RaDiCo-IDMet - National cohort on imprinting disorders and their metabolic consequences

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General

Identification

Detailed name	National cohort on imprinting disorders and their metabolic consequences
Sign or acronym	RaDiCo-IDMet
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	N° CCTIRS 16-086 / N° CPP 14129 / N° CNIL 916194 / N° MESR DC-2015-2481

General Aspects

Medical area	Biology Disability/handicap Endocrinology and metabolism Neurology Pediatrics Psychology and psychiatry Rare diseases
Study in connection with Covid-19	No
Pathology (details)	Imprinting disorders (IDs) are a group of rare genetic diseases affecting between 7,500 and 10,000 patients in the Europe. They are caused by genetic abnormalities affecting a certain type of gene, known as parental imprinting genes. Most genes in the human body are present in pairs in non-sex cells, each pair being called an allele. The genes that are subject to parental imprinting are different. They have only one allele and are subject to special regulatory mechanisms, called epigenetic modifications. Abnormalities in these mechanisms lead to abnormalities in gene expression leading to a variety of diseases whose clinical expression varies depending on whether the gene is passed on from the father or the mother. They constitute a group of 9 rare diseases (Beckwith-Wiedemann Syndrome, Prader Willi, Angelman, Silver Russel,

	Familial Precocious Puberty Limited to Boys, Pseudohypoparathyroidism, Kagami-Ogata, Temple, Transient Neonatal Type 1 Diabetes) that are probably underdiagnosed and affect growth, metabolism (lipid, carbohydrate, bone), feeding and social behavior, and reproduction.

Scientific investigator(s) (Contact)

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Organization	French National Institute for Health and Medical Research (Inserm)

Collaborations

Participation in projects, networks and consortia	Yes
Details	Rare diseases health networks FIRENDO and OSCAR / ENDO - European Reference Network (ERN)

Funding

Funding status	Public
Details	Funded by the French « Investissements d'Avenir » cohorts programme, Grant « ANR » 10-COHO-0003.

Governance of the database

Sponsor(s) or organisation(s) responsible	French National Institute for Health and Medical Research (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 main objective of this study is to describe the natural history of imprinting disorders (IDs) according to their metabolic profile.
Inclusion criteria	Patients (adults and children) affected with an ID regardless of the severity of the disease, - with a confirmed diagnosis of ID (based on molecular diagnosis) - with a signed informed consent for adults or signed informed consent of parents/guardians of minors/protected adult. 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) Elderly (65 to 79 years) Great age (80 years and more)
Population covered	Sick population
Pathology	Q87 - Other specified congenital malformation syndromes affecting multiple systems
Gender	Male Woman
Geography area	National
Detail of the geography area	National coverage through reference and competence centers focusing on these diseases.

Data collection

Dates	
Date of first collection (YYYY or MM/YYYY)	2017
Date of last collection (YYYY or MM/YYYY)	2032

Size of the database	
Size of the database (number of individuals)	[1000-10 000[individuals
Details of the number of individuals	2000 estimated

Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data

Details of collected clinical data	Medico-personal and family history, phenotype of the patient (clinical and morphological manifestation of the disease), ...
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Details of collected declarative data	Quality of life of patients and repercussions of the disease on socio-professional integration: quality of life questionnaires (SF-36, SF-10, HAQ, C-HAQ); Work Productivity and Activity Impairment questionnaire (WPAI); hyperphagia questionnaire of Dykens, and Binge-Eating Scale.
Paraclinical data (detail)	Eating behaviours
Biological data (detail)	Identified genetic/epigenetic abnormalities and their location
Presence of a biobank	Yes
Contents of biobank	Others
Details of biobank content	Stools
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
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-IDMet 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: idmet@radico.fr
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only