RaDiCo-ECYSCO - European Cystinosis Cohort

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General

Identification

Detailed name	European Cystinosis Cohort
Sign or acronym	RaDiCo-ECYSCO
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS n°15.954 / CNIL Decision n° DR-2016-383

General Aspects

Medical area	Disability/handicap Ophthalmology Pediatrics Rare diseases Urology, andrology and nephrology
Study in connection with Covid-19	No
Pathology (details)	Cystinosis: The disease is caused by mutations in the CTNS gene coding for cystinosin, a lysosomal carrier protein. The lysosomal cystine accumulation leads to cellular dysfunction in many organs. The first symptoms start at about 6 months of age with anorexia, polyuria, and failure to thrive, secondary to a Fanconi proximal renal tubulopathy. In the absence of specific therapy, end stage renal disease occurs between 6 and 12 years of age. Survival beyond this age is associated with the development of extra-renal complications in eyes, thyroid, gonads, endocrine pancreas, muscle and central nervous system
Health determinants	Genetic Lifestyle and behavior Medicine Social and psychosocial factors
Keywords	Renal Diseases, Effects of treatments, Rare diseases, Quality of life

Scientific investigator(s) (Contact)	
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Name of the director	Niaudet
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Collaborations	
Participation in projects, networks and consortia	Yes
Details	Healthcare Network for Rare Diseases Orkid / European Reference Network ERK-NET
Funding	
Funding status	Public
Details	RaDiCo received financial support from the French government managed by the National Research Agency (ANR) under the Investments for the Future Program (PIA), with reference <<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

Additional contact

Main features

Type of database

Type of database	Morbidity registers
Study databases (details)	Cohort study
Database recruitment is carried out by an intermediary	A selection of health institutions and services
Database recruitment is is made on the basis of:	Another treatment or procedure
Database recruitment is carried out as part of an interventional study	No
Additional information regarding sample selection.	Paediatric and adult patients will be mainly recruited through the network of reference, competence and recognised expert centres of rare kidney diseases. For some prevalent adult patients, recruitment will be through sites identified as in charge of regular care of cystinosis patients. During regular care follow-up visit for prevalent patient and during their first regular care visit (post-diagnosis) for incident patient, investigator will inform patients meeting the inclusion criteria about the RaDiCo-ECYSCO cohort and invite them to participate. All patients meeting criteria for inclusion and non-inclusion and willing to participate will be informed of the terms of the study during their consultation. Informed consent form and patient information sheet will be provided and explained by the investigator. Patients will be given as much time as necessary to evaluate their participation to the study. Participation in another study is not an exclusion criterion for this study as this is a follow-up of cohort type study. Also, participation in this study do not prevent participation in another study.

Database objective

Main objective	The primary objective of the RaDiCo-ECYSCO cohort is to understand the natural history and major long-term manifestations and outcomes of cystinosis in paediatric and adult cases. Secondary Objectives are to: ? Evaluate the impact of disease and treatments on patients' quality of life ? Evaluate the effect of treatment on the complications ? appraise the long-term safety of treatment and compliance Information Technology Objectives are to: ? Develop and diffuse an electronic tool of data collection from various sources linked to a database integrating a system of management and follow-up of data-management allowing collection of data for cystinosis paediatric and adult patients. ? Include data generated by patients and, where relevant, their parents and or carers. ? Expand the cohort to cover a broader European population. ? Promote the use of the RaDiCo-ECYSCO eCRF for mutualisation and harmonisation of data for cystinosis paediatric and adult patients within the expert sites. Improvement of standard care objectives are to: ? Develop comprehensive evidence based guidelines for treatments as well as for follow-up of patients who will switch from paediatric to adult status, ? Propose a system of audit against the guidelines ensuring overall care is of the highest standard as well as identifying areas of concern for actions.
Inclusion criteria	The RaDiCo-ECYSCO Cohort inclusion criteria are the following: ? Confirmed diagnosis of cystinosis (based on cystine dosage and/or presence of crystals at eye examination and/or molecular diagnosis) ? Signed informed consent Non-inclusion Criteria ? Patients not able to give their informed consent. No other non-inclusion criteria (patients with associated disease should be enrolled)
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) Great age (80 years and more)
Population covered	Sick population
Pathology	E72 - Other disorders of amino-acid metabolism
Gender	Male Woman
Geography area	International
Detail of the geography area	European study: France, Belgium, Italy, Spain, The Netherlands and Germany

Data collection

Dates

Date of first collection (YYYY or MM/YYYY)	2017
Date of last collection (YYYY or MM/YYYY)	2028

Size of the database

Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	244

Data

Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data Administrative data
Clinical data (detail)	Direct physical measures Medical registration
Details of collected clinical data	data on medical history, clinical evaluation (renal function, eyes, endocrine, gastro-intestinal

	symptoms, muscle symptoms, neurological assessment and skin lesions), laboratory analyses (including cystine dosage), cysteamine and other treatments prescription, RRT, social life, and molecular analysis of patients suffering from cystinosis. It will include all retrospective data previously collected in the CEMARA database (CNIL authorisation number: 1187326 for France; regulatory requirements for Belgium and Italy were the responsibility of the participating local site) and new data from follow-up visit of prevalent patients as well as from incident patients (new inclusions).
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Details of collected declarative data	SF-36 (adults) / SF-10 (childrens)
Biological data (detail)	Laboratory analyses: Leucocyte cystine level (expressed as nanomoles of half-cystine per milligram of protein, normal <0.15) is measured before cysteamine administration, and determined and collected at least once a year. As the WBC cystine assay is complex and highly variable between laboratories, plasma cysteamine concentration will also be collected. Sites are encouraged to record all annual additional laboratory analyses, as exploratory objective. Other laboratory analyses are performed according to current care of patients (creatininemia, kaliemia, glycaemia, Thyroid Stimulating Hormone?).
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality Quality of life/health perception
Quality of life/perceived health (detail)	SF-36 (adults) / SF-10 (childrens)
Procedures	
Data collection method	eCRF using REDCap; Cloud based, secure by design web accessible platform. Certified Health Data Hosting resource
Classifications used	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, Drug dictionary (DCIs)
Quality procedure(s) used	Continuous data management; Data Management

	Plan and Data Validation Plan. Native controls and Query system
Participant monitoring	Yes
Monitoring procedures	Monitoring by convocation of the participant Monitoring by contact with the referring doctor Monitoring by crossing with a morbidity register
Followed pathology	E72 - Other disorders of amino-acid 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)	Access Charter. Access requests to RaDiCo-ECYSCO data (rough / structured), or to analytic reports will be examined by the scientific committee following submission of a Specific Research Project (SRP) synopsis, as defined in the Resource Access Charter. Must be sent to ecysco@radico.fr
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only