RaDiCo-DCP - Primary Ciliary Dyskinesia: Identification of specific severity criteria and phenotype-genotype correlation study

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Général

Identification

Nom détaillé	Primary Ciliary Dyskinesia: Identification of specific severity criteria and phenotype-genotype correlation study
Sigle ou acronyme	RaDiCo-DCP
Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.)	CCTIRS n° 16.049Bis / CNIL Decision DR-2016-391 / CEEI n°15-259 bis

Thématiques générales

Domaine médical	Anatomy - Cytology Biology Otolaryngology or ENT Pediatrics Pneumology Rare diseases Urology, andrology and nephrology
Etude en lien avec la Covid-19	No
Pathologie, précisions	Primary Ciliary Dyskinesias (PCD) are respiratory diseases related to a constitutional abnormality of the cilia that affects the axonema, a structure common to cilia and flagella. PCDs provok upper and lower airway infections, secondary to alterations in muco-ciliary purification, which are classically manifested in childhood. These respiratory infections result in recurrent bronchopulmonary disease and rhinosinusitis, progressing to bronchial dilation and nasal polyposis, respectively. In half of the cases, there is a reverse rotation of the viscera (situs inversus), resulting in Kartagener's syndrome, defined by the triad of bronchiectasis, chronic sinusitis and situs inversus, which therefore constitutes a subgroup of PCDs. The first and most common ciliary abnormality, found in patients with PCD, corresponds to an absence of the outer dynein arms that carry the ATPase activity essential for

	ciliary movement. Many other ultrastructural abnormalities of the cilia were subsequently described in the context of PCDs. Most affected men are sterile due to immobility of the flagellum of the spermatozoa, which then present the same ultrastructural defect as that of the respiratory cilia. The incidence of PCDs in the general population is estimated to be 1/16000. The transmission of PCDs is classically autosomal recessive, with the frequency of affected individuals being much higher in populations where there is a high degree of inbreeding.
Déterminants de santé	Genetic Healthcare system and access to health care services Occupation Social and psychosocial factors

Responsable(s) scientifique(s)

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Collaborations

Participation à des projets, des réseaux, des consortiums	Yes
Précisions	Filière Maladies Rares pulmonaires RespiFil. / European Reference Network ERN-LUNG

Financements

Financements	Public
Précisions	Funded by the French « Investissements d'Avenir » cohorts programme, Grant « 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 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
Informations complémentaires concernant la constitution de l'échantillon	Depending on their symptomatology, the initial management of patients suspected of PCD is carried out in a paediatric, ENT, adult pneumology, paediatric pneumology, or reproductive biology department that undertakes etiological explorations, in particular ciliary investigations that make it possible to confirm the diagnosis of PCD. The RaDiCo-DCP cohort will consist of 300 prevalent and incident patients, both paediatric and adult, who will be followed for 5 years after their inclusion in the study.
Objectif de la base de données	
Objectif principal	The main objective is to identify in a large cohort of PCD patients, early predictors of severity in order to improve personalized patient management.
Critères d'inclusion	All prevalent patients and incidents included in the RaDiCo-DCP cohort must: ? Have a confirmed diagnosis of PCD based on at

	least one of the following diagnostic criteria: Kartagener syndrome (association of chronic sinusitis, bronchiectasis and situs inversus), and/or evidence of specific abnormalities of the ciliary ultrastructure, and/or identification of unambiguous mutations in a PCD gene. ? Have at least one annual follow-up visit in accordance with standard practice. Patients who meet the following criteria will not be included: ? Patient with an unconfirmed diagnosis of PCD; ? Patient with a concomitant progressive pathology that may interfere with the evaluation of PCD-related manifestations.
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) Elderly (65 to 79 years) Great age (80 years and more)
Population concernée	Sick population
Pathologie	Q34 - Other congenital malformations of respiratory system
Sexe	Male Woman
Champ géographique	National
Détail du champ géographique	Complete coverage of the French territory by the Reference and Competence Centers for Rare Lung Diseases
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	300 estimated
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	Demographics; General, respiratory and ENT assessments; Fertility; Genetics; Therapeutic
Données déclaratives, précisions	Paper self-questionnaire Internet self-questionnaire Face to face interview
Données paracliniques, précisions	Respiratory assessment (radiological and EFR); Paraclinical ENT assessment; NO assessment and ciliary explorations
Données biologiques, précisions	Bacteriological evaluation
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	Cohort-developed, age-appropriate quality of life questionnaires and SNOT ? 22 test reviewing 22 symptoms reflecting the pathological burden of patients with rhinological diseases.
Modalités	
Mode de recueil des données	eCRF in secure web access, secure cloud and HADS hosting
Nomenclatures employées	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, 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 Monitoring by crossing with a morbidity register
Pathologie suivies	Q34 - Other congenital malformations of respiratory system
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-DCP 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: dcp@radico.fr
Accès aux données agrégées	Access on specific project only
Accès aux données individuelles	Access on specific project only