can present a challenge in cases where there is no history of the pathology in previous generations. A diagnosis of a hereditary disease has implications for the family because immediate family members, such as children and siblings, have a 50% chance of developing the condition. This can trigger a range of emotions that vary in intensity (see Table 2).

Table 2
The feelings when they know the diagnosis and prognosis

Excerpt information	Verbatim	
Excerpt 1: woman, person at risk of inheriting the disease MSPR:	<i>"It took many years until I found a doctor (who helped them), then it was like finding a light when he told me: you are not alone, we are going to support you... I said to her: mom, what did the neurologist tell you? And she told me: here is the paper, and then I saw it, it said ataxia, what did he tell you, what's going to happen or what's not? Well, that he is going to die!... You can go to the dictionary and look up the word and start to understand what it was, which this means, why she had it"....</i>	Pathology in previous generations, as evidenced in the case of the MSPR sister's
Excerpt 2: woman, patient MMPE	<i>"I felt fine, I received the news, in 2008, but I considered at that time, that there was still a little time left for me to have more serious symptoms... somehow, I felt good because of the following... when they gave me the test, and they explained to us that this situation is genetic, at first we were grateful not to have had them (children), because I am already sick, but I did not inherit it from anyone".</i> <i>MMPE: "As time went by, it is normal that one feels not very happy, there comes a time, like now, that, although I can move by myself, even slowly, there are many things I can no longer do... sometimes I feel very sad because I can't do anything, but in a way I am happy because the disease is over with me, on my side.</i>	Determine the cause of their discomfort
Excerpt 3: woman, person at risk of inheriting the disease MSPR:	<i>"Geez, very scared because I already had my son, he was still young, almost a child, 12 at the most 15 years old; it's very shocking, maybe at that moment it doesn't matter if I have it, but what about my son, I don't want anything to happen to him"</i> <i>HAPC: "it's a struggle of thoughts, you know you may have the disease, there is a possibility that you may not have it, the first thing is to think about what could happen, one of the hardest things for the family of an ataxic person is to see...and to know what is going on, in the case that you do not have it, you know where you are going...there comes a moment when you say: yes I have it, I cannot do anything else, I have it and that's it, you have to continue your life in a normal way knowing that there will come a point in which it will not be normal...and if I have it, who is going to take care of me? I mean, who will be there? It's even harder to know where you're going or how you're going to end up."</i>	This can trigger a range of emotions that vary in intensity

3.3. Explanation given to the disease (Origin and Meaning).

The explanation that people related to SCA7 give to the disease may vary depending on the origin they attribute to it or the meaning it has had for them. When the origin of the disease is explained or understood by heredity, they know or believe that it comes from their ancestors or other family members. Another way to explain the origin of the disease is through the findings, that is, when people notice the first symptoms or difficulties, and nothing was known about the disease yet.

The recognition of these findings and the experience of starting to live with the symptoms, in the case of patients, and living with the difficulties that arise, in the case of family members, leads them to have

different perceptions of it.

With respect to the meaning, they give to the disease, the lived experience leads them on the one hand to negative expressions, and on the other hand, to have a prospective (see Table 3).

Table 3
Explanations for the disease (origin and meaning)

