Measuring the impact of diagnosis and treatment of rare diseases

Statement of the Problem: Knowledge on the impact of Rare Diseases (RD) on people living with a RD and their families lives is essential to draft the value of diagnosis, treatment, and healthcare. There has been a significant acceleration in diagnosis and treatment development, but their impact on people lives can't be assumed. The 2nd study “Spanish People Leaving with a Rare Diseases (PLWRD) social needs” compares and analyzes the diagnosis delay and its consequences as well as treatment difficulties between 2009-2017. Outcomes from other RD impact surveys are also analyzed and compared.

Methodology: 1,576 questionnaires have been collected for PLWRD and their families, more than double collected in the first study in 2009. Furthermore, 8 discussion groups and 14 interviews have been conducted.

Main Findings: As in 2009, half of the people surveyed have been delayed in their diagnosis; almost 1 of every 5 people with RD (19%) has taken more than 10 years to get it and a similar percentage (18%) between 4 and 9 years. More than a third of the people studied say they have difficulties accessing the medical devices or medicines they need, mainly due to their high price, the absence or withdrawal of the product or bureaucratic problems. This situation has worsened compared to 2009, when was a quarter of the population studied who have difficulties with this access.

Conclusion & Significance: Obtaining a diagnosis is a determining factor for proper medical and health care, which can include symptom-driven treatments, prevention strategies, and avoidance of unnecessary interventions. Although the new diagnostic technologies for better management of diseases are promising, their impact on clinical practice is still limited. Understanding the genetic basis of diseases, coupled with advances in biotechnology and drug development, has led to progress, nevertheless, patients can benefit only to the degree that they have access to tests and interventions.

Biography

Alba Ancochea is the CEO of Spanish Federation of Rare Diseases and its Foundation. She is a EURORDIS board of Directors member, as well as member of the Rare Diseases International Advocacy Working Group, member of the DITA (Drug Information ,Transparency and Access) Task Force, representative of the Council of National Alliances and supporting the Ibero-American Alliance of RD. She belongs to the CIBERER (Spanish Biomedical Research Consortium on RD) Scientific Advisory Board. She has completed undergraduate and masters studies in psychology, special needs teaching and management of NGOs. Alba is a graduate of the EURORDIS Summer School and EUPATI expert training course among other qualifications. She represents PLWRD in a dozen of Work Committees and national and international projects regarding National Strategy, Centers of Expertise, access to Orphan Medicinal Products, treatment and research on RD. She is also teacher in Socio-Health Masters and other courses related with RD.

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