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Machado-Joseph Disease in the context of the person/family that experiences it: daily changes and future expectations

Doença de Machado-Joseph no contexto da pessoa/família que a vivencia: alterações cotidianas e expectativas futuras

Enfermedad de Machado-Joseph en el contexto de la persona/familia que la vive: alteraciones cuotidianas y expectativas futuras

Micheli Vieira Della Giustina[1], *Mara Glarete Rodrigues Marinho*[2], *Claudia Zamberlan*[3], *Elenice Spagnolo Rodrigues Martins*[4], *Silomar Ilha*[5]

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ABSTRACT

Objective: To assess the main daily changes and future expectations experienced by the person/family with Machado-Joseph disease. **Methods:** Exploratory research, a descriptive study of qualitative approach, carried out with a person with the Machado-Joseph disease and with five relatives, in a city of Rio Grande do Sul. The data was collected during a home visit conducted in April 2016 through semi-structured interviews and participant observation was subjected to content analysis. **Results:** Five categories emerged: (lack of) knowledge of the disease before diagnosis; knowledge of disease after the diagnosis; difficulties of diagnosis; changes experienced after diagnosis; expectations for the future with Machado-Joseph disease. **Conclusion:** It is necessary more investment by health professionals, especially nurses, in conducting studies aimed to help people / families living with Machado-Joseph disease.
Descriptors: Machado-Joseph Disease, Home Nursing, Family, Nursing.

RESUMO

Objetivo: Conhecer as principais alterações cotidianas e as expectativas futuras vivenciadas pela pessoa/família com a Doença de Machado-Joseph. **Métodos:** Pesquisa exploratória, descritiva de abordagem qualitativa, realizada com uma pessoa com a doença de Machado-Joseph e com cinco familiares, em uma cidade do Rio Grande do Sul. Os dados coletados durante uma visita domiciliar realizada no mês de abril de 2016, por meio de entrevista semiestruturada e observação participante, foram submetidos à análise de conteúdo. **Resultados:** Emergiram cinco categorias: (Des)conhecimento da doença antes do diagnóstico; Conhecimento da doença após o diagnóstico; Dificuldades do diagnóstico; Alterações vivenciadas após o diagnóstico; Expectativas para o futuro com a doença

[1] Nurse. Graduated from the Franciscan University Center. Santa Maria, RS, Brazil. E-mail: micheli-sm@hotmail.com

[2] Nurse. Master in Nursing. Lecturer at the Franciscan University Center. Santa Maria, RS, Brazil. E-mail: maramarinho@unifra.br

[3] Nurse. PhD in Teaching Nursing at the Franciscan University Center. Santa Maria, RS, Brazil. E-mail: Claudia_zamberlan@hotmail.com

[4] Nurse. Master in Nanosciences. Doctorate in Nanosciences at the Franciscan University Center. Lecturer at the Centro Universitário Franciscano. Santa Maria, RS, Brazil. E-mail: elenicemartins@unifra.br

[5] Nurse. Doctor of Nursing. Lecturer at the Franciscan University Center. Santa Maria, RS, Brazil. E-mail: silo_sm@hotmail.com

de Machado-Joseph. **Conclusão:** Torna-se necessário, mais investimento por parte dos profissionais da saúde, especialmente dos enfermeiros, na realização de estudos voltados a buscar auxiliar as pessoas/familiares que convivem com a Doença de Machado-Joseph.
Descritores: Doença de Machado-Joseph, assistência domiciliar, família, enfermagem.

RESUMEN

Objetivo: Conocer las principales alteraciones cuotidianas y las expectativas futuras vividas por la persona/familia con la enfermedad de Machado-Joseph. **Metodología**: Investigación exploratoria, descriptiva de enfoque cualitativo, realizada con una persona con la enfermedad de Machado-Joseph y cinco miembros de la familia, en una ciudad del estado del *Rio Grande do Sul* (*Brasil*). Los datos recogidos durante una visitación domiciliar realizada en el mes de abril/2016, por medio de entrevista semi-estructurada y observación participante, fueron sometidos al análisis de contenido. **Resultados**: Surgieron cinco categorías: (des)conocimiento de la enfermedad antes del diagnóstico; conocimiento de la enfermedad después del diagnóstico; dificultades del diagnóstico; alteraciones vividas después del diagnóstico; expectativas para el futuro con la enfermedad de Machado-Joseph. **Conclusión**: Es necesario más inversión por parte de los profesionales de la salud, especialmente de los enfermeros, en la realización de estudios direccionados al auxilio a las personas/familiares que conviven con la enfermedad de Machado-Joseph.
Descriptores: Enfermidad de Machedo-Joseph, Atención Domiciliaria de salud, Família, Enfermería.

