

# DEFINING THE GENETIC DRIVERS OF ADULT-ONSET CHOLESTATIC LIVER DISEASE

NCT07317193

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<b>Status</b>	RECRUITING
<b>Phase</b>	Not Applicable
<b>Sponsor</b>	Fondazione IRCCS Ca' Granda, Ospedale Maggiore Policlinico
<b>Enrollment</b>	60 participants

## Key Eligibility Criteria

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### Inclusion (12)

- Cases:
- Adults, aged > 18 years with:
  - persistent or intermittent elevations in serum alkaline phosphatase (ALP) or gamma-glutamyltransferase (GGT) for at least six months not explained following standard diagnostic assessment adhering to the guidelines of the European Association for the Study of the Liver (EASL), or with a positive family history of unexplained cholestasis or hepato-biliary cancer, negative to previous genetic tests (targeted panel for PFIC genes or WES);
  - primary sclerosing cholangitis (PSC) with unusual features: small-duct PSC, non-typical radiological findings according to radiological guidelines on PSC, absence of concomitant inflammatory bowel disease, negative to previous genetic tests (targeted panel for PFIC genes or WES) or who didn't perform previous genetic test;
  - primary biliary cholangitis (PBC) without specific anti-mitochondrial antibodies, negative to previous genetic tests (targeted panel for PFIC genes or WES) or who didn't perform previous genetic test.

... and 7 more (see full listing online)

### Exclusion (1)

- Cases:

## Locations (1 total)

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Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico - Istituto di Ricovero e Cura a Carattere Scientifico di natura pubblica, Milan, Milano, Italy