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Persistent suffering: Living experiences of patients with rare disease: An interpretative phenomenological study

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Abstract:

BACKGROUND: The low prevalence of rare diseases has caused the need for studies in this field to be neglected. Regardless of the prevalence of rare diseases, many people around the world have to live with the medical, psychological, and social consequences of their condition. Individuals with rare diseases may face challenges that are different from those experienced in more common medical conditions. The life experiences of patients with rare diseases have not been sufficiently investigated. The purpose of this study was to discover the meaning of living as a person with a rare disease.

MATERIALS AND METHODS: This interpretative phenomenological study was conducted in 2021–2022 on 10 patients with one of the rare diseases (registered in the Atlas of Rare Diseases of Iran). Based on purposeful sampling, people with rare diseases living in Mazandaran, Golestan, and Tehran provinces were invited to participate in the study. Data collection was done using open and semi-structured interviews. The research question was exploring understanding the experience and meaning of life as a person with a rare disease. Van Manen's interpretive phenomenological approach was used to analyze the data, and the criteria of validity, transferability, and verifiability were used to ensure the trustworthiness of the research.

RESULTS: The five main themes "permanent suffering, such as a bird in a cage, rejection, immersion in the whirlpool of thoughts, losing the feeling of life", and 10 sub-themes "nightmare, giving up, deprivation, limitation, worthlessness, being stigmatized, dark vision, confusion, continuous regret, and inferiority feeling" were extracted.

CONCLUSION: The results of this study show that the suffering of the disease casts a shadow on all aspects of a patient's life with a rare disease. The effects of illness, disability, limitations, and exclusions had created a human being in a cage, whose right to live like others has been denied.

Keywords:

Lived experience, phenomenology, qualitative study, rare disease

Introduction

Illness is a profound and life-changing event. Getting sick or being diagnosed with a serious illness requires that the sick person and those around him stop and evaluate their lives from now on.[1] Rare disease has a frequency of less than 5 in 10,000 people. So far, around five to eight thousand rare diseases have been identified worldwide. Most rare diseases are considered genetic disorders, even if their symptoms do not appear very soon, they will accompany a person throughout his life.[2]

Currently, 332 types of rare diseases have been identified in the country (Iran), and about 4,000 rare disease patients have been registered in the electronic registration system of rare diseases in Iran.[3] Rare diseases are also known as orphan diseases, and that is because there is no effective

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treatment method, and all prescriptions and treatment methods used are primarily aimed at reducing the symptoms and complications of the disease. According to the European Rare Diseases Foundation's estimate, except for 80% of these diseases, which have a genetic origin, the rest are the result of some kind of infection and allergies.[4]

Rare diseases, starting early in life, or causing chronic pain, movement, sensory or mental disorders, reducing a person's independence, and increasing the threat to life, cause difficulties for patients. Most rare diseases are chronic; thus, patients endure a lot of physical and mental suffering depending on their characteristics.[5]

There is little information among health care professionals about these diseases and their patients. A suggested way to improve the situation of people with rare conditions is to increase awareness of rare diseases. The challenges of the lack of knowledge in the health care system about rare diseases are diagnosis errors, errors in bedside care, delay in diagnosis, and lack of adequate information.[5-7]

Due to the little research about these diseases, patients and their families benefit less from the healthcare system. Because nurses have a vital role both in the medical care system and in society, they need to have more information in this field to empower them in the role of care, training, counseling, and support. Considering the lack of knowledge about rare diseases, nurses can play a vital role and add to the scientific body of rare diseases with research in this field.

