192 - SIGNIFICANCE OF CARE FOR RELATIVES OF PACIENTS WITH FRIEDREICH ATAXIA

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INTRODUCTION

We knew, beforehand, that the Friedreich Ataxia (FRDA) is considered a rare disease, registered as chronic-degenerative and that the majority of cases progress to death in adulthood. The diagnosis based only on clinical findings is limited, but facilitates better screening for selecting the suspected cases that deserve to be tested (ALBANO, L.M.J., et al. 2001). Therefore, knowledge of a family with four members carrying awakened us the curiosity to understand the universe and that family that we could provide a support from a relationship of aid to it.

Many questions arose, and to begin the search, and search by studying the few published articles on the subject, began to ask ourselves: How the family (re) organize the passing of the illness? What does the "caring" for the family mean? What health services and social media can help a family with this problem? Basically, from these three questions, we opted and aimed to seize the meaning of caring for family members of patients with Friedreich ataxia.

LITERATURE REVIEW

The Freidreich ataxia (FRDA) is a recessive hereditary disease, which is due to a modified gene - the gene X25, located on chromosome 9. Thus an individual who receives a gene from one parent patient becomes bearer of FRDA and never develop the disease being able to transmit it, if inherit two copies of the gene develops the disease amended (GONÇALVES, S.; AMARAL, C., 2000).

From a cell the most common mutation in FRDA is an expansion of a DNA segment, known as GAA repetition that is repeated from 7 to 22 times in normal people, but in people with this disease the sequential GAA are repeated between 200 to more than 1,000 times. This feature makes it difficult to transcription and translation during protein synthesis that occurs in the cells of individuals with the Friedreich ataxia. Thus, the protein frataxin, which should be produced in large amounts, not exist in sufficient quantity that will bring consequences. The frataxin is somehow involved in regulating the transport of iron out of the mitochondria. This can be compared to a central power which essentially the glucose (or other nutrients in case of lack thereof), by the process of cellular respiration, is converted into energy (ATP) which is needed to build and maintain the functioning of cells (main task of mitochondria).

According Bagliolo (1994), Friedreich ataxia is the most common site of the spinocerebellar degeneration regarded as the most well characterized, among others. There is a degradation and degeneration of the spinal nerves of the limit, the cerebellum and the nerves that carry sensations from hands to feet. Considered hereditary with autosomal recessive heritage, whose frequency is almost 1 in 100 and whose prevalence is 2 in 100,000, is probably the most common type of ataxia static and dynamic in the first sign that it is between the ages of 5 and 15 years . Changes of the word coexist, nystagmus (which appears later). Almost always, is accompanied by malformations of the skeleton, walking horse, cifo-scoliosis among others. And, even loss of hearing, vision change in the colors. (PEDROSA, C.; OLIVEIRA, E.; PIRES, R. 2002; SILVA, A.D. et al, 2001).

For Bennett and Plum (1997), it often is associated with cardiomyopathy and an insulin-dependent diabetes mellitus in 10 to 20% of patients. Heart failure is probably the main cause of death, with an average age of 37 years. The first symptoms are observed during childhood or the early years of adolescence. Presents difficulties of precise movements with the lips and tongue, making their way to speak slow and difficult to understand. This fact, that worsens over time, just like locating where your feet and hands are in space (impaired sense of position), weakness and loss of muscle mass in the legs and hands.

Smeltzer & Bare (2002) affirm that the family is the main source of support in our society, as a dynamic system. Therefore, the inability of a member affects the other members of the family. Only with the incorporation of the process of rehabilitation, the family system can adapt to change in one of its members. The family provides continuous support, participate in solving the problem and learn to take the required continuous care.

For Collière (1989), caring act is essential, not only to the lives of individuals, but to the survival of the entire social group. For the author, this act represents a vital function inherent in the survival of every living being, because "the treatment is the guarantee of direct continuation of the life of the group, the Homo species".

Thus, care can be described as a social practice that involves the manifestation of culture, behavior, the beliefs and values determined by their meanings in different social groups. An important point to be considered are the differences found in the systems of beliefs and values between the patient and the caregiver. In this case, the caregiver must be delivered in a holistic approach to help protect, restore and maintain the patient in her home.

METHODOLOGY

We opted for a qualitative research on the history of life of family caregivers of FRDA, precisely the oral history, conducted with a family in a town north of Parana. For Meihy (2002), "*is a modern method used for the preparation of documents, archiving and studies concerning the social life of people. It is always a history of the present time and also known as living history. The oral history is presented as a way to capture the experiences of people willing to talk about aspects of his life, maintaining a commitment to the social context.*".

