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

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General

Identification

Detailed name	Primary Ciliary Dyskinesia: Identification of specific severity criteria and phenotype-genotype correlation study
Sign or acronym	RaDiCo-DCP
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS n° 16.049Bis / CNIL Decision DR-2016-391 / CEEI n°15-259 bis

General Aspects

Medical area	Anatomy - Cytology Biology Otolaryngology or ENT Pediatrics Pneumology Rare diseases Urology, andrology and nephrology
Study in connection with Covid-19	No
Pathology (details)	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.
Health determinants	Genetic Healthcare system and access to health care services Occupation Social and psychosocial factors

Scientific investigator(s) (Contact)

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Unit	Inserm U 955
Organization	French National Institute for Health and Medical Research (Inserm)

Collaborations

Participation in projects, networks and consortia	Yes
Details	Filière Maladies Rares pulmonaires RespiFil. / European Reference Network ERN-LUNG

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
Additional information regarding sample selection.	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.

Database objective

Main objective	The main objective is to identify in a large cohort of PCD patients, early predictors of severity in order to improve personalized patient management.
Inclusion criteria	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.
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	Q34 - Other congenital malformations of respiratory system
Gender	Male Woman
Geography area	National
Detail of the geography area	Complete coverage of the French territory by the Reference and Competence Centers for Rare Lung Diseases
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	2017
Date of last collection (YYYY or	2028

MM/YYYY)	
Size of the database	
Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	300 estimated
Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data
Clinical data (detail)	Direct physical measures Medical registration
Details of collected clinical data	Demographics; General, respiratory and ENT assessments; Fertility; Genetics; Therapeutic
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Paraclinical data (detail)	Respiratory assessment (radiological and EFR); Paraclinical ENT assessment; NO assessment and ciliary explorations
Biological data (detail)	Bacteriological evaluation
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)	Cohort-developed, age-appropriate quality of life questionnaires and SNOT ? 22 test reviewing 22 symptoms reflecting the pathological burden of patients with rhinological diseases.
Procedures	
Data collection method	eCRF in secure web access, secure cloud and HADS hosting
Classifications used	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, 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 Monitoring by crossing with a morbidity register
Followed pathology	Q34 - Other congenital malformations of respiratory system
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-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
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only