The qualitative method is a method for a deeper understanding of experiences and searching for people to understand the meaning or nature of experiences from an ideal point of view.[8] In qualitative research, a better understanding is obtained due to the creation of new significant distinctions to get closer to the phenomenon under study. Phenomenology in nursing is related to the subject of the mind; the living person in the body with the experience of health and illness is the nurse and the patient.[9] The goal of phenomenology is to describe the meaning of this experience—both in terms of what is experienced and how it is experienced.[10] Heidegger's hermeneutic phenomenology has been widely used to understand the meaning of lived experiences in health research.[11]

Medical researchers must find out how the patient experiences his illness and what meaning he gives to it. In contrast to the mechanical concept of the body, the "living body" is considered the axis of human mentality and being in the world and includes all dimensions of human existence. Illness is not just a local disorder in a part of the patient's body; it is an all-encompassing existential concern that disrupts all daily activities, habits, plans, and goals of patients. The body is the center of our lived experience. When a person gets sick, not only the physical part is involved, but also the being-in-the-world is disturbed. Illness changes the patient's existence, and if it can be diagnosed, it can affect the relationship of others with the patient. Therefore, examining the disease from an existential point of view is necessary. Phenomenology provides a comprehensive explanation of the human experience of illness to express and describe the experience and create meaning.[12]

A review of the literature showed that the phenomenon under investigation (living with a rare disease) is a phenomenon with socio-cultural dimensions, and the majority of the evidence is related to the socio-cultural context other than Iran. Iran has its own cultural and social characteristics. It is necessary to conduct a qualitative study to achieve the various dimensions of this phenomenon and increase the body of nursing knowledge in the field of rare patient experiences.

The present study is beyond the mere description of the participants' experiences and seeks to explain and describe the lived experiences of living with a rare disease; interpretive phenomenology was the basis of the researcher's work. The researcher has the experience of taking care of a member of his family with a rare disease and also has a rare disease himself, and two other researchers are members of a rare disease association; therefore, in addition to the necessity of the subject in the society, to conduct the study, there has also been a tendency in the field of rare diseases. Because the results of this study include the challenges of living with a rare disease, the findings can be the voice of rare patients for the health care providers, and health officials, and in the case of translation of knowledge for the general public.

Materials and Methods

Study design and setting

A qualitative study of interpretive phenomenology was conducted in 2021–2022 on patients with a rare disease approved by Iran's Rare Disease Foundation.

Study participants and sampling

Determining the sample size accurately in qualitative studies cannot be predicted in advance and usually in phenomenological studies interviewing 6 to 20 participants is sufficient.[13] The participants of this research included four men and six women who were selected from a wide range of ages. Sampling was purposeful. Iranian patients suffering from a rare disease who were willing to talk about their lived experiences were invited to participate in the study. Sampling with

the maximum diversity in terms of age, sex, marriage, education, and type of occupation, the type of disease was rare. Inclusion criteria were having a rare disease, age 18 years or older, and sufficient ability to express one's experiences. No specific exclusion criteria were considered. Deep, interactive, and semi-structured interviews were used to collect the data.

Data collection tool and technique

The researcher started the interview with a general and open question and tried to have minimal interference in the interview process. Exploratory questions were asked to clear the ambiguity in understanding the experiences of research participants The interview guide is given in Table 1. Each participant was interviewed at least twice. The duration of each interview session varied from 45 min to 120 min. The interview process was recorded according to the coordination and permission of the participants. To enrich the interviews, exploratory questions such as "Give an example, why? How did you feel" were used in the interviews as necessary.

Data analysis method: Six steps of Van Manen's interpretative phenomenology were used to perform and analyze the data: 1) turning to the nature of lived experience; 2) exploring the experience under study as it was lived; 3) reflecting on the intrinsic elements that characterize the phenomenon; 4) the art of writing and rewriting: 5) maintaining a strong and directional relationship with the phenomenon: 6) correspondence of the context of the research by considering the components and the whole.[14]

Scientific accuracy and validity of data (Rigor)

To ensure the reliability of the research (accuracy and appropriateness of the research methodology), four criteria were used: 1) validity, 2) reliability, 3) transferability, and 4) verifiability.[15,16]

Ethical consideration

All ethical issues in human research were considered. This study was approved by the Ethics Committee of Golestan University of Medical Sciences (IR.GOUMS.REC.1400.117).

Table 1: The interview guide

Question
What comes to your mind when I say rare disease?
What is it like to be a patient with a rare disease? Explain it to me.
Tell me about your life experiences as a patient or being a rare patient.
Tell me about your life experiences living with a rare disease in different situations in your personal and social life.
Tell me about your life experiences as someone living with a rare disease considering medical situations.