Excerpt information	Verbatim	
Excerpt 1: woman, person at risk of inheriting the disease, MSPR:	"My grandfather married his second wife years later, and from that marriage, I have five uncles, three of whom suffer from the disease.... I came to think and suppose that the day my father had an accident and fell, it was because maybe he already had ataxia manifestations"	They know or believe that it comes from their ancestors or other family members
Excerpt 2: woman, patient MMPE	"For her (referring to her mother), the disease was passed from my grandfather to me. I don't know if she (mother) just understood that I have it for her" or if they didn't develop it before they died".	
Excerpt 3: woman, patient MMPE:	<i>"Well, at that time I didn't know I had the condition, but I had noticed that it was hard for me to do something, I had a little difficulty in doing certain things"</i>	When people notice the first symptoms or difficulties, and nothing was known about the disease yet.
Excerpt 4: woman, person at risk of inheriting the disease, MSPR:	MSPR: "my sister was very young when she started to wear glasses, the ophthalmologist observed how she walked, we started the pilgrimage, my mom took her to one and another, the neurologist who saw my cousin, told her she has a hereditary disease called ataxia".	
Excerpt 5: woman, person at risk of inheriting the disease, MSPR:	<i>"Ataxia is the worst thing that, I believe, can happen to a human being "," to end up turned into a rag "," to depend 100% on the other person, not to see, not to move, what do you want to live for?... there is no way to defend yourself, it is what hits you the hardest, knowing that whatever you do there is no cure and at the end of the day it is not a peaceful death, it is not a human death".</i>	The meaning they give to the disease, the lived experience leads them on the one hand to negative expressions and on the other hand, to have a prospective
Excerpt 6: woman, patient MMPE:	<i>"they told me it was confirmed and that, with this type of condition, little by little, I was going to have a little more difficulty in several areas: my vision, my motor skills, my speech... I already knew what to expect because of how my cousins' condition had developed, so I thought, well, sooner or later it will have to be like this".</i>	

3.4. Changes in interpersonal relationships

Neurodegenerative diseases entail a change in interpersonal relationships (family and community). In the case of SCA7, as the patient's requirements for care increase, changes are observed in their

relationships. These changes are the result of the patient's shifting role, resulting in their disconnection from the community. Changes in communication and in family relationships (rapprochement or distancing) are observed. At the community level, greater closeness or isolation can be observed.

Regarding communication between family, there are situations that make it necessary to share information about the existence of the disease in the family with other people, such as when one desires to have children. In the same way, changes in the family relationship can be observed, either of rapprochement and/or distancing and it is even possible that a pact of silence could be reached.

Finally, regarding the community relationship, a dual response was also observed. On the one hand, there was a rapprochement between the community, and on the other hand to separate from the community for protection (see Table 4).

Table 4
Changes in interpersonal relationships with family and the community

Excerpt information	Verbatim	
Excerpt 1: man, carer HACP	HAPC: <i>"I talk to my girlfriend, look, in my family there is this disease, it is a process, it leads to this, this and this, it takes years, it is not from one day to the next, physically it starts a decline, if you want to be with me, go ahead, if not I am not going to hold you back or force you, it is difficult, very difficult".</i>	It is necessary to share information about the existence of the disease in the family with other people
Excerpt 2: woman, carer MKPC:	<i>"We have been together for 2 years, but still... it is an issue with him, with him it has been complicated with my previous partners no, but with him yes, he wants to have a child"</i>	
Excerpt 3: woman, person at risk of inheriting the disease MSPR:	<i>"My brothers were never interested in knowing what was going to happen, they didn't even care what might happen to them, they said: "my sisters have the disease and me, why do I want to know if I have it? when I told him that we can enter the protocol to know if I have it or not and he said: "I am not interested, what do I gain if they tell me that I have it, besides, he is the one with more children, seven, and they have not been interested in finding out either, he says: ...I am over 60 and I have not had any symptoms, so it is most likely that I don't".</i>	Changes in the family relationship can be observed, either of rapprochement and/or distancing
Excerpt 4: woman, person at risk of inheriting the disease MSPR:	<i>"The final part of H's agony was shared, because my sister-in-law arrived, a niece, those were the final 5 days", as</i>	In their relationship with the community, they observe either approaching or distancing themselves from it to protect themselves.
Excerpt 5: woman, patient MMPE:	<i>"Few neighbors know that I have a condition that prevents me from being fully mobile"</i>	

3.5. Economic, Occupational, Social Implications for Treatment and Care

3.5.1. Sickness related expenses

Because SCA7 has no pharmacological treatment, the economic expenditure in this area, is directed mainly towards rehabilitation therapies and home modifications, for example, rearranging furniture to allow sick people some mobility within the home.

