Rare Disease Day: a new EU platform to support better diagnosis and treatment

Currently a vast amount of data on patients with specific conditions is scattered across Europe in about 600 'registries' – databases that hold information on patients with specific conditions. Data is not collected EU-wide and there are no shared standards to analyse the information that is available on rare diseases. The new European Platform on Rare Diseases Registration will bring this data together supporting the quality research that can enhance diagnosis and treatment outcomes – helping to improve the lives of patients and their families.

Vytenis **Andriukaitis,** Commissioner for Health and Food Safety, said: *"Rare diseases present a health challenge where action and collaboration at EU level has clear added value. Connected, we are so much greater than the sum of our parts. This new EU Platform on Rare Diseases Registration will address the fragmentation of rare diseases data, promote the interoperability of existing registries and will help to create new ones Moreover, the Platform will also be useful for the work of* European Reference Networks*, real EU success story, allowing them make use of anonymised information from a large pool of patients and offer better treatment to those in need."*

Tibor **Navracsics**, Commissioner for Education, Culture, Youth and Sport, responsible for the Joint Research Centre, said: *"This platform will help scientists, policymakers and patients alike make the most of data on rare diseases that have, until now, remained largely untapped. By setting EU-wide standards for data collection and exchange, the platform will also mean that information collected in the future can more easily be compared across Europe. And patients can rest assured that their private data will remain private – while benefiting from improved diagnosis and treatment."*

A rare disease can affect someone from birth, like cystic fibrosis, or it can develop later in life, like Huntington's disease. Despite the large total numbers of those affected in Europe, information on effective diagnosis and treatment strategies are not collected in a uniform way and are often not shared among registries or across countries. As a consequence, patients often suffer alone with little or no hope of being cured.

There are in fact very few national rare disease registries in the EU – most are managed by individual hospitals, research institutions, pharmaceutical companies or patient advocacy groups. The type of data collected varies widely. Some are focused on developing medicines for particular diseases, while others may be more interested in tracking instances of rare diseases over time, for example.

The new platform will substantively improve this situation. Available online and open to the public, it merges registry data sources to foster the critical mass of patient data needed to trigger pharmacological, translational or research studies.

The Platform includes a registry infrastructure consisting of:

- the European Directory of Registries, which gives an overview of each participating registry;
- the Central Metadata Repository, which stores all types of variables used by the registries;
- a data protection tool, which makes sure patient data is held under a pseudonym and cannot be traced back to the individual.

By providing EU standards for data collection and data sharing, the platform will for the first time make it possible to search data of rare disease patients. This significant achievement will allow the creation of critical knowledge for a given disease, enabling research and supporting patients, health care providers and policy-makers.

The platform will be an important asset for the European Joint Programme on Rare Diseases which aims to establish a research and innovation pipeline for rapid translation of research results into clinical applications and uptake in healthcare. Through this programme, the platform resources can be used in future research projects and disseminated to a wider community of rare disease researchers, clinicians and patients in the EU and beyond.

Background

The European Platform on Rare Diseases Registration supports the objectives laid out in the Commission Communication on Rare Diseases: Europe's challenges and the Council Recommendation on an action in the field of rare diseases. These documents have guided the European response to rare diseases over the past decade, identifying codification and inventory of rare diseases as elements of such a response.

It also supports the implementation of the Directive on the application of patients' rights in cross-border healthcare, notably the rare diseases-related work of the European Reference Networks. These connect medical specialists and researchers and provide advice to patients based on the principle that the knowledge travels, not the patients.

Rare diseases have been a priority for EU Framework Programmes for Research and Innovation for over two decades. By enhancing the interoperability and re-usability of registry data, the platform also supports EU funded research and innovation to respond to rare diseases-related challenges.

As the European Commission's science and knowledge service, the Joint Research Centre supports EU policies with independent scientific evidence throughout the whole policy cycle.

Further information

European Platform on Rare Diseases Registration

Factsheet on the platform

Commission information on World Rare Diseases Day

Commission activities in the area of rare diseases

European Reference Networks