Results

Of the 10 individuals who participated in this study, 4 were male and 6 were female [Table 2].

According to the results of the study, 5 main themes, 10 sub-themes, and 53 mini-units were extracted [Table 3].

Permanent suffering

The main theme "permanent suffering," was explained with two sub-themes, "Being like a nightmare" and "to give up." This theme and its sub-themes represent the life experiences of people with rare diseases and are given with direct quotes in quotation marks. The participants' experiences indicated a life of pain and suffering caused by the disease for their whole life, which has been unbearable suffering, without hope, with a bag full of pain, fatigue, and sadness.

"This disease will bother me for the rest of my life; what should I do with all this difficulty, what should I do with all this pain and suffering for the rest of my life…I am a collection of pain and suffering; the disease is attached to me; I will live with the disease until the end of my life." (P: 10)

In any way, suffering from a rare disease has imposed the effects of suffering on patients, which could be caused by the complications of the disease, people's lack of familiarity with the disease, or its incurability.

a. Like a nightmare: The sub-theme of being a nightmare shows that the disease had shadowed them like a nightmare throughout their lives, and they had little to live like other people.

Participants described various challenges of living with their disease. The disease was like a garment on their body that was never removed from their body; it was with them in different periods of their lives, it never ended, it caused a lot of mental suffering, and as a result, they experienced gradual decay and desperation to live with the disease.

"the pain and suffering of our disease doesn't like us at all. It says that I will be with you until the end of your life, whether you want it or not." (P: 10)

These patients had experienced the disease in their lives as if it was inseparable from them and they should not wait in the hope of recovery because they lose years of life and they wanted their family and surrounding people to see and accept them as they are with no hope that they will get better, to live like other people because there is no hope for them to recover and get rid of the disease.

Table 2: Demographic and clinical characteristics of participants in the research

Participant code	Name of disease	Age	Gender	Education	Duration of disease
1	Alkaptonuria	28	Male	Diploma	Since birth
2	Dent	31	Male	Primary school	Since birth
3	CIPA*	21	Female	Illiterate	From 2 years old
4	Early-onset Parkinson's	57	Female	Illiterate	14 years
5	Achondroplasia	18	Male	Diploma	Since birth
6	Achondroplasia	18	Female	Diploma	Since birth
7	Neurofibromatosis	50	Female	Illiterate	From 10 years old
8	Ichthyosis	49	Male	Diploma	Since birth
9	Neurofibromatosis	27	Female	Diploma	Since birth
10	Epidermolysis bullosa	33	Female	Diploma	From 2 years old

*Congenital insensitivity to pain with anhidrosis

Table 3: Themes, sub-themes, and mini-units

Mini-units	Sub-themes	Themes
Like wearing a dress, living with a disease is not a 2- or 3-day disease, I will burn and die with this disease, the disease has no final date, the chronicity of the disease, exhausting mental pressure, being exhausting, continuous physical disability, fatigue, pain, suffering	Like a nightmare	Permanent suffering
Desperation, meltdown bit by bit, to be incurable	To give up	
Communication deprivation, emotional deprivation, social deprivation	Deprivation	Like a bird in a cage
Disability at a young age, poverty	Limitation	
To be ignored, to leave the world (recluse), to be isolated, to be humiliated, to be worthless, to have low self-esteem, to be pushed out of society, to be pushed to the sidelines, family support diminishing, government support diminishing, to be shy, to be disillusioned, heartbreak	Worthlessness	Rejection
Being pointed out in the society, being strange to people, taffeta separate, as if I came from the moon, being different from others.	Being stigmatized	
It is impossible to see the achievement of dreams, to have a vague future, to have a dark future, to have nothing but a black future, not to have a future for yourself.	Dark vision	Immersion in the whirlpool of thoughts
Humiliation, confusion from the pain of the times, astonishment from the bad behavior of the society	Confusion	
Regretting, losing dreams	Continuous regret	Losing the feeling of life
Feeling ugly, being a black fish in an ocean of beautiful fish, feeling like an ugly fish among beautiful fish, a second-class citizen, being ashamed.	Inferiority feeling	