3.5.2. Employment implications

The sick people were working before the disease caused serious mobility or vision problems. When they could no longer do so, they quit their jobs.

For caregivers, the work continues, with the help of other family members, and caregiving duties rotate. One caregiver took three years to decide to go back to work, and one of his sick relatives had to be taken to a nursing home.

3.5.3. Changes In Social Activities

Social activities of the family of people with SCA7 changed, it can be observed that the changes are focused on satisfaction of the affected persons, with the purpose of compensating for the daily routine prior to the loss of vision or mobility. Caregivers adapt to continue sharing common activities (see Table 5).

Table 5
Economic, Occupational, Social Implications for Treatment and Care

Excerpt information	Verbatim	
Excerpt 1: man, carer HACP:	<i>'My mother's house had to be remodelled so they could climb the stairs, enter the bathroom, and leave the bedroom, tubes were installed throughout the house; the house had to be modified for a blind person.</i> <i>"You have to make adjustments in your life, in your economy, in your infrastructure, and know beforehand that you're going to have a burden"</i>	Sickness related expenses (modifications)
Excerpt 2: woman, carer MKPC:	<i>"The bedroom is close to the bathroom and the kitchen. The only thing that was put there were two chairs on opposite sides for her to hold on to, but now, after her fall, we've made some handrails... to see if she feels safer getting up and going alone without supervision in her room."</i>	
Excerpt 3: woman, patient MMPE:	<i>"I can no longer work, and I really liked my job, but well, you must let everything go little by little... I feel very sad now, sometimes very sad because I can't do things"</i>	Employment implications (it was necessary for them to leave their jobs)
Excerpt 4: woman, carer MKPC:	<i>"I have a degree in criminology and criminalistics. I was in the process of joining the forensic services, but with my uncle and my mother, I couldn't do anything, so I spent three years taking care of them, until one day I said: 'what am I doing? I don't want to live to take care for someone, no, no, regardless of whether it's my mum, my uncle, whatever, I want to make a life for myself and not my life in a sentimental sense but my personal professional growth, so I decided to take him to a nursing home, since I started working I have been more distant from my mum, which was what I was looking for".</i>	
Excerpt 5: man, carer HACP:	<i>"I had to adapt everything so that they could be well, as much as possible to give them quality of life". HACP: "at the time we have to take care of my aunts, it was years of not going out, the trips were to San Luis [San Luis Potosi, Mexico] to see the family, and they could say goodbye."</i>	Changes In Social Activities (for satisfaction of the affected persons)
Excerpt 6: woman, patient MMPE:	<i>"My husband bought a big screen at home, to watch a movie that we might like, at least I can see the images a little better, because we used to love going to the cinema, but now I can't go anymore".</i>	

3.6. Perceived discrimination or stigma among family members and their community.

People related to SCA7 did not report experiences in this regard, they did not refer any discriminatory treatment, what they perceive is lack of knowledge and understanding of the disease on the part of the community (see Table 6).

Table 6
Perception of discrimination or stigma

Excerpt information	Verbatim	
Excerpt 1: woman, person at risk of inheriting the disease MSPR:	<i>MSPR: "my uncle, he was a teacher and then they said that he went to school drunk, they lived in a small town, so he had to walk from his house to school, and that was where people said that, how is it possible that the teacher comes to school already drunk!". MMPE: "No, of course not, the only thing here is that even one's friends don't quite understand".</i>	lack of knowledge and understanding of the disease on the part of the community

4. Discussion

According to the Everyday life experiences in relation to disease Berger and Luckmann (1998) argue that the everyday life of subjects is an ordered, coherent, and objective knowledge of the reality in which they are immersed [11]. Everyday life does not need verification; living with SCA7 leads people to reconstruct relationships, actions and give them meaning. Anything is done to "give quality of life" to those being cared for and strategies are generated to "adapt". When the disease occurs, it is seen as a "process".

