RaDiCo-COLPAC - RaDiCo-COLPAC - National Cohort on the Epidemiology, Clinical and Genetic Heterogeneity of "Low Phospholipid?Associated Cholelithiasis" (LPAC) Syndrome

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General

Identification

Detailed name	RaDiCo-COLPAC - National Cohort on the Epidemiology, Clinical and Genetic Heterogeneity of "Low Phospholipid?Associated Cholelithiasis" (LPAC) Syndrome
Sign or acronym	RaDiCo-COLPAC
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	N° CCTIRS 16-361 / N° CPP 14238ND / N° MESR DC-2015-2480 / MR-001

General Aspects

Medical area	Gastroenterology et hepatology Pediatrics Radiology and medical imaging Rare diseases
Study in connection with Covid-19	No
Pathology (details)	LPAC syndrome is a rare and symptomatic form of intrahepatic gallstones of cholesterol in young subjects (< 40 years) of genetic origin, recurrent characteristically after cholecystectomy and often responding favorably to treatment with ursodeoxycholic acid. LPAC syndrome is associated with an abnormally low concentration of phospholipids in the bile. It preferentially affects women between the ages of 20 and 40 and is often associated with a history of gestational cholestasis. A family history of symptomatic gallstones is present in 40% of cases and a germline mutation (most often of the missense and heterozygous type) of the ABCB4 gene, which encodes MDR3, the biliary transporter of phospholipids, is found in 30% to 50% of cases. Apart from the ABCB4 gene, there is no other predisposition gene currently identified. Its clinical expression is variable, ranging from the

	microlithiasis form responding well to the ursodeoxycholic acid to the calculus of the bile ducts responsible for recurrent angiocholitis. Some rarer forms can be complicated by cirrhosis or cholangiocarcinoma. LPAC syndrome is therefore a rare disease, clinically and genetically heterogeneous, whose course and prognosis are not predictable and whose prevalence in the general population remains unknown.
Scientific investigator(s) (Contact)	
Name of the director	Corpechot
Surname	Christophe
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Unit	UMR_S938
Organization	Institut National de la Santé et de la Recherche Médicale (Inserm)
Collaborations	
Funding	
Funding status	Public
Details	The RaDiCo-COLPAC cohort is funded by the French « Investissements d'Avenir » cohorts programme, Grant « ANR » 10-COHO-0003. This study also received a grant from the "COMAD" call for projects launched by the SNFGE in 2016.
Governance of the database	
Sponsor(s) or organisation(s) responsible	Institut National de la Santé et de la Recherche Médicale (Inserm)
Organisation status	Public
Presence of scientific or steering committees	Yes

Labelling and database evaluation	Security audit certification of the database. Data management and continuous quality control of data.

Additional contact

Main features

Type of database

Type of database	Morbidity registers
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

Database objective

Main objective	The main objective is to describe the different clinical, biological and radiological manifestations of LPAC syndrome defined according to current diagnostic criteria or according to extended criteria to any recurrent symptomatic gallstone disease and to delineate the different possible evolutions.
Inclusion criteria	Inclusion Criteria: Any patient over 13 years of age, prevalent or incident, meeting the usual diagnostic criteria (Patient Category 1: symptomatic gallstones with at least 2 out of 3 LPAC syndrome criteria, see below) or extensive (Patient Category 2: symptomatic gallstones with only one out of 3 LPAC syndrome criteria, see below) diagnostic criteria for LPAC syndrome. Diagnostic criteria for LPAC syndrome (symptomatic patients): 1) First symptoms before the age of 40 2) Radiological images consistent with the existence of intrahepatic lithiasis (stones, sludge, hyper-echoic foci, "comet tails") 3) Recurrence of symptoms after cholecystectomy Exclusion Criteria: Patients who have undergone liver transplantation.

Population type

Age	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
Pathology	K83 - Other diseases of biliary tract
Gender	Male Woman
Geography area	National
Data collection	
Dates	
Date of first collection (YYYY or MM/YYYY)	2017
Date of last collection (YYYY or MM/YYYY)	2028
Size of the database	
Size of the database (number of individuals)	[500-1000[individuals
Details of the number of individuals	550 to 650 patients estimated
Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data
Clinical data (detail)	Direct physical measures Medical registration
Details of collected clinical data	Demographic data, diagnostic characteristics, medical history related to the pathology, comorbidities not related to the pathology, family history, symptoms, LPAC specific treatments and other, surgical or endoscopic interventions, ...

Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Details of collected declarative data	Quality of life (SF-10 for minors / SF-36 for adults) and pain assessment
Paraclinical data (detail)	Imaging data (liver ultrasound, liver scan, cholangiography by MRI, biliopancreatic endoscopic ultrasound, endoscopic retrograde cholangiography), ...
Biological data (detail)	Haematological and biochemical results, biliary analyses, ...
Presence of a biobank	Yes
Contents of biobank	DNA
Details of biobank content	The project includes the creation of a genomic DNA bank to search for new susceptibility genes (or modulators) for LPAC syndrome.
Health parameters studied	Health event/morbidity Health event/mortality Quality of life/health perception
Procedures	
Data collection method	eCRF in secure web access, secure cloud and HADS hosting
Quality procedure(s) used	Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and query system)
Participant monitoring	Yes
Monitoring procedures	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Links to administrative sources	No
Promotion and access	
Promotion	
Access	
Presence of document that lists variables and coding procedures	Yes

Terms of data access (charter for data provision, format of data, availability delay)	Requests for access to RaDiCo-COLPAC data (aggregated or individual) will be considered by the Scientific Committee following the submission of a summary of a specific research project, as defined in the Charter of access to resources. Requests should be sent to: colpac@radico.fr
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only