

# - Survey Observatory of Rare Diseases 2012

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| General                                 |                                                                                                                                                                                                           |
|-----------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Identification                          |                                                                                                                                                                                                           |
| Detailed name                           | Survey Observatory of Rare Diseases 2012                                                                                                                                                                  |
| General Aspects                         |                                                                                                                                                                                                           |
| Medical area                            | Anatomy - Cytology<br>Cardiology<br>Endocrinology and metabolism<br>Hematology<br>Immunology<br>Infectious diseases<br>Neurology<br>Ophthalmology<br>Otolaryngology or ENT<br>Pneumology<br>Rare diseases |
| Health determinants                     | Genetic<br>Iatrogenic<br>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)<br>(Contact) |                                                                                                                                                                                                           |
| Name of the director                    | Heuyer                                                                                                                                                                                                    |
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| Unit                                    | Observatoire des Maladies Rares                                                                                                                                                                           |
| Organization                            | Maladies Rares Info                                                                                                                                                                                       |
| Collaborations                          |                                                                                                                                                                                                           |

| Funding                                                                |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
|------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Funding status                                                         | Mixed                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| Details                                                                | AFM-Téléthon (donations) Fondation Medtronic<br>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.                     | <p>The survey was proposed to each person contacting Maladies Rares Info Services (via email or phone) if he/she corresponded to the inclusion criteria.</p> <p>During this period, the survey was proposed to 361 people having contacted Maladies Rares Info Services.</p> <p>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.</p> |
| Database objective                                                     |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
| Main objective                                                         | <p>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</p>                                                                                                                                                              |

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