INTRODUCTION

Machado-Joseph disease (MJD), also known as Spinocerebellar Ataxia (SCA) III, is characterized as a rare hereditary neurodegenerative disease, autosomal dominant,[1] which affects neurological structures responsible mainly for motor coordination and balance, but can also affect parts of deglutition and speech.[2] This disease can affect people of both sexes at different ages, averaging 40 years old, but may vary between extremes of age, such as seven to 70 years old.[3] The average life expectancy of a person with this disease is about 20 years old.[4] Death usually arises from pulmonary complications and from the cachexia that the person develops, which can occur between six and 29 years after the onset of the disease.[5]

MJD is considered the most prevalent SCA, accounting for 15 to 45% of the cases globally.[6] Nonetheless, they are considered rare diseases, with a prevalence of 0.3 to 2.0 per 100,000 people. Among the SCAs, the relative frequency of MJD in the world is as follows: Brazil (69-92%), Portugal (58-74%), Singapore (53%), China (48-49%), Germany (42%), Japan (28-63%), Canada (24%), USA (21%), Mexico (4%) and Italy (1%).[7]

In mainland Portugal, MJD has a prevalence of 1: 100,000 and is considered a rare disease, except for the Vale do Tejo area (1:1.000).[7] In the Azores, the prevalence of the disease is about 1: 2,402 and affects a group of 34 families. It is on the island of Flores (Azores) that the disease has a higher prevalence worldwide (1:239).[4]

In Rio Grande do Sul, the minimum prevalence of SCA III or MJD - the most common type in the state - is one case per 30,000 inhabitants. However, within the affected families, one case of ataxia may happen to every two people. Although there are studies based on the disease, there are currently no medications that have been approved for the treatment of MJD and the disease remains untreated.[7] In clinical practice, the use of levodopa, antispasmodic drugs and botulinum toxin, benzodiazepines, antidepressants, among others. .[8] However, for most drugs, there are no studies demonstrating efficacy, specifically in individuals with MJD / SCA III .[9]

However, there are methods that reduce some symptoms, allowing the delay of the signs.[7] Symptomatic interventions and rehabilitation of the physical aspect can weaken the manifestations presented by people with the disease, consequently improving the quality of voluntary movements and quality of life (QoL) of these people.[10] In this context, the multidisciplinary interaction is highlighted, as a possibility of allying the possible types of rehabilitation, both physical and psychological.

The biggest problem that involves MJD is the difficulty in walking and movements, so there are treatments in conjunction with motor physical therapy, which can strengthen the muscles and still help balance, reducing the risk of falls and helping in the difficulties related to daily living activities.[8,11] Speech therapy also plays an important role in helping to reduce dysarthria and dysphagia.[11] In this context, the collaboration of the different health areas with their different types of action is also of paramount importance to the comprehensive care for the person with the MJD and their families.

As it is characterized as a neurodegenerative disease, with the evolution of the disease, the person will need more and more constant care, demanding from him/her and his/her family preparation, patience, resilience and a lot of dedication. In this aspect, the importance of family support from diagnosis, treatment/care, until the signs and symptoms begin to manifest more clearly is essential.[12] In the process of cohabitation/care of a person with MJD, the family experiences diverse feelings, such as sadness, hopelessness, certainties and uncertainties about the diagnosis and the future. For this reason, it is considered that MJD, as well as other chronic and neurodegenerative diseases, affect not only the person who develops it, but all of its family context, justifying the need and relevance of this study in the care of the person and family with MJD.

It was not found in the literature studies related to nursing, only with other health areas, justifying its importance, also, to strengthen knowledge in the nursing area. Thus, it is expected that this study may produce subsidies to strengthen and expand the knowledge about MJD, which impacts the person / family.

Based on what was said above, the following question is asked: what are the implications of Machado-Joseph's Disease in the context of the person / family that experience it? In an attempt to answer the explicit questioning, this study aimed to assess the main daily changes and future expectations experienced by the person / family with Machado-Joseph's Disease.

METHOD

It is an exploratory research, of descriptive- qualitative approach, carried out with a person with MJD and with the relatives, living in a city in the Missions region. Initially, a telephone contact was made with the person with MJD and with the relatives, making the invitation to participate in the research. After the acceptance of the participants, a home visit was scheduled.

