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NHS
Guy's and St Thomas'
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

GENETICS: *T:* 020 7188 1696/1709 *F:* 020 71881697
dnadutyscientist@viapath.co.uk
cytodutyscientist@viapath.co.uk
BIOCHEMICAL GENETICS: *T:* 020 71882591 *F:* 020 71887275
CLINICAL GENETICS: *T:* 020 71881364 *F:* 020 71881369

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: Clinical Genetics Department at Guy's Hospital, 5th Floor, Tower Wing Great Maze Pond, London, SE1 9RT

Signed: **Date:**

Name **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)	☑ 3-5 ml	Date of collection
Blood in lithium heparin (Chromosome rearrangements / Biochemical Genetics)	☐	Time of collection
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.

Store DNA

EDTA SAMPLE FOR WGS PLEASE

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? No

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 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 behavioural 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		