Expected outcomes

1. An integrated and harmonized set of instruments to assess and monitor socio-economic burden and HRQOL of patients affected by RD and their caregivers. These instruments will help measure the impact of new policies, interventions, treatments and diagnostic techniques, adapted to different RD and EU member states/languages, permitting the extrapolation to the rest of the EU and to a wide range of RD.

2. A detailed analysis of the services (health and social care) received by people with specific RD in different EU countries, including the identification of formal and informal care.

3. A report on the current socioeconomic and HRQOL status of RD patients and caregivers for the selected RD and EU countries.

The results and deliverables that emerge from this project will stimulate the future comparability and monitoring of RD in Europe as well as anticipate future information needs. The outcomes will be made readily accessible and posted on the websites of the participating Patient Associations, BURQOL-RD, EUROPLAN and EC public websites. The outcomes will also be distributed to health policy makers in the EU, while the scientific outcomes will be disseminated by means of scientific papers and meetings.
BURQOL-RD
www.burqol-rd.com

Despite the low prevalence of Rare Diseases (RD) (5/10,000 persons), more than 5000 RD are recognised and hence, over 30 million EU citizens suffer from these conditions. Most cases RD are life-threatening, chronic and debilitating, requiring long term specialist and costly formal and informal care. The lack of effective diagnoses and treatments often underlies the shortened life expectancy and quality of life of these patients.

These characteristics mean that RD require the combined efforts of health and social care professionals, politicians, managers and researchers to increase the availability of effective disease management tools to improve care and to extend both life expectancy and Health Related Quality of Life (HRQOL).

Target RD

· Cystic Fibrosis
· Prader-Willi Syndrome
· Haemophilia
· Epidermolysis Bullosa
· Histiocytosis
· Juvenile Idiopathic Arthritis
· Mucopolysaccharidosis
· Duchenne Muscular Dystrophy

A survey on patients and caregivers will be held in these countries:

- Spain
- France
- UK
- Italy
- Sweden
- Germany
- Hungary
- Bulgaria

The RD targeted in the pilot study of BURQOL-RD are:

- Cystic Fibrosis
- Prader-Willi Syndrome
- Scleroderma
- Epidermolysis Bullosa
- Histiocytosis
- Juvenile Idiopathic Arthritis
- Mucopolysaccharidosis
- Duchenne Muscular Dystrophy

Aims

The main aim of BURQOL-RD is to generate a model to quantify the socio-economic costs and HRQOL of both patients and caregivers, for ten RD in eight different European countries.

This model will be adaptable and sufficiently sensitive to capture the differences in the distinct Health and Social Care Systems in the EU Member States. As such the BURQOL-RD model will provide an integrated and harmonised means to assess the impact of new public health policies, interventions and treatments for RD “in” and “among” EU Member States.

The tools developed by BURQOL-RD will build on the ongoing EUROPLAN project and BURQOL-RD will also incorporate strategies previously developed by EURORDIS. Moreover, the associated dissemination activities undertaken by BURQOL-RD will also improve RD awareness and literacy among European citizens.

Given the nature and the goals of the project, it is clear that the fundamental beneficiary of the results of this project will be the families and caregivers of those affected by RD, a group that is often overlooked when considering such devastating diseases.

The collaboration of national patients’ associations and federations for the specific RDs to be addressed in the project is fundamental to ensure that all the objectives are successfully reached.