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ABSTRACT

Next February 28 will be celebrated once again the international day of rare diseases, it is my intention to expose the reasons why these diseases are forgotten, silenced and underdiagnosed in developing countries, with the consequent delay in diagnosis, cure or relief and co or consequence late incorporation into society to the extent of its possibilities and as unfortunately often occurs discrimination and stigmatization of patients.

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According to the World Health Organization (WHO), rare diseases (RD) are those that occur in fewer than five people per ten thousand inhabitants and there are more than seven thousand diseases. Rare Disease's Day will be commemorated on the last day of February. The main goal is to recognize the existence of these "silent conditions" to mention the reason why are for and involve doctors and civil society. [1]

Although there is no single definition for the term "Rare Disease", all definitions are based on the frequency with which they occur and also include elements such as the severity of the manifestations and/or the availability of treatment. The definitions used in the medical literature and the different national health programs fluctuate between prevalence's of 1:1,000 to 1:200,000 inhabitants. In the United States, the Rare Diseases Act of 2002 defines a rare disease strictly according to its prevalence and specifically says that it refers to any disease or condition that affects fewer than 200,000 people in the United States, which is equivalent to about of 1:500 people [3-5], in Japan, the legal definition states that a rare disease is one that affects fewer than 50,000 patients, which is equivalent to 1:2, 500 people. [2] The European Union defines rare diseases (RDs) as life-threatening or chronically debilitating conditions whose prevalence is less than 5 per 10,000.

Moreover, for many RDs, including those of genetic origin, combined efforts are required to reduce morbidity or perinatal and early mortality, and address the considerable decline in an individual's quality of life and socioeconomic potential. [6-10] Excluded from this definition are diseases that are statistically rare, but that are not life-threatening or chronically debilitating. Nevertheless, definition of RD demands the existence of a reliable statistical system that reflects the true impact of the disease on society. But at least in this West African country (Gambia) the study of rare diseases is a challenge for health services, and I will present my personal considerations.

Some of the countries of the region lacks reliable statistics and those that do exist do not collect all the information available in the different, dispersed, and little cohesive health structures of the public system and in addition the poor interconnection with private health entities just aggravates the problem. [11-15] On the other hand, the information collected often does not match with international nomenclature system and the deficit in technology and communications dragged on for centuries takes its toll on the credibility of the information collected. Without underestimating the human factor that lack of knowledge, experience, and lack of communication between municipal, regional, and national entities only worsen the current situation.

On the other hand, since the 1980s, rare diseases have been acquiring a priority place in health programs and in public opinion. In general, they are chronic, invalidating diseases and in more than 80% of genetic origin. Given the low specific prevalence of each condition, there is extraordinarily little knowledge on the part of the medical community in relation to its diagnosis and management and the impact at social, familial and society honestly is underestimated. [2,4,5].

Although some rare diseases may be detected quickly, in other cases years may pass between the first appearance of symptoms and a correct diagnosis of a rare disease, and misdiagnoses—and treatments based on them—occur with documented frequency. According to the literature GAO reviewed and GAO's interviews, those with undiagnosed, misdiagnosed, or untreated rare diseases may face various negative outcomes. For example, a person's health can suffer when appropriate, timely interventions are not provided or when interventions based on misdiagnoses cause harm. In addition, multiple diagnostic tests, medical appointments, and unwarranted interventions can add to the costs of the disease. [2,5,7,12].

People with rare diseases in underdeveloped countries suffer from stigmatization, depression and even persecution due to multiple social, religious, and other factors, for which their most basic rights continuously violated and discriminated against, leading to depression, humiliation, and many occasions suicide. There are no national programs or projects for the diagnosis, treatment, and protection of these patients.

In the scientific medical field, the lack of reference centers and qualified personnel, who are at the same time familiar with this type of ailment, is a handicap for the new generations of physicians who are not accustomed or prepared, familiar or prepared to face these diagnostic entities which In turn, they require very expensive studies, many of which are exclusive to developed countries, unjustifiably lengthening the time for diagnosis, worsening the prognosis and limiting the quality of life and social integration of these patients. Peer-reviewed studies of specific rare diseases estimated costs for people with rare diseases that are multiple times higher than costs for people without those diseases. One recent study, which has not yet been peer-reviewed, estimated $966 billion as the total cost (including medical and other nonmedical and indirect costs) in the United States for an estimated 15.5 million people with 379 rare diseases in 2019. [2,4,7,16-19].

In short, rare diseases in developing countries may not be so "rare", the rare thing is that there is the human, technical, social, and political capacity to address them in a comprehensive, holistic, and humanistic way. It is that this February 28th, when the WHO commemorates the day of rare diseases, not only take into account these patients who are already diagnosed, integrated or under treatment, but also help include all these people who suffer and will suffer rare diseases in their respective countries euphemistically called "developing" countries and for which there are no opportunities for diagnosis, treatment and social integration, due to the intrinsic economic, technical, human and scientific problems that chronically suffer.

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