

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

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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)

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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