

# GENETICS SPECIMEN FORM

Genetics Laboratories, 5<sup>th</sup> 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

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[cytodutyscientist@viapath.co.uk](mailto:cytodutyscientist@viapath.co.uk)

BIOCHEMICAL GENETICS: T: 020 71882591 F: 020 71887275

CLINICAL GENETICS: T: 020 71881364 F: 020 71881369

Surname:		Sex:	<input type="checkbox"/> M <input type="checkbox"/> F
First Name:		Ethnic origin:	
Previous Name:		Hospital number:	
DOB:		PRU Number:	
Address:		Post code:	
NHS Number (Mandatory):		Private patient (please attach invoicing details):	<input type="checkbox"/>
GP name:		GP Post code:	
Consultant:		Referring Hospital:	

Full address for returning report including Department: Clinical Genetics Department at Guy's Hospital, 5<sup>th</sup> 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 <b>EDTA</b> DNA / MLPA / array CGH)	<input checked="" type="checkbox"/> 3-5 ml	Date of collection
Blood in lithium heparin (Chromosome rearrangements / Biochemical Genetics)	<input type="checkbox"/>	Time of collection
Prenatal sample Please tick one:	CVS <input type="checkbox"/> AF <input type="checkbox"/> POC <input type="checkbox"/>	
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:			
<b>CLINICAL INFORMATION – for chromosome imbalance testing</b> <i>Place an X in the box if statement applies to the subject.</i>			
<b>1 Cognitive Development</b>	<input type="checkbox"/> Typical		
	<input type="checkbox"/> Delay (Atypical)		
	<input type="checkbox"/> Mild (IQ 50-69; for adults mental age 9-12 yrs)		
	<input type="checkbox"/> Mod (IQ 35-49; for adults mental age 6-9 yrs)		
	<input type="checkbox"/> Severe (IQ 20-34; for adults mental age 3-6 years)		
	<input type="checkbox"/> Profound (IQ <20; for adults mental age <3 years)		
<b>2 Specific Developmental Disorder</b>	Speech and language <input type="checkbox"/>	Reading and spelling <input type="checkbox"/>	Arithmetic <input type="checkbox"/> Motor Skills <input type="checkbox"/>
<b>3 Neurodevelopmental/Behavioral Problems</b>	Autistic Spectrum Disorder	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	ADHD	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Tics	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Sleep	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Feeding	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Psychosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Other behavioural problems	<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>4 Neurological Disorders</b>	Vision <input type="checkbox"/> Hearing <input type="checkbox"/> Abnormal tone/involuntary movements <input type="checkbox"/> Structural brain lesion <input type="checkbox"/>		
	Cerebral Palsy Unilateral <input type="checkbox"/> Cerebral Palsy Bilateral <input type="checkbox"/>		
	Epilepsy <input type="checkbox"/> Age of onset <3 months <input type="checkbox"/> 3-24 months <input type="checkbox"/> > 24 months <input type="checkbox"/>		
<b>5 Growth Abnormalities</b>	At birth Small for gestational age (<10th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	At birth Large for gestational age (>90th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Current:		
	Tall stature (height >95th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Short Stature (height < 5th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Macrocephaly (>95th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Microcephaly (<5th centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>6 Congenital Malformations/Dysmorphism</b>	Heart disease (e.g. ASD, VSD)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Renal and Urogenital malformations	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Brain Malformations	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Eye malformations (e.g. anophthalmia, microphthalmia)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Ear malformations	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Cleft lip <input type="checkbox"/> Cleft palate <input type="checkbox"/>		
	Micrognathia	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Limb abnormalities (e.g. short or long bones)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Digital abnormalities (e.g. syndactyly, polydactyly)	<input type="checkbox"/> Yes	<input type="checkbox"/> No
	Facial dysmorphism e.g. hypertelorism	<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>7 Endocrine and metabolic conditions</b>		<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>8 Cutaneous stigmata/skin lesions</b>		<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>9 Hair, nail, teeth abnormalities</b>		<input type="checkbox"/> Yes	<input type="checkbox"/> No
<b>10 Other Skeletal abnormalities eg scoliosis</b>		<input type="checkbox"/> Yes	<input type="checkbox"/> No