RaDiCo-MPS - RaDiCo-MPS - Mucopolysaccharidosis patients in France in the era of specific therapeutics

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General

Identification

Detailed name	RaDiCo-MPS - Mucopolysaccharidosis patients in France in the era of specific therapeutics
Sign or acronym	RaDiCo-MPS
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS n° 16-570 / CPP n°DC-2015-2482

General Aspects

Medical area	Cardiology Dermatology, venereology Disability/handicap Endocrinology and metabolism Gastroenterology et hepatology Neurology Odontology Ophthalmology Otolaryngology or ENT Pediatrics Pneumology Psychology and psychiatry Rare diseases Rheumatology Urology, andrology and nephrology
Study in connection with Covid-19	No
Pathology (details)	The mucopolysaccharidoses (MPS) are lysosomal storage disorders caused by accumulation of sulphated carbohydrate polymers in the lysosomes leading to a cascade of multisystemic disease manifestations. The sulphated polymers are composed of a central core protein attached to disaccharide branches deriving from sulphated monosaccharides or glycosaminoglycans (GAGs, formerly termed mucopolysaccharides,). The

	primary storage products are: dermatan sulphate, chiefly a constituent of connective tissues; heparan sulphate, chiefly a constituent of cellular membranes; and keratan sulphate and chondroitin sulphate, found abundantly in the cartilages and in the cornea. GAG excretion in urine allows screening for MPS both quantitatively (elevated urinary GAG content) and qualitatively (characteristic profile of sulphated derivatives). MPS are rare diseases; their overall incidence varies over the countries and ethnicities but is estimated to be approximately 1:25 000 to 1:30 000 births. Inheritance is autosomal recessive for all but MPS-II (or Hunter disease) that is an X-linked disorder. The genes responsible for the 11 enzyme deficiencies corresponding to the following 7 clinical subtypes have been identified. MPS are chronic, progressive multivisceral diseases. Age at first symptoms may vary according to the severity of the disease. They can occur in early infancy or early childhood in the severe cases (the most severe forms can even manifest antenatally).
Scientific investigator(s) (Contact)	
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Surname	Bénédicte
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Name of the director	Billette
Surname	Thierry
Unit	UMR 1141
Organization	Institut National de la Santé et de la Recherche Médicale (Inserm)
Collaborations	
Funding	
Funding status	Mixed

Details	The RaDiCo-MPS cohort is funded by the French « Investissements d'Avenir » cohorts programme, Grant « ANR » 10-COHO-0003. This study is also supported by industrial funding through a public-private partnership.

Governance of the database

Sponsor(s) or organisation(s) responsible	Institut National de la Santé et de la Recherche Médicale (Inserm)
Organisation status	Public
Presence of scientific or steering committees	Yes
Labelling and database evaluation	Security audit certification of the database. Data management and continuous quality control of data.

Additional contact

Main features

Type of database

Type of database	Morbidity registers
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	The primary objective of the RaDiCo-MPS cohort is to characterize the epidemiology and natural history of MPS diseases by building a retrospective and prospective collection of extensive phenotypic data from French MPS patients.
Inclusion criteria	The RaDiCo-MPS Cohort inclusion criteria are the following: ? Confirmed diagnosis of MPS based on clinically relevant enzyme deficiency, with abnormally elevated GAG urinary excretion and/or identification of pathogenic mutations. ? Signed informed consent or parents/guardian non-opposition for deceased patients (minor or protected major)

	There are no non-inclusion criteria.
Population type	
Age	Newborns (birth to 28 days) 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)
Population covered	Sick population
Pathology	E76 - Disorders of glycosaminoglycan metabolism
Gender	Male Woman
Geography area	National
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	2017
Size of the database	
Size of the database (number of individuals)	< 500 individuals
Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data
Clinical data (detail)	Direct physical measures Medical registration
Details of collected clinical data	Growth, signs, symptoms and complications for each system (cardiologic, pulmonary, neurologic, gastrologic,...), psychomotor milestones and cognitive evolution, molecular data ...
Declarative data (detail)	Paper self-questionnaire

	Internet self-questionnaire Face to face interview
Details of collected declarative data	Vineland II, Quality of life questionnaires, Patient Global Impression of Improvement (PGI-I), ...
Paraclinical data (detail)	Echocardiography, cerebral imaging, pulmonary function testing, ...
Biological data (detail)	Urinary GAG, enzyme activities, before and during specific treatment, ...
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality Quality of life/health perception
Procedures	
Data collection method	eCRF in secure web access, secure cloud and HADS hosting
Classifications used	Drug dictionary (DCIs)
Quality procedure(s) used	Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and query system)
Participant monitoring	Yes
Monitoring procedures	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Followed pathology	E76 - Disorders of glycosaminoglycan metabolism
Links to administrative sources	No
Promotion and access	
Promotion	
Access	
Presence of document that lists variables and coding procedures	Yes
Terms of data access (charter for data provision, format of data, availability delay)	Requests for access to RaDiCo-MPS data (aggregated or individual) will be considered by the Scientific Committee following the submission of a summary of a specific research project, as defined in the Charter of access to resources. Requests

	should be sent to: mps@radico.fr
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only