From biomedical models, the dimension of illness implies the convergence between discomfort and the world of the sick person, which has had a negative connotation, since the meanings are usually linked to loss, pain, suffering, disability, or death, and involves both the subjective sphere of the individual and the social and cultural spheres [12].

Intersubjectivity, according to Schütz (2003), is linked to the essence of caregivers' thinking and, in the case of people with SCA7, it is linked to belonging to that social and family world that allows them to generate first-order constructs that are formulated from everyday experience [13]. The loss of mobility and the potential loss of independence can be regarded as the most unfortunate outcome for a human being. However, it can also be seen as a period in which people adapt and find ways to continue living, despite the challenges they face (Shütz, 1993). This perspective highlights the ability to face and adapt to situations that arise [14].

When there is a chronic patient at home, who requires more care as time goes by, caregivers often manifest lack of support and feelings of abandonment, as well as an overload of work, according to Giralt & Franco (2008) and what this implies [16]. The care required for the person with SCA7 increases as the disease progresses, requiring continuous 24-hour care and in the terminal phase of the disease, some family members may experience thoughts and/or wishes for the patient to die, which may generate discomfort among other family members. The breakdown of family subsystems due to exhaustion, communication problems, and financial hardship is one of the main challenges for intervention from a social work perspective. Institutional and organizational management facilitates real connections between the individual experience of the patient, the caregiver, and their family.

Strengthening a broader social world facilitates shared care. Family mediation promotes the distribution of roles and time and allows for the generation of shared meanings.

The feelings when they know the diagnosis and prognosis. The comprehension of the subjective sense is associated with the diagnosis and prognosis of individuals related to SCA7 (patients, caregivers and subjects at risk). This is characterized by what Schütz (1993) denominates "common sense" [14]. The future is conceptualized based on the data that preceded it, related to everyday life experiences, that is, the "lived world" (Husserl, 2013, p. 167) [8]. This is achieved by observing and living with sick relatives (if any) during childhood or adolescence, or as parents, grandparents, sisters, brothers, etcetera.

The interpretation or configurations of meaning and sense that people give to SCA7, take elements of the "known", according to Schütz (1993) allow to place the experiences in different spheres of meaning that can be within logical formulations or in the emotions [14], the here and now, in the resolution of problems, reason and will, each subject from a different world, considering their priorities that constitute their social world, where the SCA7 can generate a process of "adaptation" to a situation that leads them to think about "suicide", reflect on 'euthanasia' for sick people or for themselves, in the case of people at risk to make life prospects and how they want to live "to the fullest". The receipt of a diagnosis and prognosis of a hereditary neurodegenerative disease, such as SCA7, can result in a range of emotional responses in the individual and/or their family, including anger, sadness, hopelessness, and frustration.

Understanding discrimination according to Solís (2017) as the set of practices, informal or institutionalized, that deny equal treatment or produce inequalities for certain social groups [17], however, the people interviewed did not report any experiences in this regard, nor did they indicate any discriminatory treatment; they only perceived a lack of knowledge and understanding of the disease on the part of the community.

When those involved with SCA7 talk to people who are not related to the disease, they try to give explanations that at the time do not reflect what they are feeling or thinking, they do not validate their thoughts or feelings, they give explanations for actions from a different social world, taking up Schütz (1993, p. 39), "when I turn to another person and ask him or her about some topic, I am assuming a social structure in which I recognize the other, I assume that we share certain codes, we join them in some common activity", who influence or allow themselves to be influenced, since they do not share the same codes The interpretation given is: "they are in denial", "they don't know what they are experiencing", "they don't know the process", distancing the link even more [14].

Limitations

The study exhibits significant methodological deficiencies, stemming from the collection techniques employed and its cross-sectional design, which precludes the capture of participant experiences throughout the progression of the disease. This is due to the substantial fluctuations in needs, emotions, and coping strategies that occur between the initial diagnosis, the moderate dependency phase, and the total dependency phase. The presence of selection bias is a possibility, given that the participants in this

study exhibited specific characteristics, including higher levels of educational attainment, more robust support networks, and enhanced capacity for reflection on their experiences. Diagnostic experience has the potential to engender psychological burdens, as evidenced by the observations made in the case of the participants. Consequently, it is recommended that subsequent studies be conducted with the participants address any emerging concerns.