The oral history begins from the choice of the person or a group to be interviewed. This theoretical and methodological feature requires minimally three elements: the interviewer, the interviewee (witness) and the recording equipment, in addition to the authorization, called letter of transfer that ensures the researcher using the archiving and publication of oral history. The deponent has an important role, as part of the beginning of the story until the authorization for publication, which should be informed about some aspects about the Res. N. 196/1996/CNS. After recording, the transcript is done, the textualization and transcription (MEIHY, 2002). We also recommend the use of field copybook, which act as a diary.

INTERLACING LIVING HISTORY FROM REALITY TO LITERATURE

By articulating the alive story alive with literature in which patients and caregivers speak of its past and present, their

doubts and uncertainties, we seek to recover the sense of living for caregivers. We identified individuals with roman letters in capitals, the family caregivers as witness plus the family [mother / sister] and the professionals who occupies the office followed by a letter of the alphabet at random.

Regarding the sister we felt the need to talk, wreak, to refer to significant changes in the style of your life from the moment when his brothers were sick.

"... I can not go anywhere, I can not get much time away from home. I have no pleasure, because step all the time taking care of the boys. One of the only places that I will go is the church "[deponent sister].

Apart from the changes related to the lack of leisure and the deprivation of work outside the home, appear in this verbalization, aspects involved in the conflict in family relationships, especially between the caregiver and spouse.

"... my first marriage did not work, because even then I was not living with them, I lived there more than at home, had almost no time to devote myself to my husband. It was then that started the conflict, fights and finally the separation. I got married again, lived a time living at home separated from my mother, but always kept helping her, but I saw that was not enough, because my brothers needed the care 24 hours a day. My mother is already old and is not capable of taking care of them alone, because you can not count much with relatives. Today, I, my husband and my son live in the house with them, to help more, but realized that my husband is no longer the same, I think he basically, in essence, it likes to live there and does not accept the in fact sometimes I leave it aside, you take care of my brothers. " [Deponent sister]

Other causes also contribute to a deterioration of physical and emotional caregiver, due to the expenditure of energy that demands the process of care, since this condition almost always falls on only a family member, who often can be seen alone, helpless in front of the problem.

Regarding the mother, we see a wonderful person, who despite all suffering, maintains an ogle and serene look. And we cannot avoid the question: "How can you express this peace, in your face, despite all suffering that has gone by and which continues moving in your life?" Calmly replied:

"... I was always a poor person, my husband lived changing around, had not a fixed service, always had financial difficulties, then never lost the faith in God. When moved to Rolândia, we live in a small house with no comfort, no window in the room, so without the minimum condition to care well for my children. This season, the four were bedridden, urine and evacuate the bed, and I did not have anyone to help me, the only thing I did was sit in a corner of the room and cry, not that they know, asking God's help. I could not cry in front of them, because even without talking, they noticed on my face when I was sad, and it's sad seeing they were also showing sometimes through tears dripping from the face" [Deponent mother].

According to FRITZEN (2000), when possible we must smile in the midst of the trials. Because after all, the ordeal is the expression of the will of God, therefore, is a gift from God, just like what we like. The sick person is extremely sensitive, although sometimes be apathetic, indifferent or even aggressive, like a sponge absorbs what is happening around you. Even without being able to express verbally, to capture what is happening around you, reacting in different ways.

We saw ambivalence on the part of a deponent-sister who is now reflected by the presence of a feeling of love, sometimes by the revolt, both very intense.

"... Sometimes I stayed with them very nervous, because even when they moved their arms, they made aggression acts towards the children and me. They hit with great force, which at times I was angry, but at the same time I felt pity, because they were perceived willingness to walk, get out of bed, because neither the other rooms of the house they know " [Deponent sister].

We observed that in this patient-family interaction, love, affection and tenderness overcome the anger and hate. To FRITZEN (2000), the synthesis of all laws and norms governing the relations of a community is love. Love is the great mainstay of care, especially the love of a mother for a considered special son, is for a genetic disease, for a physical disability, for a chronic disease, hence the love everything can, including the miracle, when necessary.

We note also the sister in uprising, for what it sees as the neglect of health professionals.

"... we need to be strong for bear, bear it all, because when we take my brothers to the doctor, we sometimes stay all day and when we come back we have the feeling that nothing was done. A feeling of emptiness and anger because my brothers are discarded in the hours stretcher in the same position, tired, hungry, and when the doctor arrives just gives a peek, there is nothing to us and then he is gone. (...) We know that the disease has no cure, but it is difficult to accept, and this situation worsened after the doctor who came here at home and began researching the disease. The doctors who cared him din not like, so my brothers are treating with contempt and indifference" [Deponent sister].

