

# RARE DISEASES: BACKGROUND INFORMATION

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## The context

As many as 8,000 rare diseases affect the lives of around 30 million people in the EU every day. Many of those affected by a rare or complex disease do not have access to diagnosis and high-quality treatment, since no country alone has the knowledge and capacity to treat all rare diseases and conditions. **European Reference Networks (ERNs)** are virtual knowledge-sharing networks involving healthcare providers across Europe. These networks give patients and doctors across the EU access to the best expertise, while facilitating the timely exchange of life-saving knowledge, often without the need to travel to another country. The EpiCARE network brings together 28 highly specialised health centers in 13 European countries, all of whom have expertise in rare and complex forms of epilepsy.

## The story

Last year, EpiCARE took on the case of a 4-year-old Finnish boy with a rare abnormality in the brain, causing him to have between 20-30 seizures each day. This was a very specific disorder of which only a small number of medical centres in the world have experience. A decision therefore had to be made on what kind of treatment to suggest – Gamma Knife® surgery, only available in 2 centres, or surgical resection, only performed in 3-4 centres. The doctors in Finland presented the case to the network, with 7 other countries present (Italy, UK, France, Spain, Romania, Sweden, and the Netherlands). After reviewing the different treatment options, it was decided that the child should go to Marseille for Gamma Knife surgery®. The surgery was successful, and although the boy still experiences seizures, their number has been drastically reduced.

## The objective

The story aims to introduce readers to the principle of European Reference Networks and the value of the knowledge sharing they enable. Through the story of this particular young patient, audiences will understand that cooperation at an EU level makes a real difference to both patients and their families, as well as to the health professionals that care for them. Due to the rarity of such complex conditions specialist knowledge is scarce, fragmented and often not available in the patient's region or even country. Many patients therefore do not get satisfactory explanations for their symptoms and have difficulties finding a specialist with the necessary knowledge about treatment options. By consolidating knowledge and expertise scattered across countries, the ERNs give healthcare providers access to a much larger pool of expertise. This story demonstrates that the ERNs improve the chances of patients receiving accurate diagnoses and advice on the best treatment for their condition.

## The European reference networks

European Reference Networks (ERNs) are unique and innovative cross-border cooperation platforms that enable specialists to diagnose and treat rare or low prevalence diseases. They aim to provide highly specialised treatment and to concentrate knowledge and resources. ERNs are being set up under the EU Directive on Patients' Rights in Healthcare ([2011/24/EU](#)), which also makes it easier for patients to access information on healthcare and increase their treatment options. There are 24 ERNs involving 25 European countries (including Norway) and more than 300 hospitals with over 900 healthcare units covering all major disease groups. These 24 specialised networks [are working on a range of medical issues](#) including bone disorders, childhood cancer and immunodeficiency. The ERN initiative receives support from several EU funding programs, including the [Health Programme](#), the [Connecting Europe Facility](#) and [Horizon 2020](#).