

- 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

Medical area
Anatomy - Cytology
Cardiology
Endocrinology and metabolism
Hematology
Immunology
Infectious diseases
Neurology
Ophthalmology
Pneumology
Psychology and psychiatry
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

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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	
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.
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 mitigate them.</p> <p>This survey studied three topics:</p> <ul style="list-style-type: none"> - Misdiagnosis; - Access to information; - Financial support for healthcare, products and

service delivery.

Inclusion criteria

- Status: patients, their spouses, their children or their parents.
- 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
- 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...

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 whether it was worth collecting data.

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)

03/2011

Date of last collection (YYYY or MM/YYYY)

01/2012

Size of the database

Size of the database (number of

< 500 individuals

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

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

Care consumption (detail)

Hospitalization
Medical/paramedical consultation
Medicines consumption

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