

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

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Général	
Identification	
Nom détaillé	RaDiCo-COLPAC - National Cohort on the Epidemiology, Clinical and Genetic Heterogeneity of "Low Phospholipid?Associated Cholelithiasis" (LPAC) Syndrome
Sigle ou acronyme	RaDiCo-COLPAC
Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.)	N° CCTIRS 16-361 / N° CPP 14238ND / N° MESR DC-2015-2480 / MR-001
Thématiques générales	
Domaine médical	Gastroenterology et hepatology Pediatrics Radiology and medical imaging Rare diseases
Etude en lien avec la Covid-19	No
Pathologie, précisions	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.

Responsable(s) scientifique(s)

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Laboratoire	UMR_S938
Organisme	Institut National de la Santé et de la Recherche Médicale (Inserm)

Collaborations

Financements

Financements	Public
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Précisions	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.
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Gouvernance de la base de données

Organisation(s) responsable(s) ou promoteur	Institut National de la Santé et de la Recherche Médicale (Inserm)
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Statut de l'organisation	Secteur Public
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Existence de comités scientifique ou de pilotage	Yes
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Labellisations et évaluations de la base de données	Security audit certification of the database. Data management and continuous quality control of data.
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Contact(s) supplémentaire(s)

Caractéristiques

Type de base de données

Type de base de données

Morbidity registers

Origine du recrutement des participants

A selection of health institutions and services

Le recrutement dans la base de données s'effectue dans le cadre d'une étude interventionnelle

No

Objectif de la base de données

Objectif principal

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.

Critères d'inclusion

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.

Type de population

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 concernée Sick population

Pathologie K83 - Other diseases of biliary tract

Sexe Male
Woman

Champ géographique National

Collecte

Dates

Année du premier recueil 2017

Année du dernier recueil 2028

Taille de la base de données

Taille de la base de données (en nombre d'individus) [500-1000[individuals

Détail du nombre d'individus 550 to 650 patients estimated

Données

Activité de la base Current data collection

Type de données recueillies Clinical data
Declarative data
Paraclinical data
Biological data

Données cliniques, précisions Direct physical measures
Medical registration

Détail des données cliniques recueillies 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,

Données déclaratives, précisions Paper self-questionnaire
Internet self-questionnaire
Face to face interview

Détail des données déclaratives recueillies	Quality of life (SF-10 for minors / SF-36 for adults) and pain assessment
Données paracliniques, précisions	Imaging data (liver ultrasound, liver scan, cholangiography by MRI, biliopancreatic endoscopic ultrasound, endoscopic retrograde cholangiography),
Données biologiques, précisions	Haematological and biochemical results, biliary analyses,
Existence d'une bibliothèque	Yes
Contenu de la bibliothèque	DNA
Détail des éléments conservés	The project includes the creation of a genomic DNA bank to search for new susceptibility genes (or modulators) for LPAC syndrome.
Paramètres de santé étudiés	Health event/morbidity Health event/mortality Quality of life/health perception
Modalités	
Mode de recueil des données	eCRF in secure web access, secure cloud and HADS hosting
Procédures qualité utilisées	Data Management Plan and Data Validation Plan. Continuous data management (automatic control rules and query system)
Suivi des participants	Yes
Modalités de suivi des participants	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Appariement avec des sources administratives	No
Valorisation et accès	
Valorisation et accès	
Accès	
Existence d'un document qui répertorie les variables et les modalités de codage	Yes
Charte d'accès aux données (convention de mise à disposition, format de données)	Requests for access to RaDiCo-COLPAC data (aggregated or individual) will be considered by the Scientific Committee following the submission of a

et délais de mise à disposition)	summary of a specific research project, as defined in the Charter of access to resources. Requests should be sent to: colpac@radico.fr
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Accès aux données agrégées	Access on specific project only
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Accès aux données individuelles	Access on specific project only
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