

Rare diseases: diagnostic and therapeutic journey of the families of affected people

Doenças raras: itinerário diagnóstico e terapêutico das famílias de pessoas afetadas

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Keywords

Public health nursing; Pediatric nursing; Maternal-child nursing; Rare diseases; Health services

Descritores

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Abstract

Objective: To characterize the diagnostic and therapeutic journey of families of people with rare diseases within the network of Brazilian public services.

Methods: This was a qualitative research project. The bio-ecological theory of human development, by Urie Bronfenbrenner, was used to understand the data. The research instrument was a semi-structured interview, and data were analyzed using the content analysis method.

Results: Three central themes were grouped: "Journey of families in search of a diagnosis"; "Journey of families after the diagnosis of the disease"; "Journey of therapeutic maintenance".

Conclusion: The access to specialized services enabled diagnosing of the rare disease. The treatment was a challenge, because there were few drugs available within the therapeutic options for these diseases. Legal recourse was essential for therapeutic access and maintenance.

Resumo

Objetivo: Caracterizar o itinerário diagnóstico e terapêutico realizado pelas famílias de pessoas com doenças raras, no âmbito da rede de serviços públicos brasileiros.

Métodos: Trata-se de pesquisa qualitativa. Utilizou-se a teoria bioecológica do desenvolvimento humano, de Urie Bronfenbrenner, para a compreensão dos dados. O instrumento de pesquisa foi uma entrevista semiestruturada, e os dados foram analisados pelo método de análise de conteúdo.

Resultados: Foram agrupados três núcleos temáticos: "Itinerário das famílias em busca do diagnóstico da doença"; "Itinerário das famílias no pós-diagnóstico da doença"; "Itinerário de manutenção terapêutica".

Conclusão: O acesso aos serviços especializado possibilitou a obtenção do diagnóstico da doença rara. O tratamento foi um desafio, pois há poucos medicamentos disponíveis na escolha terapêutica para essas doenças. A judicialização foi fundamental para o acesso e a manutenção terapêutica.

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Introduction

The birth of an ill child can cause changes within family life, giving rise to odd behaviors and feelings, which reflect the impact of coping with this new condition.

Families are faced with the need to readjust the roles of its members, to take on new responsibilities beyond the usual ones when a child is born, and to seek social and health services that offer them social, financial and emotional support. Rare diseases are particularly considered a continuous learning experience, not only for the affected individuals, but also for their family members who face numerous challenges, especially in the micro context of the family, and in the relationship with the health services to which they undoubtedly will be connected for a long time.⁽¹⁾

Families of people with rare diseases are usually treated unequally in health services, but not exactly due to prejudice. Their rights related to access to quality health services, to equity, to resolution, and to integrality of interventions are not always respected, either because the services do not have the technological capabilities to manage a rare condition or because professionals are not prepared. Thus, the “negotiation” of family rights is denied from the beginning of the child’s life. After the diagnosis of the disease, the battle still continues, as these families need to seek ways to venture outside of the health system to continue with the proper treatment, which is usually expensive and often only possible through judicial means.

Importantly, some diseases are classified as rare due to the low frequency with which they occur in the population. However, the affected individuals do not always receive an early diagnosis and, moreover, there are few therapeutic options and rare scientific research in this area.⁽²⁾ These diseases contribute to increased morbidity and mortality, particularly in children. The risk of preventable complications and deaths due to a late diagnosis can weaken the entire family system of the affected person.⁽³⁾ There are more than 7000 different types

of rare diseases. This amount is unstable, since it tends to increase, with the addition of five new diseases listed weekly. Thus, rare diseases are much more common in society than the name apparently suggests, because they constitute 6-10% of disease worldwide.⁽⁴⁾

The advancement of research in genetics, particularly on the human genome, made it possible to better understand the world of rare diseases. It was found that 80% of rare diseases to date are of genetic origin, involving one or several genes or chromosomal abnormalities that represent between three to four percent of births. Others are caused by viral or bacterial infections or allergies, or by degenerative, proliferative or toxic processes (chemicals, radiation etc.). In developed countries, infant mortality among people with rare diseases reaches 30%. This percentage may be even higher in Brazil, since many of these children are not properly diagnosed and therefore do not receive appropriate treatment.^(3,4) As an example, we cite the case of cystic fibrosis, because the clinical trajectory of diagnosed patients is often identified as intermittent pneumonia.

The diagnostic and therapeutic journey of people with rare diseases can be a major challenge in relation to health services. The therapeutic journey is characterized by a succession of steps, from the onset of the disease, with the use of traditional medicine (self-medication and traditional healers) and modern medicine (modern care facilities). It represents the path to try to solve health problems, according to individual and socio-cultural practices.⁽⁵⁾ This is a concern of studies that seek to know which paths users go through when they do not identify with the health system schemes or flows.^(5,6) To date, there are no studies about the diagnostic or therapeutic journey of families of people with rare diseases, which evidences little research conducted on this phenomenon.

