|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **GENETICS:** *T:* 020 7188 1696/1709 *F:* 020 71881697  [dnadutyscientist@viapath.co.uk](mailto:dnadutyscientist@viapath.co.uk)  [cytodutyscientist@viapath.co.uk](file:///\\gstt.local\shared\Genetics\Quality%20Management\Quality%20Management%20Documents\cytodutyscientist@viapath.co.uk)  **BIOCHEMICAL GENETICS:** *T:* 020 71882591 *F:* 020 71887275  **CLINICAL GENETICS:** *T:* 020 71881364 *F:* 020 71881369  **GENETICS SPECIMEN FORM**  **Genetics Laboratories, 5th Floor, Tower Wing, Guy’s Hospital,**  **Great Maze Pond, London, SE1 9RT**  <http://www.viapath.co.uk/departments-and-laboratories/genetics> | | | | |  | | | | | | |
| Surname: |  | | **Sex:** | | | M  F | | | | | |
| First Name: |  | | Ethnic origin: | | |  | | | | | |
| Previous Name: |  | | Hospital number: | | |  | | | | | |
| DOB: |  | | PRU Number: | | |  | | | | | |
| Address: |  | | Post code: | | |  | | | | | |
| NHS Number (Mandatory): |  | | Private patient (please attach invoicing details): | | |  | | | | | |
| GP name: |  | | GP Post code: | | |  | | | | | |
| Consultant: |  | | Referring Hospital: | | |  | | | | | |
| Full address for returning report including Department: | | | | | | | | | | | |
| Signed: Date: | | | | | | | | | | | |
| Name (print) Email: | | | | | | | | | | | |
| **Invoice address if different from referral address:** | | | | | | | | | | | |
| Samples please ensure specimens are dispatched to the laboratory promptly after sampling | | | | | | | | | | | |
| Blood in potassium EDTA (DNA / MLPA / array CGH) | | | |  | | | | | | | |
| Blood in lithium heparin (Chromosome rearrangements / Biochemical Genetics) | | | |  | | | | | | | |
| Prenatal sample Please tick one: | | | | CVS  AF  POC | | | | | | | |
| Other – Please state | | | |  | | | | | | | |
| **Tests requested**  **NB** For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form. | | | | **Clinical Details** Please include full details of patient, with pedigree if relevant)  **NB** For testing for chromosome imbalance (array CGH/chromosome analysis), please provide clinical details on the reverse of this form. | | | | | | | |
| In submitting this sample, the clinician confirms that **consent has been obtained**:  (a) for testing and possible storage  (b) for the use of this sample and the information generated from it to be shared with members of the donor’s family and their health professionals (if appropriate).  (c) we assume that consent has been obtained for sensitive disposal of any fetal remains unless otherwise stated. **Please do NOT send the consent form** | | | | Has this case been discussed with the Genetics Department? If so, with whom?  Is the patient pregnant?  If YES: how many weeks gestation? | | | | | | | |
| **All fields above are mandatory. Samples supplied with inadequate or illegible information, will be subject to delay or rejection.** | | | | | | | | | | | |
| |  |  | | --- | --- | | For Departmental use only |  | | | | | | | | | | | | |
| NHS Number: | | | | | | | | | | | |
| CLINICAL INFORMATION – for chromosome imbalance testing *Place an X in the box if statement applies to the subject.* | | | | | | | | | | | |
| 1 Cognitive Development | | | Typical | | | | | | | | |
|  | | | Delay (Atypical) | | | | | | | | |
|  | | | Mild (IQ 50-69; for adults mental age 9-12 yrs) | | | | | | | | |
|  | | | Mod (IQ 35-49; for adults mental age 6-9 yrs) | | | | | | | | |
|  | | | Severe (IQ 20-34; for adults mental age 3-6 years) | | | | | | | | |
|  | | | Profound (IQ <20; for adults mental age <3 years) | | | | | | | | |
| 2 Specific Developmental Disorder | | | Speech and language  Reading and spelling  Arithmetic  Motor Skills | | | | | | | | |
| 3 Neurodevelopmental/Behavioral Problems | | | Autistic Spectrum Disorder | | | | |  | Yes |  | No |
|  | | | ADHD | | | | |  | Yes |  | No |
|  | | | Tics | | | | |  | Yes |  | No |
|  | | | Sleep | | | | |  | Yes |  | No |
|  | | | Feeding | | | | |  | Yes |  | No |
|  | | | Psychosis | | | | |  | Yes |  | No |
|  | | | Other behavioral problems | | | | |  | Yes |  | No |
| 4 Neurological Disorders | | | Vision  Hearing  Abnormal tone/involuntary movements  Structural brain lesion | | | | | | | | |
|  | | | Cerebral Palsy Unilateral  Cerebral Palsy Bilateral | | | | | | | | |
|  | | | Epilepsy  Age of onset <3 months  3-24 months  > 24 months | | | | | | | | |
| 5 Growth Abnormalities | | | At birth Small for gestational age (<10th centile) | | | | |  | Yes |  | No |
|  | | | At birth Large for gestational age (>90th centile) | | | | |  | Yes |  | No |
|  | | | Current: | | | | |  | | | |
|  | | | Tall stature (height >95th centile) | | | | |  | Yes |  | No |
|  | | | Short Stature (height < 5th centile) | | | | |  | Yes |  | No |
|  | | | Macrocephaly (>95th centile) | | | | |  | Yes |  | No |
|  | | | Microcephaly (<5th centile) | | | | |  | Yes |  | No |
| 6 Congenital Malformations/Dysmorphism | | | Heart disease (e.g. ASD, VSD) | | | | |  | Yes |  | No |
|  | | | Renal and Urogenital malformations | | | | |  | Yes |  | No |
|  | | | Brain Malformations | | | | |  | Yes |  | No |
|  | | | Eye malformations (e.g. anophthalmia, microphthalmia) | | | | |  | Yes |  | No |
|  | | | Ear malformations | | | | |  | Yes |  | No |
|  | | | Cleft lip  Cleft palate | | | | |  |  |  |  |
|  | | | Micrognathia | | | | |  | Yes |  | No |
|  | | | Limb abnormalities (e.g. short or long bones) | | | | |  | Yes |  | No |
|  | | | Digital abnormalities (e.g. syndactyly, polydactyly) | | | | |  | Yes |  | No |
|  | | | Facial dysmorphism e.g. hypertelorism | | | | |  | Yes |  | No |
| **7 Endocrine and metabolic conditions** | | |  | | | | |  | Yes |  | No |
| **8 Cutaneous stigmata/skin lesions** | | |  | | | | |  | Yes |  | No |
| **9 Hair, nail, teeth abnormalities** | | |  | | | | |  | Yes |  | No |
| **10 Other Skeletal abnormalities eg scoliosis** | | |  | | | | |  | Yes |  | No |