

THE FORM MUST BE COMPLETED FULLY IN BLACK INK. THE SPECIMEN CONTAINER AND FORM MUST BE LABELLED WITH TWO MATCHING PATIENT IDENTIFIERS, OTHERWISE THE SPECIMEN WILL BE REJECTED. SEE OVERLEAF FOR SAMPLE REQUIREMENTS

PATIENT DETAILS (Fields marked with * are mandatory)

STICK YOUR PRINTED LABEL OVER THIS SECTION IF AVAILABLE

FAMILY NAME*

FIRST NAME(S)*

DATE OF BIRTH*

CHI/NHS NUMBER*

POSTCODE*

PEDIGREE/REFERENCE

SEX* MALE FEMALE UNKNOWN

ETHNIC ORIGIN if relevant e.g. for CF

REFERRING CONSULTANT (FOR REPORTING)

CLINICIAN

TELEPHONE

EMAIL

ADDRESS

POSTCODE

IF TESTING IS CHARGEABLE, PLEASE SUPPLY A PURCHASE ORDER OR REFERENCE NUMBER

REASON FOR REFERRAL/TEST REQUEST

TESTING REQUESTED (Specify gene/variant if known)

No testing, storage only

CLINICAL DETAILS (including age at presentation and related symptoms)

IS THERE A FAMILY HISTORY? YES NO

(include name and CHI/DOB of index case)

IS THE PATIENT CLINICALLY AFFECTED? YES NO

IS THE PATIENT/PARTNER PREGNANT? YES NO

HOW MANY WEEKS GESTATION? WEEKS URGENT

Please note that additional clinical information or completion of a pro-forma may be required for specialist tests (see page 2)

SAMPLE DETAILS

BLOOD EDTA (KE) (DNA Tests / Microarray)

BLOOD Li Heparin (Cell culture based tests / Karyotyping)

OTHER Specify: DNA, AF, CVS, Tissue etc.

TAKEN BY DATE

PHONE/PAGER TIME

LOCATION

CONSENT & RECORD OF DISCUSSION

Please ensure testing and /or storage of genetic material is discussed with the patient and that a summary of clinical consent is included in the patient's health record. Further information regarding consent can be found at <https://www.bsgm.org.uk/healthcare-professionals/confidentiality-and-genetic-information>. The patient should discuss and understand the following:

- Family implications.** The results of my test may have implications for other members of my family. I acknowledge that my results may sometimes be used to inform the appropriate healthcare of others. This could be done in discussion with me, or in such a way that I am not personally identified in this process.
- Uncertainty.** The results of my test may reveal genetic variation whose significance is not yet known. Deciding whether such variation is significant may require sharing of information about me including (inter)national comparisons with variation in others. I acknowledge that interpretation of my results may change over time as such evidence is gathered.
- Unexpected information.** The results of my test may reveal a chance of a disease in the future, and nothing to do with why I am having this test. This may be found by chance, while focusing on the reason for my test, and I may then need further tests to understand what this means for me. If these additional findings are to be looked for, I will be given more information about this.
- DNA storage.** Normal laboratory practice is to store the DNA extracted from my sample even after the current testing is complete. My sample might be used as a 'quality control' for other testing, for example, that of family members.
- Data storage.** Data from my test will be stored to allow for possible future interpretations.
- Health records.** Results from my test and my test report will be part of my patient health record.

FOR LAB USE ONLY

SHEETS SCANNED

INSTRUCTIONS AND SPECIMEN REQUIREMENTS

All specimens should be clearly labelled with surname, forename and date of birth/CHI number and these details must match those supplied on the test referral form. If the specimen label does not match the information that is supplied on the test referral form, **the sample will be discarded.** Please ensure other labels e.g. those which are added to the specimen tube by other laboratories, **do not obscure the original specimen label** containing the patient identifiers (name and date of birth/CHI number) or the specimen may be rejected.

The specimen should be placed in a sealed plastic bag containing absorbent material, with the accompanying referral form placed in a separate compartment, preventing contamination in the event of leakage. Blood and fluid specimens sent through the post should be packaged in accordance with PI 650 and UN3373 regulations.

If a delay in sending a specimen is unavoidable, blood and solid tissue specimens should be refrigerated overnight, specimens for prenatal diagnosis (AF or CVS) should be stored at room temperature. **DO NOT FREEZE.**

BLOOD SPECIMENS

To enable automated processing, please use 4ml containers for collection of adult/paediatric blood specimens.

Molecular DNA investigations (and/or storage)	EDTA (KE)	Adult/Paediatric-3ml Newborn-1ml
Micro array analysis Patients > 6 Months of age Patients < 6 Months of age only	EDTA (KE) Lithium heparin and EDTA	Adult/Paediatric-3ml Newborn-1ml
Karyotype (chromosome analysis) only	Lithium heparin	Adult/Paediatric-3ml Newborn-1ml
Rapid aneuploidy/chromosomal sex screen	Lithium heparin and EDTA (KE)	Newborn-1ml (in each tube)

PRENATAL DIAGNOSIS

Amniotic fluid (AF)	Sterile 20ml universal	15-20ml
Chorionic villus biopsy (CVS)	Sterile 20ml universal containing CVS transport medium (supplied by the Laboratory)	10 – 20 mg of villus for most referrals
For AF and CVS referrals (parental bloods): Non-Abscan - Maternal blood Abscan for prenatal micro array- Maternal and paternal bloods	EDTA (KE) blood Lithium heparin and EDTA (KE) bloods	3ml 3ml in each tube

OTHER REFERRALS

Products of conception (PoC) Include appropriate consent forms.	Dry, well-sealed container	Should NOT be placed in formalin fixative.
Skin/other tissue	Sterile 20ml universal containing transport medium (supplied by the Laboratory)	

ADDITIONAL INFORMATION/PRO-FORMA

Some tests may require approval by a Clinical Geneticist before testing can be performed. You may also be asked to complete a further specialist request form/pro-forma e.g. Glasgow Epilepsy Service, Disorders of Sexual Development. These and an electronic copy of this form can be downloaded from the Department's web page www.nhsggc.org.uk/medicalgenetics. Please ensure an email address is provided to allow the Duty Scientist to contact you if further clinical details or completion of a specialist form is required.

If further copies of this form are required please contact the laboratory on 0141 354 9300 or email genetic.laboratories@ggc.scot.nhs.uk