5. Conclusions

After carrying out this research, it was observed that SCA7 represents a daily process for patients and their families that, in turn, gives meaning to life and the disease with a certain future.

Since there is no cure, the world of common sense and everyday life are reconfigured from the knowledge of diagnosis and the presence of symptoms to the loss of independence in the sick person and the caregiver. For everyone involved with the illness, it gives meaning to life, from "trying to live life to the fullest" to considering "suicide" and reflecting on "euthanasia."

The disease in turn, rebuilds relationships and actions mainly in caregivers who seek to "give quality of life to their relatives", having SCA7 is considered "the worst thing that can happen to a human being", and generates a construct of 'strengthens' and adaptation to "see how to get ahead".

For the people involved with SCA7 it is necessary to have intersubjective spaces for dialogue and validation of the feelings and perceptions that relate to the reality they live with the outside world, allowing them to share common codes and activities that bring them closer to the rest of the people who do not have the disease.

This study provided an overview of people's experiences with a specific phenomenon, in this case, SCA7. This will allow healthcare personnel to understand that a disease, like this, alters their family and community relationships, requiring multidisciplinary care to intervene in the different areas affected by the disease.

Declarations

Conflicts of interest

The researchers state that they have no conflict of interest.

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Figures

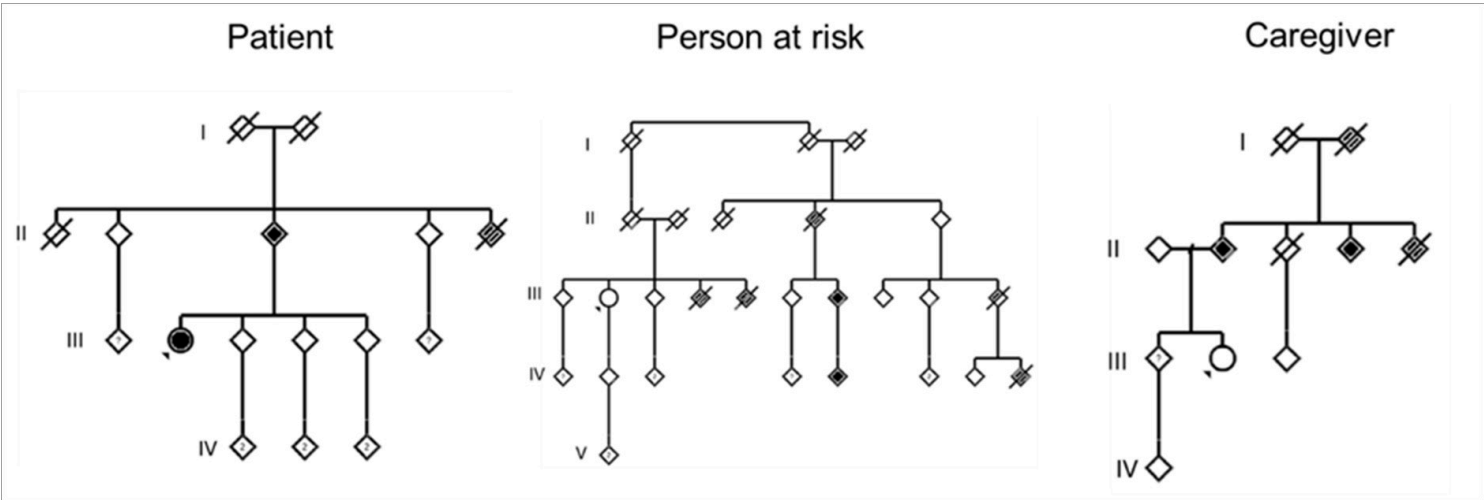


Figure 1

Participants' pedigrees (figure created by the authors)