The testimonies mentioned show that the caregivers fulfill a difficult task to bring them to the hospital because depend on the availability of ambulances and other persons, to carry them, beyond the expected and the "goodwill" of professionals to help them .

The manner the doctors communicate with patients and the degree of perception of patients and family are important factors in the doctor-patient relationship. The positive behavior of doctors strengthens this relationship. So the feeling of comfort and trust between doctor and patient is a component well accepted and desirable for any medical consultation. Communication between doctor and patient must care is increasingly on the basis of information that patients want to know, as details of his illness.

Another common type of feeling that appeared to be frequent in the relationship between caregiver, the family and the process of care of bedridden patients was fear, as we will see in these verbalizations:

"... I'm already old, and I have great health, the only thing I ask is that God give me strength to take care of my dear children, what I can do for them I will ... For now I have my daughter and assistant "L", a better angel, a great friend who helps taking care of them, but I'm afraid of losing the Auxiliary "L" that takes care of them so well that despite them being fat has no wound along their bodies" [Deponent mother]

The feeling of fear which is revealed by the mother reflects a concern of being unable to maintain the care, if she becomes ill. That feeling is a constant threat felt by the caregivers, resulting in an unstable state of health. The important message is that the revelation of consciousness that the mother has the responsibility of its role, and when unable to care for, who will replace her? What will be the fate of these children?

The feelings of compassion and sadness that are not verbalized, are themselves in the form of silencing, so subliminal messages issued by the caretakers, observed especially when the mother speaks, touches and looks at them.

We agree with Boff (1999) by stating that the front of the other makes it impossible to indifference. The front of the other forces me to take a position because speeches, provokes and evokes, especially in the face of the impoverished, marginalized, sick and excluded.

As we saw earlier, many of the sentiments expressed by family members, but beyond these, there are other emotional reactions, because they face a big challenge that is the deal with a new situation of life in the face of the consequences of Friedrich's ataxia, because the neighborhood the death is a next step.

People are threatened with death in different ways. According to Kübler-Ross (1989), the emotional responses of a person facing death can be traced through five stages: denial and isolation, anger, bargaining, depression and acceptance. These

five stages does not always happen in sequence, they may overlap each other. Patients and their families staying in a round cycle experience and may be at different stages in a given time. Thus, for the caring parent, "*it was very difficult at the beginning of the disease, not accepted, because they were healthy boys until all sixteen years*".

The denial and isolation occur as a first reaction to the possibility of imminent death. A person cannot accept the fact that death is near, the denial allows the existence of hope.

We noticed that in the process of family caring many factors add to the existing ones that interfere negatively in their development according to the overload of missed opportunities and the intense mobilization of forces supported, and defensive (re)establishment of defense mechanisms to keep up is balanced in front of so many demands. These are limitations that prevent developing a more effective role of caregivers.

To develop the care at home, the caregiver is faced with various kinds of difficulties, especially those that invariably limit the quality of care, which range from internal aspects such as socio economic and cultural level, inadequate space at home for the patient, relationships family, to external factors such as access to health services, low income to buy essential medicines to maintaining the lives of individuals.

"*... the disease of my brothers is not only driving, with the passage of time they spend to suffer from heart, the pressure rises and diabetes appears. The medicines we get in the clinic, but many times they make it without it, so we have to buy them, and often we do not, because we do not have money" [Deponent sister].*

"*... my brothers need a physical therapist accompany, because they are shriveled, their hands locked, the feet turned behind. At the beginning until one appeared , but she went only along the first days, then they came back, and we do not have conditions to do physiotherapy on them because they are very heavy. If we had money, everything would be easier, so sometimes I revolt, because unfortunately the money can it all. They also needed a speech therapist, a psychologist, finally, a team to work together to alleviate what they feel " [Deponent sister].*

For Smeltzer & Bare (2002) the rehabilitation is a creative and dynamic process that requires a team of professionals working with the patient and the family. Team members represent a variety of specific on which each health worker makes his particular contribution. Accordingly, each evaluates the patient and identifies their needs within the scope of its area. They also argue that patients confined to bed for long periods, with motor or sensory dysfunction and those with muscular atrophy and reduction of padding between the top layer of skin and underlying bone are prone to ulcers of pressure. These are factors contributing to immobility, impairment of sensory perception and cognition, decreased tissue perfusion, decreased nutritional status, friction, shear forces and increased humidity.

