RARE DISEASES
A major unmet medical need
There are some 6000 to 8000 life-threatening or chronically debilitating diseases.

27–36 million people in the EU have a rare disease.

80% are genetic diseases.

Some examples: cystic fibrosis, Duchenne muscular dystrophy, Huntington’s disease, acute myeloid leukaemia.

The main challenge for policies and related research and innovation is to **improve the lives of patients** with rare diseases. This requires cooperation between research and healthcare.
Rare Diseases – a major unmet medical need

P4P analysed the EU-funded project portfolio and identified five policy challenges:

▶ improving diagnosis, prevention and treatment
▶ facilitating the regulatory pathway for potential treatments
▶ effective and equal provision of healthcare for patients
▶ effective management and pooling of research and medical data
▶ contributing to and benefiting from global collaboration.
ACTIONS FOR POLICYMAKERS

- support integration and networking among EU research, patient and healthcare organisations
- support health technology assessment, standards and the evidence-base to guide policy
- develop legal and ethical agreements for collecting and exchanging health and genetic data
- adapt the implementation of legal requirements, especially for clinical trials
- collaborate globally through the International Rare Diseases Research Consortium (IRDiRC) to accelerate research.
RESEARCH FOR NEW AND BETTER SOLUTIONS

The EU has invested more than €1 billion in research and innovation on rare diseases through FP7 and Horizon 2020.

RESULTS SO FAR:

› gathering a critical mass of rare diseases cases and expertise for collaboration

› better understanding of the pathophysiology and natural history of rare diseases

› new and better prevention, treatments and diagnostics

› tools and approaches relating to effective and ethical data sharing

› new approaches for clinical trials in small populations.

24 European Reference Networks involving more than 900 healthcare units across Europe to tackle rare or low prevalence and complex diseases.
MORE INFORMATION

Rare diseases

P4P Report: Rare Diseases
a major unmet medical need

European Reference Networks

The International Rare Diseases Research Consortium

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