### Molecular Genetic Testing for Alagille Syndrome

### Analysis of the *JAG1* and *NOTCH2* genes

*Please send* ***EDTA*** *blood (1ml minimum for neonates, 5-10ml for children and 10-20ml for adults) or DNA to Prof. S. Ellard, Molecular Genetics Laboratory, RILD Level 3*, *Royal Devon & Exeter NHS Foundation Trust, Barrack Road, Exeter EX2 5DW with this form*.

rde-tr.MolecularGeneticsAdmin@nhs.net

**Please fill in as fully as possible and tick boxes where appropriate.**

Lead Clinical Geneticist: Dr P Turnpenny (01392-405726 or peter.turnpenny@nhs.net)

Consultant Molecular Geneticist: Professor S Ellard (01392-408259 or sian.ellard@nhs.net)

**Patient details Requestor Details**

|  |  |
| --- | --- |
| **SURNAME:**      | **CLINICIAN NAME:**      |
| **FIRST NAME(S):**      | **TELEPHONE:**      |
| **DATE OF BIRTH: (dd/mm/yyyy)**      | **E-MAIL ADDRESS REQUIRED:**       |
| **PATIENT’S POSTCODE (UK ONLY):**      | **REPORT ADDRESS:**      | **INVOICE ADDRESS:**      |
| **NHS NUMBER:**      |
| **GENDER:**Male[ ]  Female[ ]  |
| **ETHNIC ORIGIN**       | **GENETICS NO.:**      |

**Clinical Information**  Age at diagnosis:

|  |  |
| --- | --- |
| **Liver**  Prolonged neonatal jaundice [ ]   | Liver failure [ ]   |
|  Conjugated hyperbilirubinaemia [ ]   | Biopsy findings (if performed) |       |
|  Other liver features [ ]  |       |
| **Heart:** Pulmonary stenosis [ ]   | Tetralogy of Fallot [ ]   |
|  Atrial Septal Defect [ ]   | Aortic stenosis [ ]   |
|  Ventricular Septal Defect [ ]   | Other (please specify) [ ]   |       |
| **Face:**  Facial features suggestive of Alagille syndrome [ ]  |        |
|  Facial features suggestive of Hajdu Cheney syndrome [ ]  |        |
| **Eye:** Posterior embryotoxon [ ]   | Other eye abnormality (please specify) |        |
| **Skeletal:** Vertebral abnormality (please specify) [ ]  |       |
| **Renal:** (please specify) [ ]  |       |
| **Other:** (please provide details)[ ]   |       |
| **Family History:** Yes [ ]  No [ ]  Details of affected family members**:**     **NB. Please include a pedigree indicating relationships and details of affected family members.***If genetic testing has been carried out in this family please give details below* |
| **Test requested**: Mutation analysis of the *JAG1* and *NOTCH2* genes by Next Generation sequencing (£750\*) [ ] *JAG1* sequencing and dosage analysis (£600\*) [ ] *JAG1 (*exons 1-26) sequencing andMLPA **then***NOTCH2* (exons 1-34) sequencing if *JAG1* negative (£1200\*)[ ] *NOTCH2* sequencing (patients with previous *JAG1* negative result) (£600\*) [ ] \* For current prices please use the latest version of the request form and note that a 25% overhead will be applied for non-NHS referrals.  |
| **Testing for known familial mutation**Is this patient Affected [ ]  Unaffected [ ]  (If this patient is affected please provide clinical details above) |
| Name of index case/proband |       | **Laboratory where testing of index case was carried out:**       |
| Relationship to patient |       |
| Mutation details |       |
| **Please include a copy of the laboratory report for the index case/proband** |