- French cystic fibrosis register (qualified register)

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Last update : 02/11/2014 | Version : 4 | ID : 219

General

Identification

Detailed name	French cystic fibrosis register (qualified register)
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	Autorisation CNIL n° 1202233 du 2 mars 2007

General Aspects

Medical area	Pneumology Rare diseases
Health determinants	Genetic

Scientific investigator(s) (Contact)

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Organization	Association Vaincre la Mucoviscidose
Collaborations	
Funding	
Funding status	Mixed
Details	Association Vaincre la MucoviscidoseCentre de référence de NantesINED
Governance of the database	
Sponsor(s) or organisation(s) responsible	Association Vaincre La Mucoviscidose
Organisation status	Private
Additional contact	
Main features	
Type of database	
Type of database	Morbidity registers
Additional information regarding sample selection.	Annual identification of cases since 1992 from CRCMs and local health information centers (centres relais) as well as, more rarely and since 2008, by the transplant teams' file and French Association for Screening and Prevention of Disabilities in Children (AFDPHE)
Database objective	
Main objective	1) Descriptive and analytical epidemiology ? Estimate the prevalence, incidence, geographical distribution and number of patients suffering from cystic fibrosis in France ? Estimate mortality and life expectancy at different ages ? Determine the mortality risk factors ? Describe the medical and socio-demographic characteristics of the population, especially elements concerning diagnosis, anthropometry, spirometry, microbiology, morbidity, transplants and treatment, as well as the educational and professional situations of patients. The descriptive analysis of this data is finalized by the publication of an annual report and center reports and is subject to other publications, posters, communication during congresses and

information given to families and patients via the association's magazine.

2) Evaluation of healthcare practices
The data is used to evaluate the quality of treatment, health care and diagnosis of cystic fibrosis. It also provides responses to the fundamental question on equity in these three fields, by comparing these practices to the national diagnosis and care protocol for this rare disease.
The following is also carried out:
? analysis of survival factors by introducing the treatment location and type of caring for example, in addition to the traditional variables.
? evaluation of the socioeconomic cost of cystic fibrosis by trying to match resources to the constantly changing needs.
? evaluation of neonatal screening (Cazes et al, ECFS 2005)
? development of a Program for Improving the Quality of cystic fibrosis health care by looking to the benchmarks extracted from the registry (mainly the FEV1 and BMI).

3) The registry's objectives in the fields of therapy and research
? Provision of a database for physicians and researchers.
? Search for genotype-phenotype correlations (Duguépéroux et al, 2002, 2004, 2005).
? Setup of thematic surveys, for example:
? "Pregnancy" survey: an initial stage has consisted in a retrospective study of pregnancies listed in France between 1980 and 1995. At the end of this study, a prospective registry was set up in 1996 under the observatory, involving an additional questionnaire for any pregnancy reported in the observatory's data collection. The retrospective study and first few years of prospective data collection led to a publication (Gillet et al, 2002).
? Cross-disciplinary study on "transplants" conducted in 2000 in liaison with the French Biomedicine Agency. In addition to the findings, it determined a number of important variables to collect and integrate into the registry.
? "Cepacia" survey: the Cepacia observatory was set up in 1993 by the "Vaincre la Mucoviscidose" association. Its Scientific Director is Prof. G. Chabanon. This observatory is one of the thematic observatories of the registry, with which it shares some data, including
the main characteristics for identifying patients; the Cepacia observatory networks with clinicians and

	microbiologists in healthcare centers. Its remit involves, on the one hand, conducting epidemiological surveillance of colonizations and infections by Burkholderia cepacia complex and similar bacteria in cystic fibrosis sufferers; on the other hand, building a national reference strain bank for use by the scientific community. ? "Mucoviscidose, Famille et Société" survey: organized by the French National Institute for Demographic Studies (Ined), Reference Center for Rare Diseases, "Cystic Fibrosis", of Nantes teaching hospital and the "Vaincre la Mucoviscidose" association. This aims to gather the views of the patients themselves - children, teenagers and adults - of their lifestyles and conditions: What are their family situations? What are their residential, academic and professional pathways? What aspects are linked to social recognition of the disease, and the benefits and help received? What are their possible functional limitations and/or activity restrictions? How is their social life characterized, how do they perceive the disease and how do they get on with their usual care center? What are their needs, expectations and aspirations? This information gleaned from patients will then be compared against medical data in the French Cystic Fibrosis Registry from the Cystic Fibrosis Skills and Resources Centers (CRCMs). The purpose of this study is to set up a five-year national survey of the Registry, created and managed by the "Vaincre la Mucoviscidose" association. The project comprises two stages: firstly a preliminary feasibility stage, when a pilot survey is conducted among patients followed up in the Roscoff and Strasbourg CRCMs; secondly a national survey will be conducted among all of the patients followed up by the CRCMs across France and re-launched every five years.
Inclusion criteria	The criteria for inclusion in the registry are - in addition to consent by the patient and/or parents for data to be used - those defined by the 1998 consensus conference of the Cystic Fibrosis Foundation and reported by Rosenstein and Cutting. These combine clinical and biological criteria: 1) the presence of one or more phenotypic characteristics or a family history of cystic fibrosis among siblings, or an increased rate of immunoreactive trypsin (neonatal screening) and 2) two positive sweat tests, or two mutations identified of the CFTR gene, or a pathological nasal transepithelial potential difference.

	This definition of cystic fibrosis is currently being amended in the registry following the recommendations in J Ped 2008:153:S4-S14
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)
Population covered	Sick population
Gender	Male Woman
Geography area	National
Detail of the geography area	Metropolitan France (all the 22 regions) as well as Reunion island
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	1992
Size of the database	
Size of the database (number of individuals)	[1000-10 000[individuals
Details of the number of individuals	Number of patients notified since the beginning of the recording year by year: Year : 1992 1993 1994 1995 1996 1997 1998 1999 2000 2001 2002 2003 2004 2005 2006 2007 2008 2009 Number of patients : 1641 1849 2032 2215 2406 2551 2707 3231 3377 3589 3936 4111 4544 4745 4994 5140 5357 Analysis in progress
Data	
Database activity	Current data collection
Type of data collected	Clinical data Paraclinical data Biological data

	Administrative data
Clinical data (detail)	Direct physical measures Medical registration
Paraclinical data (detail)	FEV1: volume that has been exhaled at the end of the first second of forced expirationFVC: forced vital capacity
Biological data (detail)	Cytobacteriological sputum examination. Blood gases (SaO2, PaO2, PaCO2)
Administrative data (detail)	Identification data socio-demographic data, follow-up location (center), date and department of birth, home department
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality Health care consumption and services
Care consumption (detail)	Hospitalization Medical/paramedical consultation Medicines consumption
Procedures	
Data collection method	Annual questionnaires (3 types of collection: import from the patient software, online data entry and paper questionnaire)
Participant monitoring	Yes
Details on monitoring of participants	Follow-up by the skills centers, transplant teams and local health information centers
Links to administrative sources	Yes
Linked administrative sources (detail)	Inserm - CépiDC data
Promotion and access	
Promotion	
Link to the document	http://www.registredelamuco.org
Link to the document	http://www.ecfs.eu/projects/ecfs-patient-registry/intro
Link to the document	http://www.centre-reference-muco-

	nantes.fr/index.php/epidemiologie/registre-francais-de-la-mucoviscidose

Access	
Terms of data access (charter for data provision, format of data, availability delay)	Annual report. Data demand on : http://www.vaincrelamuco.org/ewb_pages/d/donnees_registre.php
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only