"I also have a humorous definition for myself, which says that whenever Pinocchio becomes a person, we also become a person." (P: 8)

"These kinds of diseases are not solved in 3 or 3 days, they affect you for a lifetime, and little by little your body becomes wasted. You think you are not like the others, your tolerance decreases, and you do not laugh. I used to cry with every laugh..." (P: 1)

b. To give up: Participants experienced giving up in life along with desperation, feelings of hopelessness and depression, and tiredness. Life has been a collection of pain and suffering for them, although they tried to be in a good mood, many worries along with mental pressures have led them to a sense of despair and destruction, unlucky at birth, misfortune during life, and not being like others, melting bit by bit and becoming impatient.

A rare patient is like a butterfly with a broken wing or like someone who is left in a dark cave and is cut off from life and from heaven and earth. They felt that they were melting bit by bit and had no choice but to endure, although they reached a stage where they no longer had the patience to live. The heavy cost of medicines and the inability to buy or understand that the disease is incurable also created a sense of desperation and helplessness in them. One of the participants who have ichthyosis said:

"There was a period when I had reached the peak of difficult conditions. I was coming from the direction of Piroozi Street to go to my home. Although I was driving I saw a water tanker (truck) watering the last lane of the street garden. I was also behind it. In my mind, I was picturing myself hitting it, but it did not work {I did not commit suicide}, but later it became such a tool for me, that whenever things went wrong for me, I said go kill yourself." (P: 8)

This theme and its sub-themes show that patients with rare diseases endure a lot of mental and physical suffering with a load of fatigue, and the shadow of this disease destroys all aspects of their life.

Like a bird in a cage

This main theme was explained with two sub-themes of deprivation and limitation. Having a rare disease and living it for a human being is like depriving a human being of a normal life. Although rare patients were living,

they felt as if they were a bird left alone in a cage, only alive but limited and deprived of their natural rights.

"Many pleasures are gone, my health is gone, my happiness is gone, I am alone like a bird, of course in a cage!..." (P: 1)

a. Deprivation: Being a rare patient was accompanied by deprivations in many different aspects of communication, emotional and social family, childhood, at school, in terms of recreation or deprived of a suitable job, and even deprived as a human being of a normal life, marriage, and childbearing.

". the blisters increased day by day and my deprivations also increased. It was these deprivations that made me unable to be in society" (P: 10)

The emotional deprivation of patients with a rare disease was such that they were deprived of friendship with other children due to the disease.

"For example, the children of our school were taking pictures together, I could not take pictures with them, I mean no one said to come and join us" (P: 5)

"… sometimes I say that I might not even get married at all. Who lives with us? We are different from the others..." (P: 6)

"The children avoided me, their families were not kind to me, sometimes they looked at me badly, I kept saying to myself, maybe it is just me who thinks so cause, I did not do anything that would make anyone want to insult me, I'm a sick person, I'm in a lot of pain myself." (P: 10)

Living with a rare disease deprived them of social rights such as eating in restaurants, becoming a salesperson, and conducting some other jobs.

"One day we (the children and I) went to the crossroads of our neighborhood. When I wanted to buy a sandwich, we ordered the sandwich and the children were also sitting... Then he put it in a plastic bag and I came to take it away. He said no, you can not eat it here! I said ok, I do not want to and I did not pay, I came out!" (P: 8)

"I would like to work in the community and have my own source of income, but it is not possible. People in the community do not accept us, that is, they do not buy anything from our blistered and peeling hands, they even do not support us much..." (P: 10)

The deprivation stated by one participant who had epidermolysis disease:

"At school, my blisters did not allow me to sit. My hands were stuck together. I wanted to paint. Sometimes I could not even write. I'm very miserable. So far, I have had four surgeries to be able to write with my hands. These hands are stuck together. I cannot describe the pain. I cannot do my job, I cannot laugh." (P: 10)

b. Limitation: Patients faced restrictions due to physical problems, such as short stature that prevented them from choosing their favorite job, or their appearance, which they considered as a limitation for them to be able to marry the person they love.

