NHS
South East
Genomic Laboratory Hub

GASTROHEPATOLOGY GENETICS REQUEST FORM

Liver Molecular Genetics, Institute of Liver Studies,
3rd Floor, Cheyne Wing, King's College Hospital,
Denmark Hill, London, SE5 9RS

T: 020 3299 4625/2253
kch-tr.kchlmgadmin@nhs.net

All fields are mandatory. Illegible, unclear or incomplete forms will result in delays or rejection.

CONSENT STATEMENT: It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future diagnostic testing. In signing this form the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the donor's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. If the patient does not wish information to be shared please write this clearly in the clinical summary box.

PATIENT DEMOGRAPHICS	
First name:	
Last name:	
DOB:	**Gender:** Male ☐ Female ☐ Other ☐
NHS number:	
Hospital no:	**Family ref no:**
Postcode:	**Life status:** Alive ☐ Deceased ☐
Non-NHSE funded (please attach invoicing details): ☐	

PATIENT ETHNICITY	
White:	British ☐ Irish ☐ Any Other White Background ☐
Mixed:	White And Black Caribbean ☐ White And Black African ☐ White And Asian ☐ Any Other Mixed Background ☐
Asian or Asian British:	Indian ☐ Pakistani ☐ Bangladeshi ☐ Any Other Asian Background ☐
Black or Black British:	Caribbean ☐ African ☐ Any Other Black Background ☐
Other Ethnic Groups:	Chinese ☐ Any Other Ethnic Group ☐ (please specify: _____)
Not stated ☐	**Not Known** ☐

CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILTY

Please demonstrate how your patient meets the eligibility criteria for this test. Interpretation of results depends on the quality of clinical information provided. Find the eligibility criteria here: https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-V7.1-OFFICIAL.pdf

Is patient pregnant? Y/N If yes how many weeks gestation?_____

Have other members of this family had gene testing? Y/N
Please provide details:

For familial cases, please include a pedigree with the patient clearly marked:

Affected ☐ Unaffected ☐ | Age of onset: | Patients to be tested: Patient only ☐ Patient and both parents ☐ Other ☐

CLINICALLY URGENT? ☐

CLINICIAN DETAILS	
Requesting clinician / consultant Name: Hospital & department: NHS email: Phone:	Responsible clinician / consultant *(if different)* Name: Hospital & department: NHS email: Phone:

SAMPLE TYPE:

Blood EDTA ☐ for DNA or gene tests

CVS ☐ **Amnio** ☐ **Fetal blood** ☐ **POC** ☐

Other (please state)

Date of collection:

Time of collection:

For Departmental Use Only:

TEST REQUEST:

Gene test: _____

Test directory clinical indication and code (reason for testing) https://www.england.nhs.uk/publication/national-genomic-test-directories/

R171.1 Cholestasis (NGS Panel) ☐

R172.1 Wilson Disease (*ATP7B* single gene sequencing) ☐

R173.1 Polycystic Liver Disease (NGS Panel) ☐

R175.1 Pancreatitis (NGS Panel) ☐

R175.2 Pancreatitis (*CFTR* single gene sequencing) ☐

R177.1 Hirschsprung (*RET* single gene sequencing) ☐

R331.1 Intestinal Failure (NGS Panel) ☐

Crigler-Najjar Syndrome (*UGT1A1* full gene sequencing performed as part of Large Panel R171.1) ☐

If the clinical indication and code are not provided, a panel will be applied based on the clinical information provided.

DNA storage only ☐

Other (please specify) _____

Note: Please ensure the latest version of this request form is used, found on our website: www.southeastgenomics.nhs.uk

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Patient first name:	Patient last name:	DOB:	NHS no:

HPO terms phenotypes and presence in this individual – please tick
Please confirm the HPO terms that have been assessed, and select whether they are present or absent

R171 Cholestasis	Present	Absent
Bile duct proliferation		
Cholestasis		
Cirrhosis		
Conjugated hyperbilirubinemia		
Diarrhea		
Elevated gamma-glutamyltransferase level		
Elevated hepatic transaminase		
Failure to thrive		
Failure to thrive in infancy		
Hearing impairment		
Hepatic failure		
Hepatic steatosis		
Hepatocellular carcinoma		
Hepatomegaly		
Heterogeneous		
Hyperbilirubinemia		
Hypercholesterolemia		
Hypoglycemia		
Increased LDL cholesterol concentration		
Increased serum bile acid concentration		
Intermittent jaundice		
Intrahepatic cholestasis		
Jaundice		
Malabsorption		
Normal/low gamma-glutamyltransferase level		
Oedema		
Pancreatitis		
Prolonged prothrombin time		
Pruritus		
Short stature		
Splenomegaly		

R172 Wilson	Present	Absent
Cirrhosis		
Decreased serum ceruloplasmin		
Hemolytic anemia		
Hepatic failure		
Hepatomegaly		
High nonceruloplasmin-bound serum copper		
Kayser-Fleischer ring		

R173 Polycystic Liver Disease	Present	Absent
Abdominal pain		
Congenital hepatic fibrosis		
Gastrointestinal hemorrhage		
Hepatomegaly		
Increased total bilirubin		
Malformation of the hepatic ductal plate		
Multiple renal cysts		
Polycystic liver disease		
Portal fibrosis		

R175 Pancreatitis	Present	Absent
Abdominal pain		
Acute pancreatitis		
Chronic pancreatitis		
Diabetes mellitus		
Exocrine pancreatic insufficiency		
Pancreatic calcification		
Pancreatic pseudocyst		
Recurrent pancreatitis		
Steatorrhea		

R177 Hirschsprung	Present	Absent
Constipation		
Abdominal pain		
Diarrhea		
Aganglionic megacolon		
Intestinal obstruction		
Aganglionosis of the small intestine		
Total intestinal aganglionosis		

R331 Intestinal Failure	Present	Absent
Abdominal distention		
Abdominal pain		
Abnormality of the pancreas		
Alkalosis		
Cholestasis		
Cirrhosis		
Colitis		
Dehydration		
Diarrhea		
Failure to thrive		
Growth delay		
Hepatic failure		
Hepatic fibrosis		
Hepatosplenomegaly		
Hypochloremia		
Hypokalemia		
Hyponatremia		
Immune dysregulation		
Immunodeficiency		
Inflammation of the large intestine		
Intractable diarrhea		
Intrauterine growth retardation		
Metabolic acidosis		
Polyhydramnios		
Premature birth		
Recurrent fever		
Secretory diarrhea		
Short stature		
Small for gestational age		
Trichorrhexis nodosa		

Crigler-Najjar Syndrome	Present	Absent
Encephalopathy		
Jaundice		
Kernicterus		
Unconjugated hyperbilirubinemia		

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