

FOCUS - Cohort of Patients with Hypertrophic Cardiomyopathy: Prospective Screening for Fabry Disease

Head :Hagège Albert, Inserm U633 Cardiology Department at Georges Pompidou European Hospital [Hôpital Européen Georges-Pompidou]

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General

Identification

| | |
|-----------------|--|
| Detailed name | Cohort of Patients with Hypertrophic Cardiomyopathy: Prospective Screening for Fabry Disease |
| Sign or acronym | FOCUS |

General Aspects

| | |
|---------------------|--|
| Medical area | Biology Cardiology Rare diseases |
| Pathology (details) | Fabry disease, hypertrophic cardiomyopathy |
| Keywords | filter paper, α -galactosidase A, enzyme, screening, prevalence |

Scientific investigator(s) (Contact)

| | |
|----------------------|---|
| Name of the director | Hagège |
| Surname | Albert |
| Address | 20 Rue Leblanc, 75015 Paris |
| Email | albert.hagege@egp.aphp.fr |
| Unit | Inserm U633 Cardiology Department at Georges Pompidou European Hospital [Hôpital Européen Georges-Pompidou] |
| Organization | Paris Public Hospitals |

Collaborations

Funding

| | |
|----------------|---------|
| Funding status | Private |
|----------------|---------|

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|--|---|
| Details | Reference Centre for Fabry Disease and Hereditary Connective Tissue Diseases?Genzyme SAS |
| Governance of the database | |
| Sponsor(s) or organisation(s) responsible | Paris Public Hospitals (AP-HP) |
| Organisation status | Public |
| Additional contact | |
| Main features | |
| Type of database | |
| Type of database | Study databases |
| Study databases (details) | Cohort study |
| 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 |
| Database objective | |
| Main objective | To estimate the prevalence of Fabry disease in a population of men and women with hypertrophic cardiomyopathy (HCM) consistent with primary cardiomyopathy. |
| Inclusion criteria | ? men and women ? between 18 and 79 years old ? with hypertrophic cardiomyopathy |
| Population type | |
| Age | Adulthood (19 to 24 years) Adulthood (25 to 44 years) Adulthood (45 to 64 years) Elderly (65 to 79 years) |
| Population covered | Sick population |
| Gender | Male Woman |
| Geography area | National |

| | |
|--|---|
| Detail of the geography area | France (30 cardiology centres) |
| Data collection | |
| Dates | |
| Date of first collection (YYYY or MM/YYYY) | 07/2006 |
| Date of last collection (YYYY or MM/YYYY) | 07/2008 |
| Size of the database | |
| Size of the database (number of individuals) | < 500 individuals |
| Details of the number of individuals | 392 subjects ? 278 men ? 114 women |
| Data | |
| Database activity | Data collection completed |
| Type of data collected | Clinical data Declarative data Biological data |
| Clinical data (detail) | Direct physical measures |
| Details of collected clinical data | ? demographic characteristics ? medical history ? echocardiographic parameters ? current drug therapies |
| Declarative data (detail) | Paper self-questionnaire |
| Details of collected declarative data | Characteristic signs or symptoms of Fabry disease. |
| Biological data (detail) | Blood sample (venipuncture for microdose screening) |
| Presence of a biobank | Yes |
| Contents of biobank | Whole blood DNA |
| Details of biobank content | Samples shipped to central laboratory within 96 hours. |
| Procedures | |

| | |
|---|---|
| Data collection method | Systematic screening using a filter paper test (to determine β -galactosidase A activity). |
| Participant monitoring | Yes |
| Monitoring procedures | Monitoring by contact with the referring doctor |
| Links to administrative sources | No |
| Promotion and access | |
| Promotion | |
| Link to the document | http://heart.bmj.com/content/97/2/131.full#ack-1 |
| Access | |
| Terms of data access (charter for data provision, format of data, availability delay) | Contact the scientist in charge. |
| Access to aggregated data | Access on specific project only |
| Access to individual data | Access on specific project only |