"In terms of appearance... naturally, if you look at the people, they are all taller... they can do all kinds of jobs... we can only do the jobs of sitting at the desk... like in banks... we cannot do other things." (P: 5)

" I could not go to the swimming pool or the gym like other children, these {lumps} bothered me a lot. I could not wear comfortable clothes. Not being able to go to the gym, or the swimming pool, were all my worries..." (P: 9)

This theme and its sub-themes show that people with a rare disease, like a bird in a cage, struggle with many restrictions and deprivations in society, so they are even deprived of living conditions.

Rejection

Living as a rare patient was perceived as rejection. The participants faced being ignored in different layers of their social life in such a way that to get rid of this high amount of rejection—humiliation, feeling of worthlessness, and low self-esteem, they were pushed into isolation and seclusion and felt themselves shy, disappointed, and heartbroken individuals.

a. Worthlessness: The participants stated that they experienced many humiliations in life, mainly because of their appearance.

"No one saw me anymore, no one even called me, I had no value, I had no price, no one listened to me wherever I spoke." (P: 1)

b. Being stigmatized: The experience of being stigmatized in society and being strange to people's eyes has been annoying for patients. They had experienced the stigma caused by their appearance strongly and at all stages of life.

"I did not have good friends. The families objected to my presence. They said that their children were dreaming at night and I caused mental problems for the children. I did not make excuses for them. I wore gloves, I wore the hijab, and I covered my wounds so that no one would see me, but unfortunately, it did not help." (P: 2)

Another painful experience of being fingered due to disease can be clearly seen in the quote below:

"I wanted to eat kebab (in a restaurant) when a mother and a daughter sat in front of me, the daughter said to her mom, "I'm afraid of this lady!" (P: 7)

Several experiences of the stigma caused by the disease were expressed in the form of being ignored and worthlessness:

"Several times I went to the bakery to buy bread and I was ignored cause I am short, no one even looked at me. How is it possible! I'm a human being..." (P: 6)

Being fingered and ridiculed during school also existed. A participant with achondroplasia said of suffering from being different and being labeled by others:

"At school, I was bothered a lot. For example, in elementary school, I was the children's plaything, the children's mockery. There were many times when I had to sit on a chair alone. (P: 6)

People with rare diseases are unwantedly marginalized in society, they feel heartbroken by the behavior of family, society, and government. In the eyes of many people, they are strange and as if they are not from the planet earth.

Immersion in the whirlpool of thoughts

Being a rare patient has been accompanied by ruminations of hopelessness, not seeing a future for themselves, or a fearful future, so that they feel trapped in a vortex of despairing thoughts. Dark vision and confusion were two sub-themes.

a. Dark vision: The participants stated that although they try to keep their spirits up, it is not possible. Pain and suffering, being ashamed of their presence, fear of themselves, feeling helpless, and fear of a dark future are their constant intellectual concerns.

"It is not at all clear how long we will live. Do we have the right to have a goal? Do we have the right to think about the future?" (P: 10)

Losing opportunities to get married due to appearance conditions and the possibility of having a child with an abnormality (disease) were tied to the feeling of regret, the unknown future, and the fear of seeing an unknown future. The dominant experience of participants was that they did not imagine a future for themselves or only saw a future where they were left alone in a dark cinema.

"I told you that both my body and soul are ruined, I feel that I have no control over my life, I have no thoughts and hope for the future, and I think that my life is doomed." (P: 2)

b. Confusion: The patients were unable to understand society's behavior; society's bad behavior made them confused, and they were bewildered by the pain of the times.

"For example, I went to the pool and explained about my disease, the official said, "

Look, I get it and I can let you go, however, the one who does not know can have any objection. I really cannot answer to all of them. Now, for example, I'm convinced that your disease is not contagious, but I cannot convince all these people." (P: 9)

Their life story shows them being shocked by the bad behavior of society and the strange behavior of the people.

