



Rare, but frequent diseases: increasing the efforts undertaken in France and in Europe!

Press Release of the National Academy of Medicine

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On February 28, 2021, the 14th International Day for Rare Diseases, coordinated by EURORDIS, the European Federation of Patient Organizations, will take place providing an opportunity to make aware the general public and healthcare workers.

Rare diseases are defined in Europe by a prevalence of less than 1/2000 people. In France, around 7,000 rare diseases are identified, affecting more than 3 million people. Some are relatively frequent, such as cystic fibrosis, others affect a very small number of patients, or even a single case. Eighty percent are of genetic origin and concern, in half of the cases, children under 5 years of age.

France has been a pioneer in structuring the care of rare diseases through the deployment, since 2005, of National Plans for Rare Diseases (NPRDs). One hundred and thirty-one reference centers (CRMR), with proven expertise in one or more rare diseases, bring together a network of 500 regional centers of competence that collaborate in care, teaching and research activities.

These centers are integrated into national rare disease health networks (FSMR). Cross-border cooperation takes place within the European Reference Networks (ERNs). The 24 ERNs are virtual networks using a computer platform and telemedicine tools to examine patients' cases. France is a major player in these ERNs as shown by its participation to each ERN.

Research in the field of rare genetic diseases remains a huge task. It also concerns rare cancers, which mainly affect children, and the development of orphan drugs, which has made considerable progress in recent years. For example, cholic acid (under the name Orphacol®) is used to treat two extremely rare liver diseases due

to an enzyme deficiency that prevents the synthesis of the bile cholic acid. Prior to this drug, the disease progressed to cirrhosis and the need for a liver transplant.

Orphanet is an information server, with free access for the general public, dedicated to rare diseases and orphan drugs.

Wandering time for diagnosis is estimated at 2 years on average in France, but this average hides wide disparities; almost a quarter of patients still wait more than five years to find out what their illness is.

Patients' associations have played a major role in the development of the diagnosis and management of rare diseases, by making them known to the general public and by forming alliances with healthcare professionals. Many of them are grouped within the “Alliance Maladies Rares”, the “Alliance Française des Myopathies (AFM)-Téléthon”, and “Vaincre la mucoviscidose (VLM)”. “Maladies Rares Info Services” provides patients and their families with simple and accessible information, with the possibility of direct telephone dialogue.

Strong points:

1. Minimise diagnostic wandering and deadlock faced by patients.
2. Structure the transition from child to adult as optimally as possible for patients with a rare disease that appeared in childhood.
3. Allow better identification of the reference and competence centers for rare diseases in each university hospital center.
4. Evaluate the quality of ENRs.
5. Periodically renew the posting of posters in public places in order to raise public awareness of these diseases ("I'm missing a gene, does that bother you?").

Report of the National Academy of Medicine: Rare Diseases, the French model. April 2016. National Plan for Rare Diseases 2018-2022.

Activity report 2018, health sectors rare diseases. Sharing innovation, a diagnosis and treatment for everyone.

° ° ° Press release approved by the Board of Directors on 27 February 202