Considering the difficulties of families of people with rare diseases, in relationship to the health services and the scant research performed in the field of nursing, this study aimed to characterize the diagnostic and therapeutic journeys

usually taken by the families of people with rare diseases within the network of public services.

Methods

This was an exploratory study with a qualitative approach, whose theoretical framework for data interpretation was the bio-ecological theory of human development, by Urie Bronfenbrenner, encompassing the four elements of the bio-ecological model: process, context, person and time.⁽⁷⁾

This study was developed in three reference services for rare diseases in the state of Rio Grande do Sul, in Southern Brazilian. The study participants were families of patients with rare disease enrolled in public services that provided services to people with mucopolysaccharidosis, cystic fibrosis and phenylketonuria. They included 16 families of people with rare diseases, represented by 14 mothers, one father and one grandmother.

The selection of the participants was performed by the service coordinators, who invited families to participate in the study. We sought to obtain a varied sample, in relation to families residing within the countryside and the capital, time of diagnosis, and age of those affected.

Semi-structured interviews were conducted, recorded with the consent of the participants in a single meeting, at home or in the health services. The duration of the interview was 50 to 100 minutes. Data were collected over five months, and interrupted when saturation was reached, or when enough information was provided for the researcher to respond to the objective of the study. We used a script with guiding questions grouped into four sections: (1) identification of the family representative interviewed; (2) history of the family relationship with the rare disease; (3) context of family life; and (4) family interaction with network health services. Data were analyzed by reading the empirical material, seeking the essence of the discourses using the content analysis technique.⁽⁸⁾

During the pre-analysis phase, first contact with the material was established through reading and rereading of interviews, and the encoding of information. Then the thematic nuclei were delimited, using as references the regularities and patterns identified in the words, phrases and manifested behaviors, which translated the diagnostic and therapeutic journeys of these families.

The development of the study met national and international standards of ethics in research involving human subjects.

Results

According to the adopted reference, the diagnostic and therapeutic journey of families of people with rare diseases was represented by three thematic nuclei, as shown in figure 1.

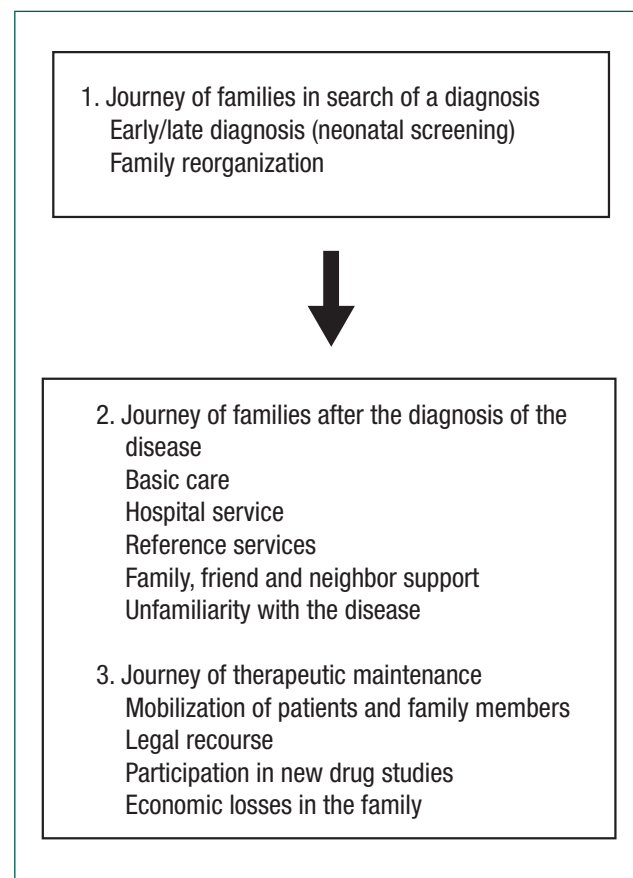


Figure 1. Journey of families of people with rare diseases

The thematic nucleus, “Journey of families in search of a diagnosis” addressed the journey of families from the moment they realized the need to seek health care until the diagnosis of a rare disease. Within the families, men mostly conducted their role as providers; mothers, who were more available to go to appointments, trips and who had a wide knowledge about the child’s health, were defined as the main characters in this stage of family life.

The thematic nucleus, “Journey of families after diagnosis of the disease” referred to the three health care levels: specialized services, hospital services, and primary care. Families also sought qualified professionals to complement the care of the ill person. There was the need of families to share household chores within its members or close friends.

The thematic nucleus, “Journey of therapeutic maintenance” identified the ways of access to treatment, which should be maintained throughout life, especially via legal recourse. Families had significant economic loss due to unfamiliarity with rare diseases within the public health system.

