

FGDR - The French Gaucher disease registry

Head :Belmatoug Nadia, Centre de référence sur les maladies lysosomales (coordinatrice)
Stirnemann Jérôme, Centre de Référence des Maladies Lysosomales (CRML) Laboratoire de
biostatistique (Pr France Mentré) - INSERM - UMR 738 - Hôpital Bichat

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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)	
Name of the director	Belmatoug
Surname	Nadia
Address	Service de médecine interne-Hôpital Beaujon - 100bd du général leclerc - 92110 Clichy
Phone	+ 33 (0)1 40 87 52 86
Email	nadia.belmatoug@bjn.aphp.fr
Unit	Centre de référence sur les maladies lysosomales (coordinatrice)

Organization	Assistance Publique des Hôpitaux de
Name of the director	Stirnemann
Surname	Jérôme
Email	jerome.stirnemann@jvr.aphp.fr
Unit	Centre de Référence des Maladies Lysosomales (CRML) Laboratoire de biostatistique (Pr France Mentré) - INSERM - UMR 738 - Hôpital Bichat
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