"I went to the laboratory, that gentleman {personnel} said I will not touch it! another one {from the staff} put on gloves so that his hands would not touch me! Now whatever you say to them is nothing!… this puts pressure on a person... one wants to run away... or when the nurse and doctor are reluctant to work for you... the patient takes refuge in them, then they ask you to run away from you! where else can I take refuge!..." (P: 8)

One of the participants said about his painful experiences during his studies:

"I used to get upset... sometimes I would sit down and cry... then I would go and tell the principal. The principal did not do anything either. I experienced most of the damage in elementary school. Mentally I saw a lot of damage; I was surprised by people's behavior." (P: 6)

People with rare diseases do not see a future for themselves due to physical, mental, and social problems, they find themselves immersed in a whirlpool of unpleasant thoughts. They have no hope for the future and are shocked by society's behavior.

Losing the feeling of life

This theme was explained with the sub-theme of continuous regression. Participants experienced the loss of life due to various causes resulting from their rare disease. Feeling lost due to lack of health, which is the basic cornerstone of living. The effects of the disease on the patient's appearance and being involved in a job and as a result not having an income, economic pressure, the heavy cost of medicine, and medical affairs have been the reason for reaching the feeling of losing a life. Not having a job, not having enough money even to buy clothes, the feeling of losing dreams, losing days of life, and overwhelming sadness, has been associated with the experience of being a loser in life.

a. Continuous regret: It was not possible to have a normal life like others or to study, have a job, get married, have children, have fun, travel, make friendships, or have

independence because of having a rare disease, as they experienced longing in their hearts. The shadow of regret over their lives was wide.

"Can my fate be like that of ordinary people? Will I have a wife with children! Not that the disease will take away the courage to think about marriage. I loved children very much. I always wished for a small child, but a child with a rare disease may be born from a genetic disease. So marriage has no meaning... I buried my dreams." (P: 10)

b. Inferiority feeling: The participants felt that people looked down on them. The feeling of inferiority was unpleasant. They perceived themselves differently from others. *"Being a rare patient is like being a black, shiny, and lonely fish in an ocean full of colorful and beautiful fish. Nobody loves you, nobody understands you. Nobody cares about you, they do not respect you." (P: 2)*

Patients with rare diseases regret that the people around them have a normal life but they do not, they see their lives lost, and they feel inferior and ashamed to see their body and appearance.

Discussion

This study depicts the lived experiences of people with a rare disease with the themes of "permanent suffering, like a bird in a cage, rejection, immersion in the whirlpool of thoughts, losing the feeling of life."

In exploring the lived experiences of people as a patient with a rare disease in society, many sufferings and challenges were discovered. Examining the evidence in other countries also shows the same bitterness. The experiences of patients with achondroplasia also showed that their main challenges were difficulties in performing daily activities, access issues, bullying or attracting unwanted attention in public, and negative effects on self-esteem.[17] This finding can indicate that patients with rare diseases all over the world have at least one thing in common, and that is the physical and mental pain they suffer.

The participants with achondroplasia in the present study also had similar experiences and suffered from the stigma of the disease, the feeling of inferiority and worthlessness, inability to perform daily activities. A child with achondroplasia was not able to use the school's toilet and buffet because of his short height and was harassed and bullied by his classmates. The experiences of patients with alkaptonuria also expressed an inability to perform normal routines, and emotional/psychological problems.[18] The quality of life of ichthyosis patients has been affected by physical health, daily life, and relationships with others or themselves.[19] All participants in this study had a low quality of life, especially in psychological aspects, and this finding has also been shown in studies conducted on patients from other countries, which can indicate that despite the availability of facilities in countries, it is rare. The presence of the disease increases the vulnerability of patients and disturbs their quality of life.