As inclusion criteria, the following were established: the person bears MJD, he/she must be considered a relative to the person with MJD and must have a physical / emotional conditions to answer the interview questions. As exclusion criterion: not being in physical / emotional conditions to answer the questions. They met the inclusion criteria, the person with MJD and five relatives form the corpus of this study, of whom, the father, the mother, a brother, two brothers-in-law.

Data were collected in April 2016 by means of a semi-structured instrument, containing two distinct parts, prepared specifically for this study and by participant observation. In the first part of the instrument, the personal data related to the relationship degree with the person with MJD, age, sex, education, profession and / or occupation were considered.

The second part addressed the following guiding questions: what do you know about MJD? Tell me about the day you received the diagnosis of MJD from your relative? How did you react to this news? Has there been any change in your life in particular, or in the family as a whole after the diagnosis of MJD? Which are? Tell me, what have you done to try to get along better with the relative bearing MJD? What are your expectations about the future of your relative with MJD?

All observations of the principal investigator during home visits were carefully recorded in a field diary. For the analysis and treatment of data, we used the Content Analysis,[13] which consists in discovering the sense nuclei that compose a communication, whose presence or frequency adds significant perspectives to the object of study. Thus, the operationalization of the analysis process followed the three steps of the method. In the first stage, an exhaustive reading of the data was sought, followed by the organization of the material and the formulation of hypotheses. Subsequently, the material was exploited, the raw data being encoded. In the third and last phase, the data were interpreted and delimited in thematic axes, according to the meanings attributed.[13]

The ethical and legal precepts involved in research with human beings were considered, according to Resolution 466/2012 of the National Health Council.[14] Clarifying the objectives and methodology of the research, the participants signed the Term of Free and Informed Consent (TFIC), in two ways, one remaining with the participant and another with the researcher. In the document, both parties were granted free access to the data collected. Participants were also informed that their names would not be disclosed and that they could withdraw from the study at any time without restriction.

The anonymity of the participants was maintained, the person with MJD was identified by the letter P (Person) and the relatives by the letter F (Family), followed by a numerical digit, according to the interview order (P1, F1, F2 ... F5). The project was submitted and approved by the Research Ethics Committee of the Franciscan University Center under the number 1.473.279.

RESULTS AND DISCUSSION

Of the six participants interviewed, four were male and two female, aged between 35 and 67 years. Regarding the educational level of the participants, two had completed higher education, two completed secondary education, one with incomplete secondary education and one with complete primary education. There were two retirees, one banker, one driver, one security guard and a housekeeper. Of those interviewed, one was the person with MJD, one was the father, one the mother, one brother and two were brother-in-law.

The analysis and categorization of the data formed five categories: (lack of) knowledge of the disease before diagnosis; knowledge of the disease after diagnosis; difficulties of diagnosis; changes experienced after diagnosis; expectations for the future with the MJD.

(Lack of) knowledge of the disease before diagnosis

It can be observed in the speeches of the participants the absence of previous knowledge about the disease, because they report that it was an unknown disease:

> *[...]Before discovering that I had the disease, I knew nothing about it. (P1)*

> *[...] I knew almost nothing, all that I know was after my brother-in-law contracted the disease. (F2)*

> *Before my brother contracted the disease, it was an unknown disease [...] (F3)*

> *Before my brother-in-law contracted the disease I had no notion, I only heard what my husband spoke about that [...] (F4)*

> *I did not know this disease, after my son took the AOP test, my daughter-in-law spoke of this disease, that they were suspecting, but I did not know anything [...] (F5)*

> *Before my son contracted the disease I did not know and knew nothing about it [...] (F6)*

In F2's report, one can observe more clearly that the knowledge about the disease came from the diagnosis and coexistence next to the brother-in-law with the disease.

Knowledge of the disease after diagnosis

The study participants recognize MJD as a progressive, hereditary, and incurable neurodegenerative disease. Following, the reports:

I know that it is a disease that has no cure, is progressive, genetic, hereditary and that has studies trying to discover the cure and stabilize the disease [...] (P1)

[...] I know that it is a hereditary disease, it passes from generation to generation and has no cure [...] (F1)

[...] today I know that it is a neurodegenerative disease that manifests itself generally from 30 years old and that affects the motor part and the balance of the person, is hereditary and can manifest itself in the new generations. (F2)

[...] I now know that it is a disease that has no cure and comes from the family, I just do not know which side. (F4)

[...] nowadays I know little about the disease, I know it's hereditary, but I do not know if it came from my side or my wife, because none of us feel anything (F5)

Participant F2 also adds the motor and balance commitments that the MJD causes to the person living with the disease. F4 and F5 end their speech stating that they do not know from which side of the family the MJD condition was inherited.