"*... Our luck is that we have the ancillary "L" that helps people to take care of them, get them from bed to get shower, putting oil on the back for not forming wound, and he looks as though they have neither sign of injury, despite getting a lot of time in bed " [Deponent sister].*

In the role of the difficulties faced by carers we highlight the communication. In the development of Friedreich ataxia speech is compromised. For caregivers, will begin a new process of care and communicate. For an act intrinsic to human existence. Even before birth we are sending and receiving messages from the world.

"*... the "G" and "M" can no longer speak, it is sometimes difficult to know what they so wished, but looking at the face, the face of them, we already know, because a parent can know what the child wants" [Deponent mother].*

Thus, the caretakers primarily the mother, have adapted to deficits in language and that over time, a unique form of communication is learned and shared between them, thus improving the quality of care. There is also the feminine intuition, because the mother can understand what the kids are wanting or feeling, even without express verbally.

We observed a link between the family care and sense of religiosity, seen as recognition by God, through the care, because they believe to receive a force, the same God who helps them in the conduct of care.

"*... Everything that I do for my children is for love, and if God gave me this cross to carry, I carry with faith and patience, and I'm sure he gives me the strength and soul that I help take care of them. I never discouraged and doubted the presence of God. And while he gives me strength, I can do what I do, but it is not easy, I think it's to support only with a spiritual force even" [Deponent mother].*

In all home visits to this family who were found with the television on, watching a mass. Once "A" said:

"*.... If I could, if I had a wheelchair, I would go to church every day, I am very devout to Our Lady, but I have no conditions, watch on television".*

We believe that from a system of beliefs, it is clear that the people are facing the most difficult situations in their lives. In this case, one can assume that there is an exacerbation religious belief and the search for supernatural forces to relieve the suffering caused by the process of care.

Perhaps, a form of transcendental facilitate the alleviation of the difficulties experienced in day to day, because the relationship between the formal health system and the caretakers are asymmetrical and off, not co-exist no interaction between them. Thus, there is the part of health professionals to work related to education on how to care for the patients with those, even in the scope of the strategy of family health.

"*... The doctors do not explain anything to us, don't teach us how to better take care of my brothers. What has been learned through other people, mainly from Dr. "H", researcher of Recife, which explained something, in relation to hygiene, nutrition and sunbath. We also got it through a dentist, with some explanation that she took a photocopy of the Internet" [Deponent sister].*

According Smeltzer & Bare (2002), currently the largest groups of people who need education to health are those with chronic diseases. Every family member responsible for care have the right to information, making it thus able to actively participate and assume responsibility for developing the best care. The health education can help patients and families to adapt to the disease, prevent complications, cooperate with the prescribed therapy and learn to solve problems when faced with new situations. This can also prevent the re-hospitalization; people with chronic diseases are frequently readmitted by the fact that they receive from relatives or caregivers an appropriate care.

Walking through the stages of transcription, and textualization, proposed by Meihy (2002) for studies of oral history, identified the "vital tone", which can be expressed thus: Brothers Melo, synonymous of hope.

FINAL CONSIDERATIONS

Looking at the Melo family, we knew it would be difficult and arduous the task of searching, we had fully aware that it would not be easy, even with the mutual-aid among us. We saw the need for family caregivers to speak, to express the feelings that were crystallized in their memories. Through the testimony of caregivers, we felt that there was great wealth in his words and that we had a lot to redeem it, since the outbreak of the disease until now.

The home visit was a therapeutic experience of operation for those involved but also a trick or a strategy of methodological support to the oral history. Through it had direct contact with reality, a true vision of a world of real carriers of

Friedreich's ataxia. We note the great chasm that separates the concrete context for these people lived, that prepared by professionals in the institution "competent" to assist them in their health care needs.

Through the testimony of his mother and sister, their caregivers, we feel quite exacerbated the presence of two main feelings: love and rebellion. For love, they no longer live their lives, to live for the sake of their children and siblings. Revolt to refer to the neglect and indifference to medical care and lack of guidance from professionals to promote the best care.

At the end we show caregivers as "special people" often abandoned to their own fate by health services, which are struggling with themselves, against the difficulties and often abandon the pleasure of living to help the other to continue living.

