

Genetic / biochemical study request form

| Medical Doctor /Researcher requesting the test: | | | |
|---|--------------|--|--|
| Name: | Surname: | | |
| Center/ Hospital: | Department: | | |
| Address: | Postal Code: | | |
| City: | Country: | | |
| Tel.: | Fax: | | |
| e-mail: | | | |
| Patient's information | | | |

| Name: | Surname: | Clinical history Number / Hospital: | | |
|-------------------|-------------------------|---------------------------------------|--|--|
| Case number: | Patient number: | (to be filled by BG) | | |
| Date of birth: | Gender: | □ Male □ Female | | |
| Sample from: 🛛 Pa | tient D Patient's relat | Patient's relative (indicate kinship) | | |

Biologic sample information

Indicate the number of tubes you are sending, the type of tube, the type of sample (peripheral blood, serum or DNA), the approximate volume of each tube, the collection date of the sample and the data for DNA samples.

| | PERIPHERAL BLOOD (Genetic or biochemical studies from plasma) | SERUM (Biochemical studies) | DNA (3 micrograms minimum) (genetics studies) |
|---|---|--------------------------------|---|
| Number of tubes (ex. 2) | | | |
| Type of tube (EDTA/without anticoagulant, eppendorf tube) | | | |
| Approximate volume per tube (ex. 10 ml, 50 ul) | | | |
| Date of sample collection | | | |
| Concentration ng / ul (ONLY to be filled in for DNA) | | | |
| Method / collection kit (to be filled in for DNA only) | | | |
| Method of quantification (nanodrop / others) (ONLY to be filled in for DNA) | | | |

- □ Informed consent for genetic diagnostic COMPULSORY
- □ Informed consent for research
- □ Clinical report
- Genetic report is **COMPULSORY** for familial Sanger studies with known mutation/s
- □ Other reports. Please specify:

Requested genetic study. Please, mark with an "X"

Genetic studies by NGS Panels

- **10010** NGS panel Hemochromatosis and Hyper-/hipoferritinemias
- □ 10020 NGS panel Congenital sideroblastic anemia and acquired sideroblastic anemia
- **10030** NGS panel Anemia due to defects in iron metabolism genes
- □ 10040 NGS panel Congenital dyserythropoietic anemia
- **10050** NGS panel Congenital erythrocytosis / Familial polycythemia

□ **10061** NGS panel Hereditary hemolytic anemias due to red blood cell enzymopathies or Glycogen storage disease

- **10062** NGS panel Hemolytic anemias due to membranopathies (spherocytosis, elliptocytosis,
- pyropoikilocytosis, ovalocytosis, stomatocytosis) & Gilbert Syndrome
- **10070** NGS panel Hereditary hemolytic anemia including membranopathies and enzymopathy (10061 + 1062)
- **10080** NGS panel NGS panel Erythropoietic protoporphyria and congenital erythropoietic porphyria
- □ **10090** NGS panel Fanconi anemia
- □ 10100 NGS panel Blackfan-Diamond anemia
- □ 10110 NGS panel Dysqueratosis congenita
- **10111** NGS panel Bone Marrow Failure (10090 + 10100 + 10110)
- □ 10120 NGS panel Neurodegeneration with brain iron accumulation (NBIA)

Genetic study by Sanger

- **20050** Sanger study of Hereditary Hyperferritinemia-Cataract (exon1 FTL)
- **20051** Sanger initial study of Hemochromatosis (exon4 HFE)
- **20052** Sanger study complete gene HBB (beta globin)

Familial genetic study by Sanger with known mutation/s (specify familial case).

Write down the gene/s to be studied: ____

Compulsory to include a genetic report from the proband

- **20010** Sanger study in a relative for one mutation in one gene
- **20020** Sanger study in a relative for two mutations in one gene
- **20030** Sanger study in a relative for three mutation in one gene

Data analysis studies (contact info@bloodgenetics.com to send data)

- **30010** NGS panels data analysis
- □ 30020 Whole Exome Sequencing (WES) data analysis

Requested biochemical study. Please, mark with an "X"

- **90010** Mesure of Hepcidin hormone in serum or plasma by ELISA
- **90020** Mesure of Labile Plasma Iron, LPI in serum or plasma

Relevant clinical information

□ Familial case

Kinship (father, mother, son, daughter, maternal/paternal uncle, aunt...):

□ Sporadic case

(In case of a family case, it is necessary to send a pedigree of the family. Send separate genetic study requests for each relative to be tested)

CLINICAL INFORMATION to include in the patient's report (fill in only the most relevant information):

(For example: 35 year old male/female with suspected disease xxxx).

| Billing information | |
|--------------------------|--|
| Institution/ Company: | |
| Authorized person: | VAT Number: |
| Address: | City/Country/Postal Code: |
| Project (if applicable): | Petition number/ order number (if relevant): |

Notes

About the service request: Please contact us before placing the order if you have any questions so we can advise you correctly on the type of test and the sample type more suitable for you.

About the personal data: Under the Law 15/1999 on Protection of Personal Data, the applicant for the test should have the patient's consent to perform the requested tests and process the personal data. Please send a copy of the signed informed consent with the application, pedigree (if applicable) and samples.

About biological samples: BLOODGENETICS keep received samples or their derivatives indefinitely for future validation and / or research, maintaining complete anonymity on the origin of each sample. The patient or his representative has the right not to consent to the use of his/her samples for research purposes even if this is not detrimental to the diagnostic tests or results. If the patient wants his/her sample to be used for research, please send the research informed consent together with the request form, the genetic informed consent, the pedigree (if applicable) and the samples. The patient may decline this consent at any time by contacting BLOODGENETICS under the following address: BLOODGENETICS, Edificio LABCO. Verge de Guadalupe, 18, 08950 Esplugues de Llobregat, Barcelona, España.

Shipment of biological samples: Please, contact us before sending samples to adequately advise the best method of preserving and sending them. We recommend blood samples be sent within 24 hours after collection keeping them at room temperature, never cold. DNA samples can be sent resuspended in TE or water, keeping them cold or at room temperature.

Right to claim: We remind you that you can exercise your right to make a complaint about a genetic study carried out by BLOODGENETICS. You should send your complaint in writing together with a photocopy of your Identity Card or passport, addressed to BLOODGENETICS. Edificio LABCO. Verge de Guadalupe, 18, 08950 Esplugues de Llobregat, Barcelona, España.

Signature