

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)

Name of the director

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Surname

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Unit

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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 MM/YYYY) 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