Difficulties in diagnosis

In the reports, it is evident the difficulty that the participants found to obtain the diagnosis of MJD:

[...] it started with loss of balance when I was 32, I went to several doctors and nobody could figure out what I had. I only received the diagnosis at 38 years, six years after the onset of my symptoms. (P1)

[...] my brother spent years looking for doctors because of the symptoms and nobody discovered anything; I realized that this was unknown even for doctors. (F2)

[...] he spent several years trying to find out what he had, I knew it was a serious illness, but he went to several doctors and each one said something different and they medicated him, but he was still the same, stumbling and falling. The doctors studied medicine and didn't know about it, there 's not a chance that I, someone that didn't study anything, would know. (F5)

In the reports it can be noticed that, after the onset of the symptoms of MJD, the person with MJD sought several professionals, but because it still was an unknown disease, this diagnosis was postponed. In the speech of P1 it can be noticed that from the beginning of the symptoms of the disease, presented by loss of the balance, six years have passed until the diagnosis confirmation.

Changes experienced after diagnosis

The life of the person with MJD, according to the participants of this research, underwent several changes, such as the end of the marriage, the separation of the children and moving to the parents' house:

After the diagnosis, I divorced, came to live with my parents, it is very difficult to live with the disease and stay away from the children. (P1)

In my life it did not change much, but it did in his. He separated and went to live with his parents in another city [...] (F1)

Yes, the concern increased, I tried to research the disease a lot to see if there is any news about the treatments. My brother broke up and came to live here in the city with my parents, here in the city he is the only case that they know with MJD, it seems that now they have discovered another person with the disease [F2]

[...] after he separated and came to live with my mother-in-law and father-in-law, I began to live with him more, I was nervous and worried because he had days he was very unstable, he stumbled and hurt himself a lot at home and in other days he was better. My husband several times thought he had the disease too, everything his brother felt, he felt it too. This was the time I was most concerned about [...] (F3)

[...] for him (person with MJD) it was very difficult because of the children, the separation! (F4)

My son separated from his wife and children, he moved in with us. Here at home, my wife does everything for him, serves the food, helps to walk when he is unbalanced, washes his clothes, he only has to worry about the treatment of the disease, which is physiotherapy and take the remedies. (F5)

In the relatives' reports, the increased concern is explicit. F2 states that after the diagnosis of MJD in his brother, he has sought to research the disease in search of novelty about the treatments. F3 demonstrates concern about his brother-in-law's imbalances with MJD. Moreover, she says that her husband, the brother of the person with MJD, thought he had the disease too, because all the signs that the brother with MJD displayed were also recognized in the brother.

Expectations for the future with Machado-Joseph disease

Regarding future expectations, all the participants expressed the desire/hope regarding a treatment that stabilizes the evolution of the disease or cure it:

I hope they find out the treatment soon enough, to stabilize the disease and maybe even cure it, because I do not want my children to go through what I'm experiencing. (P1)

I know they have studies in Porto Alegre about some medication to improve or even cure people with this disease. My brother-in-law will hardly be able to use this medication because, generally, these studies take time, but I hope that one day this disease also has a treatment and, who knows, the cure. (F1)

The expectations are good, the Brazilian associations are doing a lot of research on the subject, I hope that soon, about two years, there will be a medication to stabilize the disease and not let it evolve, I hope my brother can still use that medicine . (F2)

We must have hope, there will never be a cure, but at least some remedy that slows down the disease. (F3)

I want them to discover the cure, because I do not want to see more people suffering like my son. (F4)

I hope they find the cure for the disease, they say they are developing a remedy to stabilize the disease, so I hope this will be soon for my son to use and for his children not to bear the disease because there is a high chance of their children contracting the disease. (F5)

In the report of P1, the person with MJD, the concern with children is evident, demonstrated by the desire that they soon find a cure so that they do not go through what they are experiencing.

The MJD has not yet had a specific therapy capable of stopping the progression of the disease and the therapeutic challenges continue to attract researchers in the field of spinocerebellar ataxias.[9] In this context, Brazilian researchers have excelled in research in search of new knowledge about this disease.[15] However, even with the numerous investments, the MJD is still little discussed and known by society in general.

This question can be observed in the present research, since participants reported absence of previous knowledge about the disease, referring to it as unknown. One participant made it clear that the knowledge about MJD came only after the diagnosis and coexistence with his relative. At the time of the research, it was observed that participants already had knowledge about MJD, referring to it as a progressive, neurodegenerative, hereditary, and, until now, incurable, which causes motor and balance impairment.

