

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

Head :Corpechot Christophe, UMR_S938

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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)

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| Name of the director | Corpechot |
| Surname | Christophe |
| Address | Centre de référence des Maladies Inflammatoires des Voies Biliaires Hôpital Saint-Antoine 184 rue du faubourg Saint-Antoine 75571 Paris cedex 12 FRANCE |
| Phone | + 33 (0)1 49 28 28 36 |
| Unit | UMR_S938 |
| Organization | Institut National de la Santé et de la Recherche Médicale (Inserm) |

Collaborations

Funding

| | |
|----------------|--------|
| Funding status | Public |
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| 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. |
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Governance of the database

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| Sponsor(s) or organisation(s) responsible | Institut National de la Santé et de la Recherche Médicale (Inserm) |
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| Organisation status | Public |
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| Presence of scientific or steering committees | Yes |
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| Labelling and database evaluation | Security audit certification of the database. Data management and continuous quality control of data. |
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Additional contact

Main features

Type of database

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| Type of database | Morbidity registers |
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| Database recruitment is carried out by an intermediary | A selection of health institutions and services |
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| Database recruitment is carried out as part of an interventional study | No |
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Database objective

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| 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. |
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| Inclusion criteria | <p>Inclusion Criteria:</p> <p>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.</p> <p>Diagnostic criteria for LPAC syndrome (symptomatic patients):</p> <ol style="list-style-type: none">1) First symptoms before the age of 402) Radiological images consistent with the existence of intrahepatic lithiasis (stones, sludge, hyper-echoic foci, "comet tails")3) Recurrence of symptoms after cholecystectomy <p>Exclusion Criteria:</p> <p>Patients who have undergone liver transplantation.</p> |
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Population type

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| 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, |

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| 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 |

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| 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 |