Chapter 8
Challenges of Parents With Children With Rare Diseases in Portugal: A Relational Perspective

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ABSTRACT

This chapter makes known the reality of Portuguese families with children diagnosed with a rare disease. The parenting, romantic attachment, coping and emotional regulation strategies, resilience, and personality characteristics of parental figures are the main focus of research carried out in Portugal. In this research participated 160 parental figures (99 mothers and 61 fathers) between 22 and 81 years old. It was found that the existence of a child with a rare disease in the family can give rise to a set of disparate feelings, where the parental figures have to reorganize themselves internally and externally to respond to the needs of the child. It is important to develop intervention programs that address the needs of the parental figures underlying the act of caring.

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INTRODUCTION

Rare diseases reach significant numbers when observed globally (Luz, Silva, & DeMontigny, 2015). In the European Union, a rare disease assumes a proportion of 5 out of 10,000 people (Rare Disease UK, 2016; Silva & Sousa, 2015). But considering that there are over 7,000 different rare disease types, many of which are untitled and difficult to diagnose, these constitute 6 to 10% of diseases worldwide (Luz, et al., 2015; Pelentsov, Fielder, Laws, & Esterman, 2016b). In Portugal, there are about 800,000 patients with rare diseases and hundreds still to be diagnosed (Raríssimas, 2018).

The definition of rare diseases varies globally and is characterized by the low regularity with which they occur in the population, however, there is a wide range of different and increasingly numerous rare diseases (Dellve, Samuelsson, Tallborn, Fasth, & Hallberg, 2006; Schieppati, Henter, Daina, & Aperia, 2008). Rare diseases include, mostly, serious, degenerative or genetic diseases (Pelentsov et al., 2016b). They are often life threatening and can be debilitating as patients’ quality of life is compromised due to the lack of autonomy and the high level of pain and suffering experienced by the rare disease patient and his family (Pelentsov et al., 2016b). Diagnoses and treatments for these diseases are uncertain and / or used too late due to the lack of information and scientific knowledge. The shortage of qualified health professionals and specialized support services also makes it difficult to make progress in the health of individuals with a rare disease (Luz, et al., 2015).

The issue of rare diseases, initially with an interest in medicine, has gained emphasis in addressing the social, economic, psychological and educational implications for patients and their caregivers (Pelentsov et al., 2016b; Waldboth, Patch, Mahrer-Imhof, & Metcalfe, 2016). However, there is still a lack of research in this area, namely studies addressing the relational and affective dynamics of parents, usually the primordial caregivers, in the circumstance of having a child with rare disease. This is why there is a need for reflection and dissemination of this theme (Pelentsov, Fielder, & Esterman, 2016a). This chapter aims to make known the emotional and relational experiences of parents of children with rare diseases, through a pilot research developed in Portugal. First, this chapter focuses on the experiences and difficulties of parental figures who have children with rare diseases. Next, the chapter focuses on the study with Portuguese families, where the main research results are presented. Finally, some intervention proposals with parental figures are suggested, in order to promoting their quality of life.
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