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Rare Diseases: A Current View

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Introduction

"Thinking about the common, but remembering the strange", expression that was sometimes recalled during our undergraduate studies, because it is known that only what is thought. It is usually the common diseases that are first thought when doing the diagnosis, but those in which the prevalence is scarce, rare diseases are often not taken into account, for this reason the patients who suffer them, not only they experience the inherent conditions of their pathology, but also the lack of knowledge and specialization, which, added to the lack of specific health policies, generate delay in diagnosis and difficulty access to health services, thus accumulating physical, psychological and Intellectuals, and face inadequate or even harmful treatment [1]. Rare diseases are conditions of disease characterized by low prevalence, but also little known, little studied and most no specific treatment and diagnosis. They are also known as orphan diseases because they are unattractive for research and clinical studies [2-4].

There is no single definition for rare diseases, the most used in the literature derive from its prevalence, for example; in the United States refers to any condition that affects fewer than 200,000 people, while in the European Union defines it as that, genetic or acquired, life -threatening or chronically debilitating and whose prevalence in the general population is less To 1 in 2,000 individuals [1]. In Colombia an orphan disease is one chronically debilitating, life - threatening severe and less prevalence of 1 per 5,000 people. About 80% of rare diseases are of genetic origin, a very minor number of them, no more than 400, are treated [2]. Two out of three diseases appear before two years, representing 20% of infant mortality and 10% of pediatric hospitalizations [5].

Rare diseases have particular characteristics beyond the small number of patients affected. They often affect patients from birth, affect multiple organ systems, are severely disabling, can greatly reduce life expectancy and impair physical and mental abilities [6] Not content with this, the staff interact with the health service is an odyssey that begins with the formulation of diagnosis. Because of the low prevalence of these diseases, primary care physicians and even specialists are not properly trained to diagnose the rare disease. Some of

these disorders have a defined diagnostic test and even a treatment that has proved useful, but the difficulty in establishing the correct and timely diagnosis is a relevant issue, resulting in loss of treatment opportunities and associated with increased morbidity or the mortality [7].

As illustrated in several studies there is a delay for the diagnosis of different rare diseases. In order to reach the diagnosis in patients with achalasia, children were misdiagnosed and maltreated for more common medical conditions and in some patients the duration of symptoms before the diagnosis of achalasia was 6 to 10 years, although for their diagnosis Performed through a good symptomatic evaluation, confirmed with esophageal manometry and medical and surgical treatment has proved to be effective and safe, due to the fact of having symptoms that simulate common diseases (vomiting, dysphagia, weight loss), a misdiagnosis [8]. Like patients with telangiectasia hereditary hemorrhage, they attended the pediatric age with spontaneous epistaxis, however it went unnoticed until other features of the disease manifested clinically in adulthood, including vascular complications. The diagnostic delay time was approximately 2 decades from the onset of symptoms, prolonging the appropriate treatment, since this disease has a safe and effective preventive treatment for arteriovenous malformations, which are potentially dangerous to life and have sequelae Disabling [9].

In patients with primary ciliary dyskinesia (PCD) the median age at diagnosis was 4.4 years in the majority of patients despite the presence of typical symptoms from an early age and the prolongation of the time of diagnosis led to severe Chronic and irreversible lung damage [10]. For individuals with Pompe disease, despite the existence of options accurate diagnosis and therapy of enzyme replacement, there is a delay in diagnosis, which is higher in patients with late - onset Pompe disease. During this period without adequate diagnosis accumulation of debilitating problems and increased pathophysiological damage, causing significant morbidity and increased mortality, in addition the early initiation of therapy presents better results and can stabilize the progression in many patients. [11] Some relatively common disorder can hide rare diseases, such as autism (in Rett syndrome, Usher

syndrome type II, Cerebral gigantism Sotos, Fragility Chromosome X, Disease Angelman, phenylketonuria Adult, Sanfilippo, Etc.) or epilepsy (Shokeir Pena Syndrome, or Fetal Hypokinesia, Feigenbaum Bergeron Richardson, Kohlschutter Tonz, Dravet, etc.) [12].

Conclusion

The main reason for delaying the diagnosis of rare diseases is the insufficient knowledge of these diseases due to their low prevalence, also because they are heterogeneous disorders that some lack a characteristic seal or have clinical characteristics that overlap with several diseases Common and most do not have an effective biochemical or histological analysis for their diagnosis. For this reason, it is necessary to emphasize the critical need to increase the awareness of rare diseases by medical personnel, because despite the different advances in the diagnosis and treatment of some of them, even the correct and definitive diagnosis depends on The cognitive biases of the physician, and it is not acceptable that "those living with a rare disease in addition to suffering the pain of the disease, suffer the loneliness and vulnerability of disinformation" [13].

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