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MEANING OF CARING FOR RELATIVES OF PERSONS WITH FRIEDREICH ATAXIA

ABSTRACT

The Friedreich Ataxia is considered a rare, chronic-degenerative disease who often evolves to death in adulthood. The study aimed to understand the importance of caring for family members of individuals with Friedreich Ataxia. Qualitative research from oral history. The two subjects, mother and sister, refer significant changes in lifestyle, concern about not maintain care and becoming ill, difficulty in bringing them to the hospital, and also waiting for professionals with “goodwill" to serve them. As a supportive condition, they show an exacerbation religious belief and the search for supernatural forces to relieve the suffering caused by the care process. The home visitation was an experience and a support strategy. We note the wide gap that separates the real situation experienced by the family when facing the institution and its "competent" professionals in the care of their health needs.

KEYWORDS: Friedreich Ataxia; Family Relations; Comprehensive Health Care.

SIGNIFICATION DU SOIN POUR LES FAMILIERS DE PORTEURS DE L'ATAXIE DE FRIEDREICH

RÉSUMÉ

L'Ataxie de Friedreich est considérée une maladie rare, chronique et dégénérative qui évolue fréquemment vers le décès à l'âge adulte. L'étude s'est donné comme objectif de comprendre la signification du soin pour les familiers de porteurs de l'Ataxie de Friedreich. Recherche de type qualitatif à partir de l'histoire orale. Les deux sujets, mère et fille, relatent des changements significatifs dans le style de vie, la préoccupation quant à la possibilité de ne pas pouvoir maintenir le soin ou de tomber malade, la difficulté de se rendre jusqu'à l'hôpital, en plus de l'attente et des problèmes liés à la « bonne volonté » des professionnels pour les recevoir en consultation. Comme condition pour supporter la situation, elles démontrent une croyance religieuse exacerbée ainsi qu'une recherche tout aussi exacerbée de forces surnaturelles pour atténuer la souffrance provoquée par le processus de soin. La visite domiciliaire a été une expérience et une stratégie d'appui. Nous avons constaté le grand abîme qui sépare le contexte concret vécu par les familiers et l'institution et ses professionnels « compétents » lorsqu'ils sont reçus en consultation pour subvenir à leurs besoins de santé.

MOTS-CLES : Ataxie de Friedreich, famille, soins globaux-intégrés.

SIGNIFICADO DEL CUIDADO PARA FAMILIARES DE PORTADORES DE LA ATAXIA DE FRIEDREICH

RESUMEN

La Ataxia de Friedreich es considerada una enfermedad rara, crónica-degenerativa y que, a menudo, evoluciona a óbito en la edad adulta. El estudio tuvo como objetivo aprender el significado del “cuidar” para familiares portadores de la Ataxia de Friedreich. Investigación cualitativa desde la historia oral. Las dos declarantes, madre y hermana cuentan mudanzas significativas en el estilo de vida, preocupación de no poder mantener el cuidado y de enfermarse, dificultad en llevarlos hasta el hospital, además esperar que “les de la gana” los profesionales en atenderlos. Como condición tolerable demuestran una exacerbada creencia religiosa y en búsqueda de fuerzas sobrenaturales para disminuir el sufrimiento provocado por el proceso del cuidado. La visita domiciliar fue una experiencia y una estrategia de apoyo. Constatamos el gran abismo que separa el contexto concreto vivido por los familiares frente a la institución y sus profesionales “competentes” en la asistencia de sus necesidades de salud.

PALABRAS CLAVES: Ataxia de Friedreich, Relaciones Familiares, Asistencia Integral a la Salud.

SIGNIFICADO DO CUIDAR PARA FAMILIARES DE PORTADORES DE ATAXIA DE FRIEDREICH

RESUMO

A Ataxia de Friedreich é considerada uma doença rara, crônica-degenerativa e que, frequentemente, evoluem para óbito na idade adulta. O estudo objetivou apreender o significado do cuidar para familiares portadores de Ataxia de Friedreich. Pesquisa qualitativa a partir da história oral. As duas depoentes, mãe e irmã referem mudanças significativas no estilo de vida, preocupação de não poder manter o cuidado e de adoecer, dificuldade em levá-los até o hospital, além da espera e da “boa vontade” dos profissionais para atendê-los. Como condição suportiva demonstram uma exacerbação na crença religiosa e na busca de forças sobrenaturais para atenuarem o sofrimento provocado pelo processo do cuidado. A visita domiciliar foi uma experiência e uma estratégia de apoio. Constatamos o grande abismo que separa o contexto concreto vivido pelos familiares frente à instituição e seus profissionais “competentes” no atendimento de suas necessidades de saúde.

PALAVRAS-CHAVES: Ataxia de Friedreich, família, cuidado em enfermagem.