- French congenital neutropenia registry

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General

Identification

Detailed name	French congenital neutropenia registry
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS : 16/05/1997 (modif juin 01), CNIL : 01-1084 (26/04/2001)

General Aspects

Medical area	Cancer research Hematology Rare diseases
Health determinants	Genetic
Keywords	leukemic transformations, infectious risk, therapy, pharmacovigilance, risk factors, genetic, diagnosis

Scientific investigator(s) (Contact)

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Organization	APHP

Collaborations

Funding

Funding status	Mixed
Details	Institut Necker Centre de référence, Chugai, Amgen, Association famille

Governance of the database

Sponsor(s) or organisation(s) responsible	Hôpital Trousseau APHP
Organisation status	Public

Additional contact

Main features

Type of database

Type of database	Morbidity registers
Additional information regarding sample selection.	The cases are identified from clinical records obtained from pediatric hematology or general and specialist pediatric departments. These centers are consulted by telephone, post or on-site monitoring. Genetics laboratories are also contacted and a work meeting is organized with them on a regular basis.

Database objective

Main objective	The initial objective at the time of its creation is to ensure pharmacovigilance of the G-CSF received by these patients. It had nevertheless been designed as a disease registry, rather than a ""post-marketing"" treatment registry. The registry's objectives have since been extended: ? Determination of risk factors of leukemic transformations in patients suffering from congenital neutropenia ? Surveillance of access to genetic and antenatal diagnosis for diseases where genetic diagnoses are available ? Surveillance of the progression of the infectious risk and therapy in patients suffering from congenital neutropenia ? Pharmacovigilance of G-CSF: Benefit-risk ratio and search for optimum therapeutic approaches. ? Evaluation of the efficacy and tolerance of bone marrow transplants in congenital neutropenia ? Classification of congenital neutropenia ? Determination of the correlation between the phenotype and genotype of patients. ? Search for new genes involved in the molecular

	bases of these diseases ? Mathematical modeling of granulopoiesis
Inclusion criteria	All of the criteria must be present: 1. Patient suffering from severe chronic neutropenia: - permanent neutropenia: absolute polynuclear rate < 0.5 109/l, measured at at least three intervals over the three months prior to the study or absolute polynuclear rate < 1 109/l, measured at at least three intervals over the three months prior to the study and presence of either a severe infection (septicemia- cellulitis- bacterial or mycotic pneumonia) or chronic gingivo-stomatitis. - intermittent neutropenia: After a surveillance period of at least six weeks, the neutrophilia rate must be less than 0.5 109 /l in at least three blood counts. 2. Myelogram performed and cytological aspect compatible with the diagnosis (in the opinion of the registry's contact cytologist) 3. Subject aged over three months 4. Patients suffering from glycogen storage disease Ib, Shwachman Diamond syndrome or WHIM syndrome are all included 5. Consent by the patient and/or his/her parents EXCLUSION CRITERIA (except glycogen storage disease Ib, Shwachman Diamond syndrome, WHIM syndrome or large granular lymphocytosis LGL): - all types of neutropenia caused by drugs - all medical histories with chemotherapy - medullary aplasia, irrespective of its etiology (idiopathic, Fanconi syndrome, etc.) - anemia < 8gr/dl or thrombopenia (except inflammatory or iron-deficiency anemia, glycogen storage disease Ib and Shwachman Diamond syndrome). - progressive malignant pathology or medical history of malignant pathology - neutropenia linked to HIV infection - macrophage activation syndrome - initial myelodysplasia
Population type	
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 covered	Sick population
Gender	Male Woman
Geography area	National
Detail of the geography area	All of french metropolitan territory
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	1995
Size of the database	
Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	503 (in 12/2009)
Data	
Database activity	Current data collection
Type of data collected	Clinical data Paraclinical data Biological data
Clinical data (detail)	Direct physical measures
Paraclinical data (detail)	bone x-ray or pancreatic imaging or brain MRI
Biological data (detail)	hematology
Presence of a biobank	Yes
Contents of biobank	DNA
Details of biobank content	DNA bank
Health parameters studied	Health event/morbidity Health event/mortality Health care consumption and services

Care consumption (detail)	Medical/paramedical consultation
Procedures	
Data collection method	The data is collected for prevalent and incident cases from clinical records obtained from pediatric hematology or general and specialist pediatric departments.
Classifications used	D70.0 D72.0 D72.9
Participant monitoring	Yes
Details on monitoring of participants	Follow-up concerns progression of the disease. Data is collected on the following themes: hematological parameters, severe infections, pregnancy, therapy, degree of social integration (in the workplace and at school)
Links to administrative sources	No
Promotion and access	
Promotion	
Link to the document	http://tinyurl.com/PUBMED-SNCregistry
Description	Liste des publications dans Pubmed
Link to the document	http://tinyurl.com/HAL-SNCregistry
Description	Liste des publications dans HAL
Access	
Terms of data access (charter for data provision, format of data, availability delay)	Web
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only