

## ABOUT RDI

We are a national alliance of patient advocates representing = voluntary groups supporting people affected by or at risk of developing a rare disease.

RDI is committed to the identification, treatment, and cure of rare disease through programs of education, patient services and advocacy.

Welcoming input from all stakeholders of the rare disease community in Ireland.

We at Rare Diseases Ireland feel strongly that it will only be through the collaboration of all stakeholders that we will advance our core objective, to improve the lives of those affected by rare diseases and their families.



Rare Diseases  
Ireland

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

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



# WHAT IS A RARE DISEASE?

A disease or disorder is defined as rare in Europe when it affects less than 1 in 2,000 citizens (Orphan Drug Regulations 141/2000). Rare diseases may affect 30 million European Union citizens.

## CHARACTERISTICS OF RARE DISEASES

- Rare diseases can be chronic, progressive, degenerative, and often life-threatening
- Rare diseases disable patients as their quality of life can be compromised by the lack or loss of autonomy
- High levels of pain and suffering not only for the patient, but for their family members
- No existing effective cure
- There are between 6,000 and 8,000 rare diseases
- 75% of rare diseases affect children
- 30% of rare disease patients die before the age of 5
- 80% of rare diseases have identified genetic origins. Other rare diseases result from infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative.

## RARE DISEASE PATIENTS FACE COMMON PROBLEMS

- Lack of access to correct diagnosis
- Delays in diagnosis
- Lack of quality information on the disease
- Lack of scientific knowledge of the disease
- Significant social consequences for patients
- Lack of appropriate quality healthcare
- Inequalities and difficulties in access

## HOW CAN THINGS CHANGE?

- Implementing a comprehensive approach to rare diseases
- Developing appropriate public health policies
- Increasing international cooperation in scientific research
- Gaining and sharing scientific knowledge about all rare diseases and not just “frequent” ones
- Developing new diagnostic and therapeutic procedures
- Raising public awareness and getting people involved
- By facilitating the networking of patient groups to share their experience and best practices
- Showing support to the most isolated patients and their loved ones to create new patient communities or patient groups
- Providing comprehensive quality information to the rare disease community