

- Survey Observatory of Rare Diseases 2012

Head :Heuyer Thomas, Observatoire des Maladies Rares

Last update : 05/09/2014 | Version : 1 | ID : 8422

General	
Identification	
Detailed name	Survey Observatory of Rare Diseases 2012
General Aspects	
Medical area	Anatomy - Cytology Cardiology Endocrinology and metabolism Hematology Immunology Infectious diseases Neurology Ophthalmology Otolaryngology or ENT Pneumology Rare diseases
Health determinants	Genetic Iatrogenic Social and psychosocial factors
Keywords	Diagnosis, announcement, medical treatments, financial difficulties, coordination between physicians, support, interpersonal surrounding, orphan medicinal products., monitoring, healthcare
Scientific investigator(s) (Contact)	
Name of the director	Heuyer
Surname	Thomas
Address	96 rue Didot 75014 Paris
Phone	+33 (0)1 56 53 81 29
Email	theuyer@maladiesrares.org
Unit	Observatoire des Maladies Rares
Organization	Maladies Rares Info
Collaborations	

Funding	
Funding status	Mixed
Details	AFM-Téléthon (donations) Fondation Medtronic Fondation du LEEM
Governance of the database	
Sponsor(s) or organisation(s) responsible	Maladies Rares Info Services
Organisation status	Both
Additional contact	
Main features	
Type of database	
Type of database	Study databases
Study databases (details)	Not-repeated cross-sectional studies (except case control studies)
Database recruitment is carried out by an intermediary	A selection of health institutions and services
Database recruitment is carried out as part of an interventional study	No
Additional information regarding sample selection.	The survey was proposed to each person contacting Maladies Rares Info Services (via email or phone) if he/she corresponded to the inclusion criteria. During this period, the survey was proposed to 361 people having contacted Maladies Rares Info Services. In parallel, the survey was proposed to 259 additional people taking part in the service "Meet the patients" and agreeing to be part of the different studies of Maladies Rares Info Services.
Database objective	
Main objective	The objective of the Observatory is to provide to all stakeholders involved in the fight against rare diseases reliable data highlighting important issues to which patients and their families are confronted. These data must allow not only to identify those issues but also to bring forward proposals to

mitigate them.
This survey covered three topics:
- Communication about the diagnosis;
- Practical difficulties linked to drugs and other medical products;
- Coordination among the healthcare workforce.

Inclusion criteria

- Being a patient or a family member of the patient (parents, grand-parents, children, spouses);
- Being affected by a rare disease (prevalence rate inferior or equal to one out of 2000 people);
- Being able to participate in the survey (the survey was not proposed to people in distress, in a state of psychological fragility...)

Population type

Age

- Newborns (birth to 28 days)
- Infant (28 days to 2 years)
- Early childhood (2 to 5 years)
- Childhood (6 to 13 years)
- Adolescence (13 to 18 years)
- Adulthood (19 to 24 years)
- Adulthood (25 to 44 years)
- Adulthood (45 to 64 years)
- Elderly (65 to 79 years)
- Great age (80 years and more)

Population covered

Sick population

Gender

- Male
- Woman

Geography area

National

Detail of the geography area

France

Data collection

Dates

Date of first collection (YYYY or MM/YYYY)

09/2012

Date of last collection (YYYY or MM/YYYY)

12/2012

Size of the database

Size of the database (number of individuals)

< 500 individuals

Details of the number of

244 questionnaires were collected (137 paper

individuals

forms and 107 online questionnaires) and 239 were included in the final analysis.

Data

Database activity Data collection completed

Type of data collected Declarative data

Declarative data (detail) Paper self-questionnaire

Presence of a biobank No

Health parameters studied Health event/morbidity
Health care consumption and services
Quality of life/health perception
Others

Care consumption (detail) Hospitalization
Medical/paramedical consultation
Medicines consumption

Other (detail) Patient satisfaction.

Procedures

Participant monitoring No

Links to administrative sources No

Promotion and access

Promotion

Link to the document <http://www.maladiesraresinfo.org/services-proposes/89.html>

Access

Terms of data access (charter for data provision, format of data, availability delay) Contact the scientific investigator.

Access to aggregated data Access on specific project only

Access to individual data Access on specific project only