

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

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## Général

### Identification

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

Sigle ou acronyme RaDiCo-MPS

Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.) CCTIRS n° 16-570 / CPP n°DC-2015-2482

### Thématiques générales

Domaine médical

- 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

Etude en lien avec la Covid-19 No

Pathologie, précisions

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).

### Responsable(s) scientifique(s)

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Nom du responsable	Billette
Prénom	Thierry
Laboratoire	UMR 1141
Organisme	Institut National de la Santé et de la Recherche Médicale (Inserm)

### Collaborations

### Financements

Financements	Mixed
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Précisions	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-
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private partnership.

## Gouvernance de la base de données

Organisation(s) responsable(s) ou promoteur

Institut National de la Santé et de la Recherche Médicale (Inserm)

Statut de l'organisation

Secteur Public

Existence de comités scientifique ou de pilotage

Yes

Labellisations et évaluations de la base de données

Security audit certification of the database. Data management and continuous quality control of data.

## Contact(s) supplémentaire(s)

## Caractéristiques

### Type de base de données

Type de base de données

Morbidity registers

Origine du recrutement des participants

A selection of health institutions and services

Le recrutement dans la base de données s'effectue dans le cadre d'une étude interventionnelle

No

### Objectif de la base de données

Objectif principal

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.

Critères d'inclusion

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.

## Type de population

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 concernée Sick population

Pathologie E76 - Disorders of glycosaminoglycan metabolism

Sexe  
Male  
Woman

Champ géographique National

## Collecte

### Dates

Année du premier recueil 2017

### Taille de la base de données

Taille de la base de données (en nombre d'individus) < 500 individuals

## Données

Activité de la base Current data collection

Type de données recueillies  
Clinical data  
Declarative data  
Paraclinical data  
Biological data

Données cliniques, précisions  
Direct physical measures  
Medical registration

Détail des données cliniques recueillies  
Growth, signs, symptoms and complications for each system (cardiologic, pulmonary, neurologic, gastrologic,...), psychomotor milestones and cognitive evolution, molecular data ...

Données déclaratives, précisions  
Paper self-questionnaire  
Internet self-questionnaire  
Face to face interview

Détail des données déclaratives recueillies	Vineland II, Quality of life questionnaires, Patient Global Impression of Improvement (PGI-I), ....
Données paracliniques, précisions	Echocardiography, cerebral imaging, pulmonary function testing, ....
Données biologiques, précisions	Urinary GAG, enzyme activities, before and during specific treatment, ....
Existence d'une bibliothèque	No
Paramètres de santé étudiés	Health event/morbidity Health event/mortality Quality of life/health perception
<b>Modalités</b>	
Mode de recueil des données	eCRF in secure web access, secure cloud and HADS hosting
Nomenclatures employées	Drug dictionary (DCIs)
Procédures qualité utilisées	Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and query system)
Suivi des participants	Yes
Modalités de suivi des participants	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Pathologie suivies	E76 - Disorders of glycosaminoglycan metabolism
Appariement avec des sources administratives	No
<b>Valorisation et accès</b>	
<b>Valorisation et accès</b>	
<b>Accès</b>	
Existence d'un document qui répertorie les variables et les modalités de codage	Yes
Charte d'accès aux données (convention de mise à disposition, format de données et délais de mise à disposition)	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: <a href="mailto:mps@radico.fr">mps@radico.fr</a>

Accès aux données agrégées      Access on specific project only

Accès aux données individuelles      Access on specific project only