Discussion

The limits of this research were related to the investigation only of the families of people with three rare diseases (mucopolysaccharidosis, cystic fibrosis and phenylketonuria), which does not allow for the generalization of the journey of families with other rare diseases. However, the selection of families residing in the capital and the countryside strengthens the results, enabling inference that the families residing in the countryside, despite the various difficulties in their journeys, can meet their health needs.

Nurses, as health educators, can act on guidance to families and patients in predictive tests (e.g. newborn screening), care during treatment, and genetic counseling. In addition, they can identify and mobilize non-governmental services, health and social services, easing the isolation

that these families manifest in their diagnostic and therapeutic journeys. They can also promote the exchange of experiences among families experiencing the same condition, considering the need to project themselves on others, which can create opportunities for a safe environment for these families.

The Basic Health Unit was the first service used by eight families of this study. With the altered result of the newborn screening, they were notified of the possibility of rare disease in the newborn child. At that time, the families felt powerless, given the little information they were provided about the situation. Consistent with this observation, a study with parents of children with congenital hypothyroidism and cystic fibrosis showed that 54.5% of parents wanted more information when they were notified about the altered result of the newborn screening.⁽⁹⁾ However, studies performed with parents of screened children show that guidance provided about the exam during the prenatal period can facilitate understanding of an altered result.^(10,11)

Explanations can be given during prenatal care on the procedures and meanings of an altered test result, so that the family does not feel isolated from the context of neonatal screening.⁽¹⁰⁾ Moreover, nurses should be imbued with their health educator role and, as leaders of the Family Health Teams, provide professionals with knowledge about the current condition of rare diseases in their community.

For other families in this study, the difficulty focused on the investigation of the first symptoms, which seemed to be common in childhood, such as abdominal pain, cough, diarrhea and weight loss. Coming and going to health services became stressful moments when the diagnostic and therapeutic possibilities ended with the child’s health involution. In accordance with the results, a study with parents of children with rare diseases showed significant emotional aspects (stress, anxiety, doubt, anger and despair) in the family micro context in relation to health services.^(12,13) Another study performed in Denmark, in order to explore the interpretation of the parents at the point of the first signs and symptoms presented by children with

chronic diseases, showed that panic, anguish and anxiety were the main feelings during the course of recurrently going to the health service when the implemented therapy did not recover the child's health.⁽¹⁴⁾ Under these conditions of the family's disease experience, health services of greater complexity were needed to investigate the disease and stabilize the child's health.

The families in this study found diagnostic support and multidisciplinary monitoring in referral centers, usually located in capital cities. From the connection with these services, they felt accepted by qualified professionals. Importantly, one of the reference services cited by these families was linked to a university hospital created with funds from research funding agencies, which features the hitherto informal characteristics of care for those with rare diseases in Brazil.⁽⁴⁾ A reference center for rare diseases can offer a set of specific actions, such as: early diagnosis for the newborn, treatment and rehabilitation, multidisciplinary therapeutic follow-up, and genetic counseling for affected individuals and their families.⁽¹⁵⁾

The place of residence of eight families showed the need for qualified services close to those who do not live in large cities, where the reference services are usually located. A decentralization policy could ease the isolation and social limitations that these families experienced in accessing health services. Similarly, the provision of professionals to meet and work with people and families living with a rare disease of one of its members can contribute so that nurses can help families with their health care network journey i, from Primary Care to the reference services. Moreover, considering genetic counseling as part of the nursing process in the classification of nursing interventions, it follows that it can be a link that favors the professional-family interaction,⁽¹⁶⁾ softening the impact on the microsystem, mesosystem and exosystem.

Regarding high-cost treatment, the families of this study described their facilities and difficulties. Social activism of cystic fibrosis patient associations in Rio Grande do Sul was one characteristic that facilitated the access to high-cost treatment. This profile of social mobilization inherent in rare

diseases in the world shows that the theme has not only medical issues, but a social problem related to basic human rights of affected people.⁽¹⁷⁾ For all families of people with phenylketonuria in this study, the access to treatment had a significant judicial route. This is a questionable situation, since phenylketonuria is one of the rare diseases contemplated in specific clinical protocols, ensuring access to treatment.⁽¹⁸⁾

Yet, on the journey for the maintenance of a high-cost treatment, two families participated in research in experimental stages. Although there was no guarantee of the effectiveness of the treatment, these families referred to the research as a hope for cure and/or prevention of disease progression. However, it is observed that obtaining the correct diagnosis of a rare disease is not the end of the trajectory of these families. The judicial struggle for access to treatment is a constant in their history.

Conclusion

The access to specialized services enabled the diagnosis of a rare disease. The treatment was a challenge, since there are few drugs available within the therapeutic options for these diseases. Still, seeking legal recourse was essential for access to expensive medicines. In addition, it was realized that the Primary Care services were significant in the process of the diagnostic and therapeutic journey through the neonatal screening test.

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Collaborations

Luz GS; Silva MRS and DeMontigny F state that they contributed to the concept and design, analysis and interpretation of data; article writing, critical review of the relevant intellectual content, and final approval of the version to be published.

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