

- 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 "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

Inclusion criteria

All of the criteria must be present:

1. Patient suffering from severe chronic neutropenia:
 - permanent neutropenia: absolute polynuclear rate $< 0.5 \cdot 10^9/l$, measured at at least three intervals over the three months prior to the study or absolute polynuclear rate $< 1 \cdot 10^9/l$, 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 $0.5 \cdot 10^9 /l$ 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