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, ?-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

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 ?-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