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What is a rare disease?

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A disease or disorder is defined as rare in Europe when it affects less than 1 in 2000 citizens (Orphan Drug Regulation 141/2000). Rare diseases may affect 30 million European Union citizens.



Characteristics of rare diseases

- ▶ Rare diseases are often chronic, progressive, degenerative, and often life-threatening
- ▶ Rare diseases are disabling: the quality of life of patients is often compromised by the lack or loss of autonomy
- ▶ High level of pain and suffering for the patient and his/ her family
- ▶ No existing effective cure
- ▶ There are between 6000 and 8000 rare diseases
- ▶ 75% of rare disease affect children
- ▶ 30% of rare disease patients die before the age of 5
- ▶ 80% of rare diseases have identified genetic origins. Other rare diseases are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative.

Rare diseases are characterised by a broad diversity of disorders and symptoms that vary not only from disease to disease but also from patient to patient suffering from the same disease.

Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis.

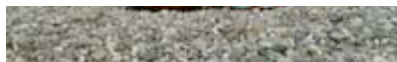
Rare disease patients face common problems:

- ▶ Lack of access to correct diagnosis
- ▶ Delay in diagnosis
- ▶ Lack of quality information on the



disease

- ▶ Lack of scientific knowledge of the disease
- ▶ Heavy social consequences for patients
- ▶ Lack of appropriate quality healthcare
- ▶ Inequities and difficulties in access to treatment and care



How can things change?

- ▶ By implementing a comprehensive approach to rare diseases
- ▶ By developing appropriate public health policies
- ▶ By increasing international cooperation in scientific research
- ▶ By gaining and sharing scientific knowledge about all rare diseases, not only the most “frequent” ones
- ▶ By developing new diagnostic and therapeutic procedures
- ▶ By raising public awareness
- ▶ By facilitating the network of patient groups to share their experience and best practices
- ▶ By supporting the most isolated patients and their parents to create new patient communities or patient groups
- ▶ By providing comprehensive quality information to the rare disease community

European Regulations and policies in place in favour of rare disease patients:

- ▶ EU Regulation on orphan Products (1999)
- ▶ EU Regulation on Paediatric Drugs (2006)
- ▶ Programme of Community Action in the Field of Public Health (2007-2013)
- ▶ EU 7th Framework Programme for Research (2007-2013)

Eurordis has published a founding paper on rare diseases: ‘[Rare diseases: understanding this public health priority](#)’

Eurordis publishes the latest figures on rare diseases in a report called The Prevalence of Rare Diseases. This report is published jointly with [Orphanet](#) on a quarterly basis.

Author: Eurordis
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