The identification of new polycystic liver disease genes.

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The identification of a new gene(s) that causes autosomal dominant polycystic liver disease.

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Hepatobiliary disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON50103

Source
ToetsingOnline

Brief title
Polycystic liver disease genes

Condition

- Hepatobiliary disorders congenital
- Hepatic and hepatobiliary disorders

Synonym
Autosomal dominant polycystic liver disease

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: ADPLD, Autosomal dominant polycystic liver disease, PLD, Polycystic liver disease

Outcome measures

Primary outcome

Identification of the pathogenic variant in PLD related genes in the patient.

Secondary outcome

Not applicable

Study description

Background summary

Polycystic liver disease (PLD) is a disorder which is characterized by the development of numerous fluid-filled cysts in the liver. PLD is a phenotype in two heritable diseases: autosomal dominant polycystic liver disease and autosomal dominant kidney disease (ADPLD and ADPKD). In ADPLD patients, only cysts are present in the liver. In ADPKD patients, numerous cysts are present in both the liver and the kidney.

ADPKD is nearly always cause by a pathogenic variant in the genes PKD1 (78%), PKD2 (15%) or GANAB (0.3%). However, ADPLD does not seem to be caused by one or a few genes. Currently, there are already 6 genes known to be the cause of ADPLD development: PRKCSH (~20%), SEC63 (~15%), ALG8 (~3%), GANAB (~2%), SEC61B (~1%), and LRP5 (~1%). However, about 60% of the ADPLD patients do not have a PLD-causing variant in one of these genes.

Study objective

The identification of a new gene(s) that causes autosomal dominant polycystic liver disease.

Study design

Dutch ADPLD patients will be selected for our ADPLD cohort based on the inclusion and exclusion criteria. From the selected patients that are willing to participate, 1 vial of 10 mL blood will be taken. This blood will be used to screen the patient for possibly pathogenic variants in genes that could be PLD-associated by modern genomic techniques like whole exome sequencing. In the laboratory, the pathogenicity of these variants will be tested by different functional analyses.

Study burden and risks

Incidental findings (knowledge of being at risk for the development of other diseases), however the discovery of these incidental findings is extremely small.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Patient has polycystic liver disease (current definition: >10 liver cysts)
Patient has visited our department in regards to polycystic liver disease
Individual is a (healthy) family member of an included patient

Exclusion criteria

Presence of ADPKD (autosomal dominant polycystic kidney disease)
Patient already has a genetic diagnosis
Patients without interest/with objection

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	06-08-2020
Enrollment:	400
Type:	Actual

Ethics review

Approved WMO

Date:	29-07-2020
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL73385.091.20