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EDITORIAL

Key priorities in rare neurological diseases: A statement from the Coordinating Panel on Rare Neurological Diseases of the European Academy of Neurology

On 28 February, we observed Worldwide Rare Diseases Day, a full day dedicated to raising awareness and generating concrete changes for the 300 million people worldwide living with a rare disease, their families, and carers.

A rare disease (RD) is defined as one that affects fewer than five per 10,000 persons in the European Union or fewer than 200,000 persons in the United States. Rare neurological diseases (RNDs) constitute a significant proportion of RDs. Almost 80% of RDs are caused by genetic anomalies, and more than half of the cases affect the central and/or peripheral nervous system, either isolated or in combination with other systems, whereas 70% start in childhood. Importantly, an increasing number of rare disorders have treatment implications and therefore should not be missed.

The European Academy of Neurology (EAN) promotes knowledge and awareness of RNDs. On average, it takes 7 years to establish the correct diagnosis, which is a great cause of uncertainty for the patients and the parents. Even more importantly, no diagnosis or a misdiagnosis may be associated with inappropriate treatment. A recent survey performed by the EAN Neurogenetics Panel [1] revealed negative scenarios. Although most of the survey responders were aware of RNDs, when it came to amenability to carrying out a complete genetic diagnosis, almost one third of the responders declared they were not happy with the current way of ordering genetic analyses in their countries. Furthermore, whole exome and genome sequencing are not easily accessible throughout Europe, with considerable variabilities among countries. Almost 10% of the responders did not know whether presymptomatic and prenatal diagnosis was available in their countries, and 47.3% were not aware of which newborn screening programs were available. Finally, 96.3% of responders declared that there is a need for education and training in neurogenetics.

Therefore, it is now time to accelerate on the field; we suggest the following key priorities to be achieved in the next 3 years:

Key Priority 1: Ensure European patients get the right diagnosis faster wherever they live, including prenatal diagnosis and newborn screening for the treatable RNDs. Fast and correct diagnosis is best provided at RND expertise centers, as is the required interdisciplinary and interprofessional care provision.

Key Priority 2: Keep increasing awareness and knowledge of rare conditions among health care professionals. This should also be achieved by systematically including RNDs in the curriculum during medical school, integrating them into the neurological and child neurology training requirements, the development of a postgraduate curriculum on rare neurological disorders, and continuous medical education programs. Furthermore, knowledge and standards of care developed in the European Reference Networks should be used throughout Europe.

Key Priority 3: Speed up the diagnosis of cases not diagnosed within their own countries, which could be done by taking advantage of European Reference Network activities (https://health.ec.europa.eu/european-reference-networks/work-erns_en), including the Clinical Patient Managements System.

Key Priority 4: Promote comprehensive and systematic collection of clinical and research data on RNDs, including use of Orphanet rare disease nomenclature (ORPHA codes) for coding purposes in the hospital information systems to render RDs visible.

Key Priority 5: Establish a more structured dialogue between all stakeholders, including patient organizations, and especially between the European Commission, national health agencies, European Reference Networks, and the EAN. Specific funding to achieve these objectives should be considered, with an ad hoc committee delineating time tables, deliverables, and milestones.

Diagnosis, care, and treatment of patients and families with RNDs are major health care challenges that have not yet been solved. Multistakeholder cooperation, significant resources, and strategic leadership are required to bring about the needed changes in health care and research. The EAN, in tight collaboration with all stakeholders including the European Reference Networks for Rare and Complex Diseases, is fully committed to this cause.

KEYWORDS

awareness, neurogenetics, rare diseases, rare neurological diseases

CONFLICT OF INTEREST STATEMENT

None of the authors has any conflict of interest to disclose.

DATA AVAILABILITY STATEMENT

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

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