

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.**

<b>PATIENT DEMOGRAPHICS</b>		<b>PATIENT ETHNICITY</b>	
First name:		<b>White:</b>	British <input type="checkbox"/> Irish <input type="checkbox"/> Any Other White Background <input type="checkbox"/>
Last name:		<b>Mixed:</b>	White And Black Caribbean <input type="checkbox"/> White And Black African <input type="checkbox"/> White And Asian <input type="checkbox"/> Any Other Mixed Background <input type="checkbox"/>
DOB:	Gender: Male <input type="checkbox"/> Female <input type="checkbox"/> Other <input type="checkbox"/>	<b>Asian or Asian British:</b>	Indian <input type="checkbox"/> Pakistani <input type="checkbox"/> Bangladeshi <input type="checkbox"/> Any Other Asian Background <input type="checkbox"/>
NHS number:		<b>Black or Black British:</b>	Caribbean <input type="checkbox"/> African <input type="checkbox"/> Any Other Black Background <input type="checkbox"/>
Hospital no:	Family ref no:	<b>Other Ethnic Groups:</b>	Chinese <input type="checkbox"/> Any Other Ethnic Group <input type="checkbox"/> (please specify: _____)
Postcode:	Life status: Alive <input type="checkbox"/> Deceased <input type="checkbox"/>	<b>Not stated</b> <input type="checkbox"/>	<b>Not Known</b> <input type="checkbox"/>
Non-NHSE funded (please attach invoicing details): <input type="checkbox"/>			

<b>CLINICAL INFORMATION AND FAMILY HISTORY</b>	
Please give as much clinical and genetic information as possible. Interpretation of results depends on the quality of clinical information provided. Please use HPO terms ( <a href="https://hpo.jax.org/app/">https://hpo.jax.org/app/</a> ) when possible, see reverse.	
<p>Have other members of this family had gene testing? Y/N Please provide details:</p> <p>For familial cases, please include a pedigree with the patient clearly marked:</p>	
<p>Is patient pregnant? Y/N      If yes how many weeks gestation? _____</p>	
Affected <input type="checkbox"/> Unaffected <input type="checkbox"/>	Age of onset:      Patients to be tested: Patient only <input type="checkbox"/> Patient and both parents <input type="checkbox"/> Other <input type="checkbox"/>
<b>CLINICALLY URGENT?</b> <input type="checkbox"/>	

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

<b>SAMPLE TYPE:</b>	<b>TEST REQUEST:</b>									
Blood EDTA <input type="checkbox"/> for DNA or gene tests	Gene test: _____									
CVS <input type="checkbox"/> Amnio <input type="checkbox"/> Fetal blood <input type="checkbox"/> POC <input type="checkbox"/>	Test directory clinical indication and code (reason for testing) <a href="https://www.england.nhs.uk/publication/national-genomic-test-directories/">https://www.england.nhs.uk/publication/national-genomic-test-directories/</a>									
Other (please state)	<table border="0"> <tr> <td>R171.1 Cholestasis <input type="checkbox"/> (NGS Panel)</td> <td>R172.1 Wilson Disease <input type="checkbox"/> (ATP7B single gene sequencing)</td> <td>R173.1 Polycystic Liver Disease <input type="checkbox"/> (NGS Panel)</td> </tr> <tr> <td>R175.1 Pancreatitis <input type="checkbox"/> (NGS Panel)</td> <td>R175.2 Pancreatitis <input type="checkbox"/> (CFTR single gene sequencing)</td> <td>R177.1 Hirschsprung <input type="checkbox"/> (RET single gene sequencing)</td> </tr> <tr> <td>R331.1 Intestinal Failure <input type="checkbox"/> (NGS Panel)</td> <td>Crigler-Najjar Syndrome <input type="checkbox"/> (UGT1A1 full gene sequencing performed as part of Large Panel R171.1)</td> <td></td> </tr> </table>	R171.1 Cholestasis <input type="checkbox"/> (NGS Panel)	R172.1 Wilson Disease <input type="checkbox"/> (ATP7B single gene sequencing)	R173.1 Polycystic Liver Disease <input type="checkbox"/> (NGS Panel)	R175.1 Pancreatitis <input type="checkbox"/> (NGS Panel)	R175.2 Pancreatitis <input type="checkbox"/> (CFTR single gene sequencing)	R177.1 Hirschsprung <input type="checkbox"/> (RET single gene sequencing)	R331.1 Intestinal Failure <input type="checkbox"/> (NGS Panel)	Crigler-Najjar Syndrome <input type="checkbox"/> (UGT1A1 full gene sequencing performed as part of Large Panel R171.1)	
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Date of collection:	If the clinical indication and code are not provided, a panel will be applied based on the clinical information provided.									
Time of collection:	DNA storage only <input type="checkbox"/>									
For Departmental Use Only:	Other (please specify) _____									

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