FGDR - The French Gaucher disease registry

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General

Identification

Detailed name	The French Gaucher disease registry
Sign or acronym	FGDR
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CNIL 05-1099

General Aspects

Medical area	Hematology Internal medicine Rare diseases Rheumatology
Health determinants	Genetic
Others (details)	Gaucher's disease
Keywords	pharmacoeconomics

Scientific investigator(s) (Contact)

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Organization	Assistance Publique des Hôpitaux de

Collaborations

Funding

Funding status	Mixed
Details	Inserm, InVS, VAINCRE LES MALADIES LYSOSOMALES

Governance of the database

Sponsor(s) or organisation(s) responsible	Assistance Publique des Hopitaux de Paris
Organisation status	Public

Additional contact

Main features

Type of database

Type of database	Morbidity registers
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.	Identifying cases is done by cross-checking the following data sources: - data from hospital doctors centralizing the data towards the reference center -data from biological laboratories specializing in medical diagnostic and biological follow-up of

	patients with Gaucher's disease - data from the CNAM via le National Secretariat for Hereditary Metabolic Disorders (SNMMH)

Database objective

Main objective	- Carry out a descriptive epidemiological analysis of the population suffering from Gaucher's disease in France (prevalence estimated at 1/100,000) which makes it possible to describe the natural history of the disease, apprehend and analyze the care practices, and evaluate them. - Develop lines of research: * Epidemiology and modeling of the change in biological and clinical markers with and without treatment * Pregnancy and Gaucher's disease * Parkinson's disease and Gaucher's disease * Monoclonal peak and Gaucher's disease * Type-3 Gaucher's disease * Gaucher's disease in children - Evaluate the efficacy and the tolerance of the treatments, optimize them and carry out pharmacoeconomic studies - Establish any genotype-phenotype relations - Detect predictive factors of complications and identify the biomarkers making it possible to follow the disease with and without treatment - Select the patients for new therapeutic options
Inclusion criteria	All of the patients diagnosed and/or followed for Gaucher's disease over all of France. Gaucher's disease is confirmed by the dosage of the enzyme activity of the glucocerebrosidase.

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)	1991

Size of the database

Size of the database (number of individuals)	[500-1000[individuals
Details of the number of individuals	560 (31/01/2011)

Data

Database activity	Current data collection
Type of data collected	Clinical data Paraclinical data Biological data Administrative data
Clinical data (detail)	Direct physical measures
Paraclinical data (detail)	Follow-up radiological paraclinical examinations (MRI, Scintigraphy, osteodensitometry, ultrasound, etc.)
Biological data (detail)	Diagnostics and biological follow-up of the disease
Administrative data (detail)	health insurance reimbursement data
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality Health care consumption and services
Care consumption (detail)	Hospitalization Medical/paramedical consultation Medicines consumption

Procedures

Data collection method	pharmacoeconomics
Participant monitoring	Yes

Details on monitoring of participants	Subjects are followed for an undetermined duration using the various sources of data which are: - medical dossiers of hospital centers and from the family doctor - the social security fund for medical care and medication reimbursement data - skills centers
Links to administrative sources	Yes
Linked administrative sources (detail)	CNAM data

Promotion and access

Promotion

Link to the document	http://www.ncbi.nlm.nih.gov/pubmed?term
Link to the document	http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2945057/
Link to the document	http://cetl.net/IMG/pdf/Stirnemann_Registre_Gaucher_CETG_2010.pdf
Link to the document	http://www.ojrd.com/content/7/1/77/abstract

Access

Terms of data access (charter for data provision, format of data, availability delay)	Access subject to authorization from the scientific board of the registry, formed by the members of the Evaluation Committee of the Treatment for Gaucher's disease
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only