- Registre des patients thalassémiques en France (Registre qualifié)

Head :Badens Catherine, Laboratoire de Génétique Moléculaire, Hôpital d'enfants de la Timone

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General

Identification

Detailed name	Registre des patients thalassémiques en France (Registre qualifié)
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CNIL 04-1396 (30/11/2004)

General Aspects

Medical area	Hematology Internal medicine Pediatrics Rare diseases
Health determinants	Genetic
Keywords	CohortClinical research

Scientific investigator(s) (Contact)

Name of the director	Badens
Surname	Catherine
Address	Laboratoire de Génétique Moléculaire, Hôpital d'enfants de la Timone, 13385 Marseille cedex 5
Phone	+ 33 (0)4 91 38 77 87
Email	catherine.badens@ap-hm.fr
Unit	Laboratoire de Génétique Moléculaire, Hôpital d'enfants de la Timone
Organization	Assistance Publique-Hôpitaux de

Collaborations

Funding

Funding status	Public
Details	Le centre de référence Thalassémies, une subvention INVS/INSERM pour 2009, 2010, 2011 et 2012 dans le cadre de la labellisation des registres maladies rares.

Governance of the database

Sponsor(s) or organisation(s) responsible	Assistance Publique-Hôpitaux de Marseille
Organisation status	Public

Additional contact

Main features

Type of database

Type of database	Morbidity registers
Additional information regarding sample selection.	Cases are identified from the following structures-sources: - Pediatric, pediatric hematology, hematology and internal medicine departments -Hematopoietic stem cell transplant departments -The 4 neonatal screening laboratories for sickle-cell anemia - French blood centers -Accredited molecular genetics laboratories for diagnosing hemoglobin anomalies Cases are validated by Dr Thuret before entry, with the attending physician being contacted when necessary.

Database objective

Main objective	-Obtain national epidemiological data on the disease, particularly its morbidity and, over the longer term, its mortality, which reflect the quality of overall patient care -Monitor treatment trends over time - especially the increasing use of orally administered active chelators and the spread of new techniques for evaluating tissue overload by heart and liver MRIs. -Compare conventional treatment results combining transfusions and iron chelation with those obtained from hematopoietic stem cell transplants. -Conduct more basic research projects

Inclusion criteria	Beta-thalassemias (TM) major and intermediate (TI), including HbE/thalassemia composite hereterozygosity and thalassemia patients who have had a HSC transplant. Diagnosis of a severe form of beta-thalassemia is formally established on the basis of data from a biochemical study of hemoglobin with no real differential diagnosis. Distinguishing between thalassemia major and intermediate requires the combination of 3 consensual criteria: ? for TM: Severe anemia indicating the setup of a systematic transfusion routine (at least 8 transfusions/year) before the age of 4. ? for TI: Moderate or medium intensity anemia that have no or only occasional need for transfusion. The date and circumstances of the diagnosis are asked for upon inclusion and the characteristics of the transfusion treatment upon inclusion and when each follow-up sheet is completed. Identification of the beta-thalassemia mutations or type of beta 0 or beta + mutation are available in most cases, allowing for a complementary approach - even if the TM or TI diagnosis retains a clinical definition. HbE/thalassemia composite hereterozygosity is of biological definition (biochemical or molecular) and can lead to an intermediate or major form of thalassemia: as a result they are subject to the same classification according to the same clinical criteria (age at diagnosis and transfusion needs).
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	France
Data collection	

Dates	
Date of first collection (YYYY or MM/YYYY)	2005
Size of the database	
Size of the database (number of individuals)	[500-1000[individuals
Details of the number of individuals	287 (2008)479 (2010)515 (2012)
Data	
Database activity	Current data collection
Type of data collected	Clinical data Biological data
Clinical data (detail)	Direct physical measures
Biological data (detail)	examination of the medical record
Presence of a biobank	Yes
Contents of biobank	Cell lines DNA
Details of biobank content	We offer to store a DNA sample, and possibly a cell line for each patient included in the registry, at the biological resource center (CRB) in Marseilles. There are currently some one hundred samples stored in this collection.
Health parameters studied	Health event/morbidity Health event/mortality Others
Other (detail)	Professional activities or schooling
Procedures	
Data collection method	Inclusion or follow-up sheet posted by standard delivery to Marseilles. The data is then entered into the database before being validated.
Participant monitoring	Yes
Details on monitoring of participants	A follow-up sheet comprising the same items as the inclusion sheet, along with noting down of the patient's death when it arises and cause of death, is

	sent to clinicians every 18 months.
Links to administrative sources	No
Promotion and access	
Promotion	
Link to the document	http://www.chu-lyon.fr/web/2652
Link to the document	http://www.haematologica.org/cgi/content/full/95/5/724
Link to the document	http://tinyurl.com/PUBMED-RPT
Description	Liste des publications dans Pubmed
Access	
Terms of data access (charter for data provision, format of data, availability delay)	Results access procedure: Publications and scientific communications at conferences Data access procedure for external teams: a request must be submitted to the registry manager (C. Badens) and to the 2 coordinators of the Thalassemia reference center (I. Thuret and C. Pondarré). Acceptance criteria currently being defined.
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only