viapath

Guy's and St Thomas' NHS
NHS Foundation Trust

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

MOLECULAR GENETICS: *T:* 020 7188 1696/2582 *F:* 020 71887273
CYTOGENETICS: *T:* 020 71881709 *F:* 020 71881697
BIOCHEMICAL GENETICS: *T:* 020 71882591 *F:* 020 71887275
CLINICAL GENETICS: *T:* 020 71881364 *F:* 020 71881369

PATIENT DETAILS

Surname:

First Name:

Previous Name:

DOB: Sex: M / F

Address:

Postcode:

Ethnic Origin:

Hospital Number:

NHS number (mandatory):

Private Patient (please attach invoicing details) ☐

G.P DETAILS

Name:

Postcode:

REFERRING HOSPITAL

Consultant:

Full address for return of report including department:

Signed: Date:

Name (PRINT): Tel no:

Fax no:

SAMPLES

Blood in potassium EDTA
(DNA / MLPA / array CGH) ☐

Blood in lithium heparin
(Chromosome rearrangements / Biochemical Genetics) ☐

Prenatal sample (Please circle) CVS / AF / POC

Other (Please state) _____

Date and time sample taken:

Please ensure specimens are dispatched to the laboratory promptly after sampling

TESTS REQUESTED

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).

Please do NOT send the consent form

CLINICAL DETAILS/REASON FOR REFERRAL

(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.

Has this case been discussed with the Genetics Department? If so, with whom?

Is the patient pregnant? Y / N

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

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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 & language ☐ **Reading/spelling** ☐ **Arithmetic** ☐ **Motor skills** ☐

3. Neurodevelopmental/Behavioural Problems

Autism Spectrum Disorder ☐ **ADHD** ☐
Tics ☐ **Sleep** ☐ **Feeding** ☐ **Psychosis** ☐ **Other Behavioural Problems** ☐

4. Neurological Disorders

Vision ☐ **Hearing** ☐ **Abnormal tone Involuntary Movements** ☐
Structural brain lesion (eg dysplasia, tumour)
Cerebral Palsy Unilateral ☐ **Cerebral Palsy Bilateral** ☐
Epilepsy ☐
Age of onset: **<3 months** ☐ **3-24 months** ☐ **> 24 months** ☐
Seizure type documented: **atonic** ☐ **myoclonic** ☐ **other** ☐

5. Growth abnormalities

At birth Small for gestational age (<10th centile) ☐ Large for gestational age (>90th centile) ☐
Current Tall stature (height >95th centile) ☐ Short Stature (height < 5th centile) ☐
Macrocephaly (>95th centile) ☐ Microcephaly (<5th centile) ☐

6. Congenital Malformations/Dysmorphism

Heart disease (eg ASD, VSD) ☐ Renal and Urogenital malformations ☐ Brain malformations ☐
Eye malformations (eg anophthalmia, microphthalmia) ☐ Ear malformations ☐
Cleft Lip ☐ Cleft Palate ☐ Micrognathia ☐
Limb abnormalities (eg short or long bones) ☐ Digital abnormalities (eg syndactyly, polydactyly) ☐
Facial dysmorphism eg hypertelorism ☐

7. Endocrine and metabolic conditions ☐

8. Cutaneous stigmata/skin lesions ☐

9. Hair, nail, teeth abnormalities ☐

10. Other Skeletal abnormalities eg scoliosis ☐