- 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 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)

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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 skateholders 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; - Pratical 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, chilren, 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