



Key Messages

- A rare disease is a condition which affects less than 5 in 10,000 people.
- There are over 6000 rare diseases affecting over 3.5 million people (1 in 17) in the UK.
- Collectively, rare diseases are not rare.
- However, NHS services to support people with rare diseases remain patchy and poorly integrated, meaning that individuals with rare diseases in the UK and their families struggle to access the help and support that they need.
- Rare Disease UK is a joint initiative of the Genetic Interest Group (the UK alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders) and others in response to these unmet health care needs.
- Rare Disease UK calls on the Government to take forward the European Council's Recommendation on a European action in the field of Rare Diseases, which calls on Member States to develop and implement plans or strategies to deal with rare disease.
- Rare Disease UK aims to create a draft strategy for rare disease in the UK with input from expert stakeholders from research, patient organisations, clinicians, the pharmaceutical industry, commissioners and government. This plan will coordinate:
 - o research into rare disease;
 - o prevention and diagnosis of rare diseases;
 - o treatment of rare diseases;
 - o information on rare diseases for patients and the public.
- The strategy will:
 - o help to ensure the targeted use of health care resources to maximise the benefits for all patients and families affected by rare disease across the UK;
 - o support the Government's health research agenda, including strengthening translational research;
 - o facilitate early and correct diagnosis and timely treatment, which could offer improved health and quality of life for people with rare diseases.
- A number of EU Member States have already developed plans or strategies for rare disease. The UK now needs to consider this as a positive opportunity for families and patients here.