

OPALE - Observatory of Patients with laminopathies and emeropathies

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Last update : 02/17/2020 | Version : 3 | ID : 5054

General

Identification

Detailed name Observatory of Patients with laminopathies and emeropathies

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)

Name of the director Bonne

Surname Gisèle

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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
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Details	French Network of Emery-Dreifuss muscular dystrophy and other nuclear envelope diseases
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Funding

Funding status	Mixed
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Details	Association Institut de Myologie, Inserm, AP-HP
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Governance of the database

Sponsor(s) or organisation(s) responsible	INSERM - Institut National de la Santé et de la Recherche Médicale
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Organisation status	Public
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Sponsor(s) or organisation(s) responsible	AP-HP
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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	
Name of the contact	Ben Yaou
Surname	Rabah
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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 muscular, 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 diseases 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