

Eidikology: proposition for a new terminology for the science of rare diseases

The European Union defines a disease as rare when it affects fewer than 5 per 10 000 persons,¹ whereas the USA considers a condition rare if it affects fewer than 200 000 individuals.² To date, however, there is no terminology to appropriately address these diseases as a category other than *rare diseases*, or in some instances *orphan diseases* when referring to conditions that lack the resources for treatment discovery. Even though these terms have helped thus far to describe the existence of a distinct group of patients, they fail in two ways:

First, they burden the patient with the stigma of living under the shadow of the *rare*. This makes social integration for these patients even harder. In our experience at the Catholic University of Honduras Research Group (GIMUNICAH), we have encountered several stories of patients who, even before diagnosis, were victims of bullying for years. Once the diagnosis is reached, the sole name of the syndrome or disease brings some peace to the patient and family; however, the categorisation as a rare disease brings back a socially marginalising label that is unfortunately recognised by medical terminology.

Second, they fail to recognise this field as a science of its own. With approximately 8000 reported conditions¹ and the particular diagnostic demands a physician faces, this set of diseases requires its own field of specialisation. Medical faculties around the world do not include them in their educational programme or dedicate only brief mentions to a few conditions. Therefore, patients are usually erroneously referred or specialists in other fields have enormous challenges in diagnosing these diseases due to the lack of familiarity with the clinical patterns. This results in

patients visiting a mean of 7.3 physicians in 4.8 years before definite diagnosis.³ Patients also require not minutes, but hours of clinical analysis to adequately approach them, which most health systems do not offer due to time constraints. All this highlights the need for training a new set of physicians to assist the more than 350 million individuals thought to be living with rare diseases worldwide.⁴

Millions of human beings around the world suffer in silence, almost forgotten by medicine with no other category to belong but to the *rare*, the *orphan* and *abandoned*. It is my dream that someday medical doctors around the world can choose to specialise in this field, in this medical science. It is my hope that the day comes when patients do not feel excluded by the very category of their disease; that they know someone dedicated and prepared themselves for the chance to help them. For them to know they matter and are *special*. Maybe it is just a small symbol, a humble step in the direction of that vision, but we all need a name when we start the journey of life to become who we are. Therefore, I propose the term *Eidikology*, from the Greek ειδικός (Eidikós), meaning *special*, and λογία (-logia), meaning *study*, as the name for the branch of medicine dealing with diseases of very low prevalence; a new term for the science of rare diseases.

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