Letter to the Editor

Suffering and Tolerance: A Look at the Lived Experiences of Patients with Neurofibromatosis

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Neurofibromatosis (NF) is a genetic disorder that causes tumors to grow on nerves in the body. It is one of the most common genetic disorders that equally affects men and women of all ethnicities. It is estimated to occur in 1 of every 3,000 births each year. Although NF is considered a common disorder, few have heard about this disease. Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is an autosomal dominant genetic disorder with 100% penetrance. The disease is caused by a mutation in the neurofibrin-producing gene located on chromosome 17 [1]. The disease affects different parts of the body including the bones, nervous system, soft tissues, skin and vessels, which can cause serious problems and complications for the patients. Café au lait spots, neurofibroma and Lisch nodules are present in most of these patients. NF1 is often associated with learning difficulties and may affect physical development. Tumors can grow along the nerves almost anywhere in the body. Some symptoms of NF1 may appear in the first year of life, while others are manifested over a person's lifetime [2]. The symptoms are quite diverse and unpredictable. Few studies have been conducted on the psychosocial effects of this disease on the patients' lives [3]. Living with NF can be challenging since the disfigurement and mental and physical problems affect the quality of life of patients. Studies on the psychosocial problems of this disease have indicated the importance of psychological therapy for the patients based on their living experience. As a person who has been in contact with NF patients for 10 years, the author draws the attention of readers to the review of the lived experiences of these patients in Iran. A qualitative study was conducted on single or married, male and female patients with mild to severe NF from different cities of Iran (from the north, south, east and west of the country). Participants shared their experiences in written form via a website (Living with Neurofibromatosis) [4]. A telegram group, and personal interviews during 2012-2017. Data were analyzed with a qualitative approach, aiming to reveal the themes of living with a rare and difficult-to-treat genetic disorder. Seven main themes were extracted from the thematic analysis: "stigma of disease, pain and suffering, being different from others, hope of recovery, vulnerability, inability to form a family, and

attachment to God and divine providence".

The stigma perceived from living with a disease that changes the appearance of an individual affects various aspects of the patients' lives, causing mental instability, loss of self-esteem, and increased burden of the disease. Family and social acceptance is an important factor to convince patients they can live a normal life. The experiences of patients indicated that the effort put for adaptation, acceptance of the disease and living with this condition are constant concerns. In addition, the low public awareness is due to the rarity of the disease. The uncertain and unpredictable course of the disease and its subsequent complications, make the patient fearful about the disease and his/her future. Hiding the symptoms from others, dependence on family, seeking ways to give identity to the disease by creating virtual social networks for NF patients, and the effort to find a cure for the disease were also evident in the patients' experiences. The results of a study on 15 Norwegian patients aged 18-37 years demonstrated the low self-esteem in NF patients from academic failure and being bullied because of the apparent neurofibromas [5]. Joining supportive virtual communities has been effective in reducing the vulnerability of patients, which gives them a sense of empathy and power to live in the society.

The present study indicates that patients with NF1 live a life dominated by pain and indescribable suffering. Considering the medical, psychological and social problems associated with this disease, the formation of supportive social networks could partly relieve patients from the pain caused by the disease. Nevertheless, there is a need for more comprehensive studies on various psychosocial aspects of the disease in different age groups.

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