DEFI-ALPHA - Cohort of Children with Alpha 1 Antitrypsin Deficiency.

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General

Identification

Detailed name	Cohort of Children with Alpha 1 Antitrypsin Deficiency.
Sign or acronym	DEFI-ALPHA
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS n°10.181 (08/04/2010), CNIL n°910279 (DR-2010-328, 29/10/2010)

General Aspects

Medical area	Gastroenterology et hepatology
Health determinants	Genetic
Keywords	occurrence of complications, ultrasound/Doppler ultrasound, FibroScan, fibrosis test, endoscopy and oesophageal video capsule, platelets, transaminase and gamma-GT, Health episodes, factors, liver transplantation

Scientific investigator(s) (Contact)

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Organization	HCL - HOPITAL FEMME-MERE-ENFANTDE LYON

Collaborations

Participation in projects, networks and consortia	Yes

Funding

Funding status	Mixed
Details	Laboratoires LFB, PHRC

Governance of the database

Sponsor(s) or organisation(s) responsible	CHU Lyon (L. Restier)
Organisation status	Public

Additional contact

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Main features

Type of database

Type of database	Study databases
Study databases (details)	Cohort study
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.	Retrospective Other bodies active in creating this cohort: CHU and CHG, parents' association (Association ADAAT ALPHA 1 France, French

	association for patients and patients' families with alpha-1 antitrypsin deficiency)
Database objective	
Main objective	General objective: To investigate prognostic hepatic factors in A1AT deficient children. Investigation of factors associated with onset of complications: portal hypertension and its complications, severe liver failure, liver transplantation, abnormalities during respiratory function exploration. Secondary objectives: - To organise an active homogenised cohort follow-up. - To ensure that new cases are recorded. - To create a reference network in order to homogenise treatment.
Inclusion criteria	Children with DA1AT born after 1989, regardless of phenotype. DA1AT is defined as an alpha-1 antitrypsin deficiency lower than 1,1 g/l, and type ZZ or SZ MZ, MS, SS, detected by participating centres (service follow-up, or detected by corresponding neonatal services, family surveys, patient associations or dosage laboratories in the geographical area of participating services).
Population type	
Age	Early childhood (2 to 5 years) Childhood (6 to 13 years) Adolescence (13 to 18 years)
Population covered	Sick population
Pathology	XI - Diseases of the digestive system
Gender	Male Woman
Geography area	National
Detail of the geography area	Multicentric cohort throughout France (15 centres)
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	09/2008
Size of the database	
Size of the database (number of individuals)	< 500 individuals

Details of the number of individuals	180

Data

Database activity	Current data collection
Type of data collected	Clinical data Paraclinical data Biological data
Clinical data (detail)	Direct physical measures Medical registration
Paraclinical data (detail)	Biochemistry and genetics, ultrasound, FibroScan, and medical imaging
Biological data (detail)	Clinical records
Presence of a biobank	Yes
Contents of biobank	Serum
Details of biobank content	Serum bank
Health parameters studied	Health event/morbidity Health event/mortality

Procedures

Data collection method	Interview: Direct input Clinical examination: Direct input Biological analysis: Direct input
Participant monitoring	Yes
Monitoring procedures	Monitoring by crossing with a medical-administrative database
Details on monitoring of participants	Until 18 years of age
Followed pathology	XI - Diseases of the digestive system
Links to administrative sources	No

Promotion and access

Promotion

Link to the document	Ruiz_et_al-2019-Liver_International.pdf

Description	Pathologies hépatiques en rapport avec le déficit en alpha1-antitrypsine dans une cohorte d'enfants en France
Access	
Terms of data access (charter for data provision, format of data, availability delay)	To be decided if data may be used by academic teams Eric Chevet, INSERM U1053, Université Bordeaux Ségalen Access conditions: for European cohort collaboration (Dino Hadzic, nedim.hadzic@kcl.ac.uk), for an adult cohort with DA1AT (Gabriel Thabut : g.thabut@bch.ap-hop-paris.fr) To be decided if data may be used by industrial teams Access for potentially interested LFB laboratories
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only