RaDiCo-ECYSCO - European Cystinosis Cohort

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Général

Identification

Nom détaillé	European Cystinosis Cohort
Sigle ou acronyme	RaDiCo-ECYSCO
Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.)	CCTIRS n°15.954 / CNIL Decision n° DR-2016-383

Thématiques générales

Domaine médical	Disability/handicap Ophthalmology Pediatrics Rare diseases Urology, andrology and nephrology
Etude en lien avec la Covid-19	No
Pathologie, précisions	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
Déterminants de santé	Genetic Lifestyle and behavior Medicine Social and psychosocial factors
Mots-clés	Renal Diseases, Effects of treatments, Rare diseases, Quality of life

Responsable(s) scientifique(s)

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Collaborations

Participation à des projets, des réseaux, des consortiums	Yes
Précisions	Healthcare Network for Rare Diseases Orkid / European Reference Network ERK-NET

Financements

Financements	Public
Précisions	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>>.

Gouvernance de la base de données

Organisation(s) responsable(s) ou promoteur	French National Institute for Health and Medical Research (Inserm)
Statut de l'organisation	Secteur Public
Existence de comités	Yes

scientifique ou de pilotage	
Labellisations et évaluations de la base de données	Security audit certification of the database

Contact(s) supplémentaire(s)

Caractéristiques

Type de base de données

Type de base de données	Morbidity registers
Base de données issues d'enquêtes, précisions	Cohort study
Origine du recrutement des participants	A selection of health institutions and services
Critère de sélection des participants	Another treatment or procedure
Le recrutement dans la base de données s'effectue dans le cadre d'une étude interventionnelle	No
Informations complémentaires concernant la constitution de l'échantillon	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.

Objectif de la base de données

Objectif principal	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.
Critères d'inclusion	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)

Type de population	
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 concernée	Sick population
Pathologie	E72 - Other disorders of amino-acid metabolism
Sexe	Male Woman
Champ géographique	International
Détail du champ géographique	European study: France, Belgium, Italy, Spain, The Netherlands and Germany

Collecte

Dates

Année du premier recueil	2017
Année du dernier recueil	2028

Taille de la base de données

Taille de la base de données (en nombre d'individus)	< 500 individuals
Détail du nombre d'individus	244

Données

Activité de la base	Current data collection
Type de données recueillies	Clinical data Declarative data Paraclinical data Biological data Administrative data
Données cliniques, précisions	Direct physical measures Medical registration
Détail des données cliniques recueillies	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).
Données déclaratives, précisions	Paper self-questionnaire Internet self-questionnaire Face to face interview
Détail des données déclaratives recueillies	SF-36 (adults) / SF-10 (childrens)
Données biologiques, précisions	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?).
Existence d'une biothèque	No
Paramètres de santé étudiés	Health event/morbidity Health event/mortality Quality of life/health perception
Qualité de vie/santé perçue, précisions	SF-36 (adults) / SF-10 (childrens)
Modalités	
Mode de recueil des données	eCRF using REDCap; Cloud based, secure by design web accessible platform. Certified Health Data Hosting resource
Nomenclatures employées	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, Drug dictionary (DCIs)
Procédures qualité utilisées	Continuous data management; Data Management Plan and Data Validation Plan. Native controls 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 Monitoring by crossing with a morbidity register
Pathologie suivies	E72 - Other disorders of amino-acid 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)	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
Accès aux données agrégées	Access on specific project only
Accès aux données individuelles	Access on specific project only