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Opinion Article

The Impact of Rare Diseases in Children and Their Families

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Introduction

A rare disease is defined as a disorder with a prevalence of less than 5 cases per 10,000 inhabitants in the European Union (EU) or less than 200,000 cases in all the United States of America (USA). However, other countries like Japan define rare diseases as those whose prevalence is below four cases per 10,000 inhabitants1. The definition of rare diseases began in the 1980s in the USA, along with the concept of orphan drugs. Altogether, these diseases evolve in a severe chronic form with deficiencies at the sensory and cognitive level. They also present great clinical complexity, making diagnosis even more difficult. The majority of rare diseases occur during childhood due to the high frequency of genetic and congenital nomalies at that age. However, the prevalence of rare diseases is greater in adults than in children due to the high mortality caused by some of those serious diseases during childhood, as well as by other clinical conditions that appear at older ages [1].

There are more than 6,000 distinct rare diseases in the EU, affecting an estimated 30 million people. Around 80% of rare diseases are of genetic origin and, of those, 70% start in childhood. Usually, rare diseases present a variety of symptoms that are different among diseases and among patients with the same disease [2]. Due to the low prevalence of each of these diseases separately, there is still not enough knowledge or scientific evidence on how to detect these cases as early as possible and, above all, how to treat them effectively. Moreover, all these affected persons (patients and families) have many unmet needs [3]. Patients with rare diseases have a series of common needs because they suffer from a very disabling chronic degenerative disease, with a lack of autonomy. Added to this is a high level of pain and suffering for both the patient and the family, as well as an important psychological burden. Therefore, professionals should made efforts to raise interest and awareness for patients and family's psychological wellbeing [4,5].

These diseases also affect work or school and social life. For instance, usually, there is a reduction or cessation of school or professional activity, as well as social relationships [2]. In this scenario, empowering patients and families through education is essential for improving their experience, health status and quality of life [5]. In order to address this type of diseases effectively, it is crucial, first, to find a common definition at the international level. Furthermore, it is necessary to carry out an interdisciplinary approach with the objective of reducing the degree of disability the disease cause, improving the quality of life of the affected people and avoiding premature mortality [3]. In such complex situations, health professionals should give a comprehensive and personalized response to people affected by these diseases and their families. However, professionals cannot give this response in a single health center. The collaboration of the different agents of society (health and social areas) is crucial in order to meet the needs of patients and families affected by rare diseases. Given this scenario, the UNICAS (UNIQUE) project, promoted by the Sant Joan de Déu Children's Hospital, in Barcelona (Spain) aims to build a network of hospitals and health centers in Spain and Europe as well as synergies with companies and research centers. Its main objective, thus, is to create an ecosystem of alliances to improve care for children and families with complex rare diseases [6].

Conclusion

it is important to combine all of society's efforts to improve the lives of those affected by rare diseases. Health professionals, managers, politicians and also patients and family members together with patient associations; all efforts are necessary and must be committed to the identification, treatment, and cure of rare diseases through education, advocacy, research, and service programs.

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