Treatabolome project designed to shorten diagnosis-to-treatment time for patients with rare diseases

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The Treatabolome project is a research initiative to develop a freely available, interoperable online platform dedicated to disseminating rare disease and gene-specific treatment information to healthcare professionals regardless of their level of specialized expertise. Developed under the Solve-RD European Research Project, it is intended to reduce treatment delays for patients with rare diseases by directly linking diagnosis and treatment information. This initiative is highly relevant to neuromuscular disorders as they are rare diseases by definition. In this special issue of the *Journal of Neuromuscular Diseases*, experts contribute Treatabolome-feeding systematic literature reviews on rare neurological and neuromuscular disorders.

In Europe, rare diseases are defined as those that affect one in two thousand individuals or fewer. Although they are individually rare, globally they affect around 6% of the population. Collectively, healthcare providers deal with a considerable number of patients with a rare disease, over 70% of which are genetically determined. Historically, around 25% of patients with rare diseases have endured a diagnostic odyssey that can last up to 30 years, according to Eurordis, an alliance of over 900 patient organizations that work together to improve the lives of people living with a rare disease.

Next generation sequencing has made it possible to efficiently diagnose a growing number of rare diseases. At the same time, new and frequently specific treatments are emerging following decades of expectant wait by patients, their families, and caretakers. To deliver such treatments to patients in an expedient manner, diagnosis and treatment now need to be linked consistently.

"The Treatabolome project arises from the improved availability of genetic diagnosis and the rapidly growing number of rare disease treatments," explained Guest Editor Gisèle Bonne, Ph.D., Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France. "There are treatments available for an increasing number of rare diseases, but there is often a substantial delay before patients receive the right treatment. Although targeted treatments are currently only available to a minority of patients with rare diseases, recent developments point towards a steep increase in the coming years, as suggested by the development of multiple gene therapies and the steady increase in the number of orphan drug applications."

This EU project involves four European Reference Networks or ERNs: ERN-RND for Rare Neurological Diseases; ERN Euro-NMD for Rare Neuromuscular Diseases; ERN-ITHACA for rare congenital malformations and syndromes with intellectual and other neurodevelopmental

disorders; and ERN-GENTURIS for patients with one of the rare genetic tumor risk syndromes.

The special issue covers gene and variant-specific treatments for rare neurological and neuromuscular disorders, highlighting the important premise that a precise genetic diagnosis may result in an equally precise therapeutic approach. It includes systematic reviews from internationally leading centers representing a concentration of rare diseases expertise involved in producing Treatabolome-feeding literature reviews, which cover:

- Genetic forms of Parkinson's disease
- Skeletal muscle ion channelopathies
- Hereditary peripheral neuropathies
- Metabolic myopathies related to glycogen storage and lipid metabolism
- Laminopathies

The issue also includes a review of the underlying disease mechanisms, potential therapeutic approaches, and the state of trial readiness of future clinical trials in facioscapulohumeral muscular dystrophy (FSHD).

The intention is that as the Treatabolome platform is finalized, the main body of data will be derived from expert-led systematic literature reviews such as those published in the current issue. They bring state-of-the-art, evidence-based information in a standardized format that is FAIR-compatible and easily uploadable to the Treatabolome database.

"When combined with diagnosis support tools like RD-Connect's Genome-Phenome Analysis Platform (GPAP), the Treatabolome prevents treatment-onset delays," noted Dr. Bonne. "It puts flesh of content onto the bones of the platform."

One of the next steps of the Treatabolome project will be to extend it to rare diseases experts from all 24 European Reference Networks and the whole scientific community, producing other datasets to enrich the database and facilitate treatment awareness for rare diseases. The project is also exploring artificial intelligence (AI) pathways for the future updates of the database that involve machine-led text mining of publications to improve the life cycle of the information displayed in the database.

"The main challenges are involving all significant rare disease groups to produce the Treatabolome datasets of their expertise, guaranteeing a yearly life cycle for updating that information, and finding continued funds to ensure the sustainability of the Treatabolome platform," commented Dr. Bonne. "Additionally, research on automated information update relying on AI is fundamental for long-term platform sustainability."

"We must strive to work towards a world with progressively fewer 'undiagnosed' and 'not yet diagnosed' rare disease and ultra-rare disease patients. Most importantly, once a diagnosis is reached, if a treatment exists for the disease/gene/variant, one should not have to wait for it to be offered to the patient," she concluded.

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