The challenges of living with epidermolysis bullosa showed that this disease has a significant burden on patients, caregivers, and their families. Both patients and caregivers must make difficult and compromising choices regarding education, career, and home life.[20] The experiences of young patients with neurofibromatosis have also been low self-confidence, failure in school, and experience of bullying.[21] In the present study, the participants experienced many physical problems in addition to painful psychological experiences. This finding is present in almost all rare diseases. For example, patients with CIPA(Congenital insensitivity to pain with anhidrosis) also had fever, pain, and tongue/hand/foot/corneal ulcers.[22]

Patients with rare diseases need to be better known and understood by both the public and health care providers. Young-onset Parkinson's disease (YOPD) is a rare disease. They also experienced psychosocial problems, anxiety, depression, cognitive disorders, damage to relationships, job damage, and emotional instability.[23] Rare diseases, also called orphan diseases, are diseases that have not been accepted by the pharmaceutical industry because there is little financial incentive for the private sector to develop and market new drugs to treat or prevent them. The findings of the current research and the available evidence also confirm that not only pharmaceutical companies but also a major part of society has rejected them and this may be due to ignorance about rare diseases. Although the researcher hopes that conducting more studies can provide health workers with a better understanding of patients.

The rare disease does not depend on the geographical location and can be in the nature of the disease. The theme of "living with the disease," which is taken from the experiences of patients with rare diseases in Italy tells about the constant suffering of the disease.[24] Despite the low prevalence of rare diseases, people living with this disease all over the world have to cope with the psychological conditions and social consequences.

Examining the needs of patients and relatives with a rare disease in Germany has also proven the challenges of life and daily activities despite the disease,[25] like a caged bird, that is, deprived of freedom and has restrictions

experienced throughout life. Some patients need support from others even for seemingly simple needs such as grocery shopping.

Humans are present in society both in terms of work and in terms of other aspects of life such as entertainment and communication. However, patients with rare diseases experience social exclusion due to the complications caused by the disease, and in some cases, their presence in society reaches zero.

A rare disease affects the patient's quality of life, daily life and ability, and emotional health; thus, most patients experience anxiety and depression.[26] In the present study, the participants experienced many deprivations and restrictions, such as the natural right to marry, the right to become a mother and father, the right to work in society, the right to recreation, prohibition from entering restaurants, swimming pools, non-acceptance by classmates, restrictions on going to gatherings, wedding, and party, and so on.

In the experiences of patients with neurofibromatosis, deprivation and restriction were the main themes, which is in line with the present study.[27] Physical, mental, psychological, social, and economic deprivation and restriction existed in the experiences of all patients, regardless of the type of rare disease. Blisters, wounds, nodules, and short stature caused their dreams and hopes to be dashed.

Growing up with a rare genetic disease is associated with patients' experiences of feeling different and fearing rejection.[28] The experiences of patients with dystonia also showed that they experienced psychosocial fears caused by the stigma of the disease and, in other words, rejection.[29] In the present study, all participants had experiences of disapproval or discrimination in their community and being strange in people's eyes. Living with a rare disease resulted in a deep feeling of losing everything, becoming worthless, inferior, regretting, and feeling like an ugly fish among beautiful fish.

It is suggested that the adaptation process of patients with rare diseases, the burden of rare diseases on the family, and cultural and social challenges faced by rare patients should be researched in the next studies.

Limitations and recommendations

One of the limitations of this study was its implementation during the COVID-19 pandemic, which made it difficult for the researchers to travel to different cities to access patients; therefore, people with rare diseases living in three provinces of Tehran, Mazandaran, and Golestan were used to provide the necessary diversity in terms of demographic and clinical variables of patients.

Conclusion

This research showed that living with a rare disease is an experience of living with constant suffering. It causes a person to find himself captive and confused and immersed in a pool of despair, worthless, deprived, and restricted, under the pressure of being stigmatized by society and with a dark vision for the future. Patients with rare diseases need comprehensive support to take advantage of life opportunities. These findings give health care providers, patients' families, and the public insight into how people living with a rare disease live as human beings with the right to human dignity.

The findings of this research can be used to improve the appropriate medical, psychological, social, and economic conditions and support them. It is suggested that more attention be paid to strategies to reduce the stigmatization of people with rare diseases in society and reduce their social isolation in future research.

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Nil.

Conflicts of interest

There is no conflict of interest.

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