

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

Last update : 11/30/2012 | Version : 1 | ID : 56

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 pharmaco-economic 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 pharmacoconomics

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
---------------------------	---------------------------------