OPALE - Observatory of Patients with laminopathies and emerinopathies

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General

Identification

Detailed name	Observatory of Patients with laminopathies and emerinopathies
Sign or acronym	OPALE
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CNIL, CCTIRS n°13.135, CPP n°58-12 (31/08/2012), ANSM n°ID RCB : 2012-A00791-42

General Aspects

Medical area	Cardiology Endocrinology and metabolism Neurology Pediatrics Rare diseases
Pathology (details)	Laminopathies and Emerinopathies
Health determinants	Genetic
Keywords	Natural history; muscular, cardiac, orthopaedic and metabolic involvements, joint contractures, cardiomyopathies, premature aging, progeria, respiratory, myopathies, lipodystrophies

Scientific investigator(s) (Contact)

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Organization	INSERM - Institut National de la Santé et de la Recherche
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Organization	AP-HP

Collaborations

Participation in projects, networks and consortia	Yes
Details	French Network of Emery-Dreifuss muscular dystrophy and other nuclear envelope diseases

Funding

Funding status	Mixed
Details	Association Institut de Myologie, Inserm, AP-HP

Governance of the database

Sponsor(s) or organisation(s) responsible	INSERM - Institut National de la Santé et de la Recherche Médicale
Organisation status	Public
Sponsor(s) or organisation(s) responsible	AP-HP

Organisation status	Public
Sponsor(s) or organisation(s) responsible	Association Institut de Myologie
Organisation status	Private
Presence of scientific or steering committees	Yes

Additional contact

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Unit	Pole Base de données, Institut de Myologie
Organization	Association Institut de Myologie

Main features

Type of database

Type of database	Study databases
Study databases (details)	Longitudinal study (except cohorts)
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 precise the natural history of musculrr, cardiac, respiratory, orthopaedic and metabolic involvements of patients suffering from laminopathies and emerinopathies. to identify cardiovascular, neurologic and respiratory prognosis factors.

	to identify obstetrical and perioperative complications related to laminopathies and emerinopathies. to identify correlations between LMNA/EMD gene mutations and the observed phenotypes. to have a repository ready for inclusion of patients in future therapeutic trials.
Inclusion criteria	all patient carrying a LMNA or a EMD gene mutation, including the paediatric population, registered to Social Security Helathcare System.

Population type

Age	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)
Population covered	Sick population
Pathology	G71 - Primary disorders of muscles
	E88 - Other metabolic disorders
	I42 - Cardiomyopathy
Gender	Male Woman
Geography area	National
Detail of the geography area	France

Data collection

Dates

Date of first collection (YYYY or MM/YYYY)	2000

Size of the database

Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	320

Data

Database activity	Current data collection
Type of data collected	Clinical data Paraclinical data Biological data Administrative data
Clinical data (detail)	Direct physical measures Medical registration
Paraclinical data (detail)	imaging
Biological data (detail)	blood and urine biological routine checkup, recorded along the followup of the patient
Administrative data (detail)	First name initial, Last name initial, gender, date of birth, place of birth, medical file number, educational maximal level
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality

Procedures

Data collection method	the data entry will be performed by investigators of rare diseaes centers, together with clinical research assistants.
Participant monitoring	Yes
Details on monitoring of participants	along the standard followup of the patient
Links to administrative sources	Yes
Linked administrative sources (detail)	town council of birth place

Promotion and access

Promotion

Link to the document	http://www.institut-myologie.org/

Access

Terms of data access (charter for data provision, format of	future publications, data access to investigators

data, availability delay)	
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only