MJD is one of the spinocerebellar ataxias that is part of a complex group of degenerative diseases, which are characterized by cerebellar degeneration and greatly affect the physical and cognitive aspects. It has a wide range of neurological symptoms, including ataxia, dysarthria, oculomotor disorders, extrapyramidal signs and several other clinical manifestations that reinforce the idea of a degenerative process.[2] In addition, psychiatric disorders, sleep dysfunction and olfactory disorders, as well as cramps, fatigue and myalgia can occur.[16-17]

Even with the signs and symptoms, the diagnosis of MJD is still difficult for professionals because, although there is an increase in the genetic findings with greater ease for the tests, it is still difficult to carry out a definitive diagnostic approach for neurological and hereditary diseases.[18] This data is in line with those of the present study, which evidenced the difficulty experienced by the participants until the diagnosis of MJD. Some participants reported having searched for several professionals, but that because it was an illness still unknown, this diagnosis was postponed. One of the participants reported that from the onset of symptoms of MJD in the family until the diagnosis, six years had elapsed.

Thus, it is evident that, because MJD is a disease that is difficult to diagnose, when evaluating a patient, professionals need a clue, a mark, a signal, in order to request a specific genetic test. Thus, it is necessary that they have a broad knowledge to be able to capture signals that sometimes appear in a discrete way, such as a palpebral retraction. Thus, they should value the patient's clinic, along with some neurological exams, which may guide genetic testing to specific diagnosis.[19]

The life of the person with MJD, according to the participants of this research, underwent several changes, among them the termination of the marriage, the rupture of the daily coexistence with the children and the transfer of the person with MJD to the parents' house. Neurodegenerative diseases greatly affect several aspects of the person's life and their families. However, a study developed with families from Rio de Janeiro affected by MJD showed that their participants, instead of only talking about the negative physical consequences inherent to the disease, were more willing to evaluate the life they may still have when living with MJD.[20]

The results of the research describe that the close relationship with parents and other relatives, who had already developed MJD but who previously had a professional career or who still had conditions to work, was taken into account in the process of acceptance and coexistence with the disease. They report that although they have become unstable with regard to balance and gait, also his ability to hold objects, they had the opportunity/time to raise their children before they became ill.[20]

These examples can be used in practice with the person / family with MJD to help them think positively, even if they are living with a disease that sometimes presents in a frightening way, since it does not have a treatment that stagnates its evolution or cure it.

Regarding future expectations, all the participants expressed the desire/hope regarding a treatment that stabilizes the evolution of the disease or cure it. They demonstrated the concern with children and the desire that they find the cure so that they do not go through what the person with MJD is experiencing. In a study conducted in the Azores Islands, Portugal, the authors identified that 61% of people at risk for MJD reported that the risk condition induces them to not wish having children because they know that they are likely to inherit the gene and develop MJD.[21]

However, in the case of the person with MJD, a participant in this study, this conscious choice was not possible, since when the symptoms started, the MJD bearer already had children. What can be done are tests of molecular genetics in the offspring to see if there is or is not the gene for MJD.

CONCLUSION

The accomplishment of this study is considered satisfactory, since it was possible to assess the main daily changes and the expectations experienced by the people / families with MJD. The main changes were the termination of the marriage, the separation of the children and the transfer to the parents' home, an increase in the concern regarding the balance and health of the person with MJD and the search for understanding more about the disease. The expectations of the person / family with MJD, in relation to the future, refer mainly to the desire / hope regarding a treatment that stabilizes the evolution of the disease or that cures it.

This study presented limitations inherent in any qualitative study, which, by nature, does not intend to generalize its results. Therefore, it is highlighted the impracticability of reproducing the data presented here if the method is applied in another reality or scenario, since they represent unique experiences of the relatives of a person with MJD. Another point to be highlighted is the scarcity of studies on the Machado-Joseph Disease, especially in the nursing area, which indicates the need and relevance of this study for the advancement of nursing knowledge as a science and profession in this context.

The results of this research can foster discussions and reflections among nursing / health professionals, family members, families and society as a whole, with a view to better care for the person with MJD and the family. Therefore, understanding the complexity that surrounds the MJD theme in the family environment, it is suggested to carry out more studies aimed at helping the people / families that experience this process.

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Contact of the corresponding author:
Silomar Ilha
Centro Universitário Franciscano - Conjunto III,
Rua Silva Jardim, nº 1175.
ZIP-code: 97010-491
Santa Maria, RS, Brazil
E-mail: <silo_sm@hotmail.com >