# - Survey Observatory of Rare Diseases 2011

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## General

### Identification

| | |
|---|---|
| Detailed name | Survey Observatory of Rare Diseases 2011 |
| CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation | -- |

### General Aspects

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|---|---|
| Medical area | Anatomy - Cytology<br>Cardiology<br>Endocrinology and metabolism<br>Hematology<br>Immunology<br>Infectious diseases<br>Neurology<br>Ophthalmology<br>Pneumology<br>Psychology and psychiatry<br>Rare diseases |
| Keywords | Long term illness, chronic disease, misdiagnosis, error, access to information, healthcare, out of pocket payments, renouncement to healthcare., ALD, medical care |

### Scientific investigator(s) (Contact)

| | |
|---|---|
| Name of the director | Heuyer |
| Surname | Thomas |
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| Unit | Observatoire des Maladies Rares |
| Organization | Maladies Rares Info |

## Collaborations

## Funding

| | |
|---|---|
| Funding status | Private |
| Details | AFM-Téléthon (donations) Fondation Medtronic Fondation du LEEM |

## Governance of the database

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| Sponsor(s) or organisation(s) responsible | Maladies Rares Info Services |
| Organisation status | Both |

## Additional contact

## Main features

## Type of database

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|---|---|
| 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. |

## Database objective

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| Main objective | The objective of the Observatory is to provide to all skateholders involved in the fight against rare diseases reliable data highlighting important issues to which patients and their families are confronted.<br>These data must allow not only to identify those issues but also to bring forward proposals to mitigate them.<br>This survey studied three topics:<br>- Misdiagnosis;<br>- Access to information;<br>- Financial support for healthcare, products and |

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| | service delivery. |
| Inclusion criteria | - Status: patients, their spouses, their children or their parents.<br>- Pathology: prevalence rate inferior or equal to one out of 2000 people, according to the data presented in Orphanet on the prevalence of rare diseases<br>- In the absence of a diagnosis: in this situation, or the patient is likely to be affected by a rare disease, or the situation referred to hypochondria, mythomania...<br>The Telephone Contact and Information person was in charge of assessing whether the situation referred to by the caller corresponded to the first or the second hypothesis and wether it was worth collecting data. |

## Population type

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| Age | Newborns (birth to 28 days)<br>Infant (28 days to 2 years)<br>Early childhood (2 to 5 years)<br>Childhood (6 to 13 years)<br>Adolescence (13 to 18 years)<br>Adulthood (19 to 24 years)<br>Adulthood (25 to 44 years)<br>Adulthood (45 to 64 years)<br>Elderly (65 to 79 years)<br>Great age (80 years and more) |
| Population covered | Sick population |
| Gender | Male<br>Woman |
| Geography area | National |
| Detail of the geography area | France |

## Data collection

### Dates

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| Date of first collection (YYYY or MM/YYYY) | 03/2011 |
| Date of last collection (YYYY or MM/YYYY) | 01/2012 |

### Size of the database

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| Size of the database (number of | < 500 individuals |

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| individuals) | |
| Details of the number of individuals | 198 individuals out of the 361 asked participated in the quantitative survey. (322 individuals participated in the qualitative study allowing to build the quantitative questionnaire) |

## Data

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| 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<br>Health care consumption and services<br>Quality of life/health perception |
| Care consumption (detail) | Hospitalization<br>Medical/paramedical consultation<br>Medicines consumption |

## Procedures

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| 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

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|---|---|
| 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 |