

- French Registry for Pompe Disease (Certified Registry)

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General

Identification

Detailed name French Registry for Pompe Disease (Certified Registry)

CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation CNIL 909139

General Aspects

Medical area Endocrinology and metabolism
Neurology
Rare diseases

Health determinants Medicine

Keywords assessment, tolerance, efficacy

Scientific investigator(s) (Contact)

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Organization Assistance Publique des Hôpitaux de

Collaborations

Funding

| | |
|----------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Funding status | Mixed |
| Details | French-Speaking Association for Glycogen Storage Disease (AFG), French Muscular Dystrophy Association (AFM), Genzyme laboratory, InVS-INSERM. |
| Governance of the database | |
| Sponsor(s) or organisation(s) responsible | Association Institut de Myologie (AIM) |
| Organisation status | Private |
| Additional contact | |
| Main features | |
| Type of database | |
| Type of database | Morbidity registers |
| Additional information regarding sample selection. | Selection of subjects fulfilling the required inclusion criteria. |
| Database objective | |
| Main objective | <p>Objectives of the registry in the field of public health:</p> <ul style="list-style-type: none"> - To collect epidemiological data on Pompe disease in France and calculate exactly how many patients are affected by this disease. - To assess the tolerance and efficacy of Myozyme, the only treatment currently effective against Pompe disease. However, standardised long-term follow-up for patients is necessary due the significant cost on the healthcare system. <p>Homogeneous and standardised information on patients suffering from Pompe disease and treated with Myozyme is collected every six months for patients undergoing treatment and every twelve months for other patients.</p> <p>Objectives of the registry in the field of clinical research:</p> <ul style="list-style-type: none"> -This registry is also aiming to improve knowledge on the natural history of the disease in untreated patients, as well as determine the frequency and severity of unusual disease manifestations (cerebral arterial malformations, digestive problems, sphincter problems, etc.). <p>Objectives of the registry in the therapeutic field:</p> |

- It is probable that other therapeutic approaches will be developed in the years to come, at which point a separate registry will certainly have a defining role in recruiting patients for these future trials, and may facilitate the comparison of results with results obtained from patients undergoing enzyme replacement therapy.

Inclusion criteria

Patients are only included in the registry when the diagnosis of Pompe disease is confirmed by biochemical (detection of acid alpha-glucosidase deficiency in the blood, muscle or fibroblasts) and/or genetic analysis.

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

Metropolitan France.

Data collection

Dates

Date of first collection (YYYY or MM/YYYY)

2004

Size of the database

Size of the database (number of individuals)

< 500 individuals

Details of the number of individuals

The calculated incidence of all forms of Pompe disease is estimated at 1/40,000. Over 100 patients have already been diagnosed with Pompe disease in France.

Data

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|------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------|
| 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) | 6-minute walking test, muscular test, timed tests, MFM scale, vital capacity measurement, respiratory pressure measurement, muscular biopsy. |
| Biological data (detail) | Acid alpha-glucosidase activity, urinary Glc4, CPK. |
| Administrative data (detail) | Identification data. |
| Presence of a biobank | No |
| Health parameters studied | Health event/morbidity Health event/mortality Health care consumption and services |
| Care consumption (detail) | Hospitalization Medicines consumption |

Procedures

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|---------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Data collection method | Collection of homogenous and standardised information on treated or untreated patients with Pompe disease according to an established protocol (recommendations from the Pompe Disease Treatment Evaluation Committee, CETP). Follow-up parameters corresponding to recommendations from the CNR-MR (National Committee for Rare Disease Registries) - CETP are collected every six months for patients undergoing treatment and every twelve months for other patients. Some of the data collected on the French registry shall also overlap with data from Genzyme's registry. As such, data sharing is planned in order to meet the requirements of the European Medicines Agency (EMA), which requests health authorities to transmit data on efficacy and tolerance to the international registry. |
| Participant monitoring | Yes |
| Details on monitoring of participants | - Annual clinical assessments for untreated patients and every six months for patients on enzyme replacement therapy - vital status. |

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|---------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------|
| Links to administrative sources | No |
| Promotion and access | |
| Promotion | |
| Link to the document | http://tinyurl.com/PUBMED-FPR |
| Description | Liste des publications dans Pubmed |
| Access | |
| Terms of data access (charter for data provision, format of data, availability delay) | Database hosted on a 4D server with access by request. Secure internet access under construction. |
| Access to aggregated data | Access on specific project only |
| Access to individual data | Access on specific project only |