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)	
Nom du responsable	Héron
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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
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-

	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 biothèque	No
Paramètres de santé étudiés	Health event/morbidity Health event/mortality Quality of life/health perception

Modalités

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

Valorisation et accès

Valorisation et accès

Accès

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: mps@radico.fr

Accès aux données agrégées	Access on specific project only
Accès aux données individuelles	Access on specific project only