### 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*. **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](mailto:peter.turnpenny@nhs.net))

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

**Patient details Requestor Details**

|  |  |  |
| --- | --- | --- |
| **SURNAME:** | **CLINICIAN NAME:** | |
| **FIRST NAME(S):** | **TELEPHONE:** | |
| **DATE OF BIRTH: (dd/mm/yyyy)** | **E-MAIL ADDRESS:** | |
| **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**:  *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** | | | | | | | | | |