

## **Summary of key findings**

### **Research**

- 1- Patients and families generally show an appreciation of the importance of research and a willingness to be involved in research
- 2- Patients are not well informed of research into their condition. This can result in low patient participation in research projects.
- 3- Patient organisations act as a vital source of information on research into rare diseases.

### **Diagnosis**

- 1- Patients and families affected by rare diseases wait too long for a correct diagnosis
- 2- A worrying number of people with rare diseases receive incorrect diagnoses before their final diagnosis is made
- 3- Patients and families worry about the level of awareness of rare disease among healthcare professionals
- 4- Patient organisations play an important role in the diagnosis of rare diseases
- 5- The experiences of patients and families of diagnosis can vary greatly raising concerns about equality of access and fair treatment in different parts of the country

### **Patient care, information and support**

- 1- Patients and families are not provided with enough information on all aspects of their condition, both at first diagnosis and subsequently
- 2- Information to patients and families must be provided in a range of formats and at various levels of medical and scientific detail to ensure full understanding and informed decision making.
- 3- Patient organisations are often the main or only source of information for rare disease patients.
- 4- There is a lack of support for rare disease patients with their medical and non-medical issues

### **Coordination of Care**

- 1- The majority of patients' care is poorly coordinated.
- 2- Patients have to attend multiple clinics for different aspects of their condition, often at a long distance from where they live
- 3- The majority of patients do not have access to a specialist centre for their condition

- 4- Patients frequently experience problems with medical, psychological, financial, social and other issues at transition periods

**Access to treatment**

- 1- Trying to obtain medicines can be distressing for some patients and families
- 2- There is no licensed treatment available for most patients with rare diseases
- 3- Some patients are informed of off-label or unlicensed medicines but often patients and families have to inform their doctor
- 4- Patients and families experience inconsistencies in access to medicines

Source: Rare Disease UK