

Sample delivery instructions

Any sample delivery must follow the next requirements. If you have any questions, do not hesitate to contact the head manager of the Molecular Diagnostics Platform.

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The format for sending samples (DNA concentrates, minimum quantities and volumes required, etc.) varies depending on the different molecular studies and the specific requirements for each technique.

In order to carry out individualized studies, it is possible to send either **peripheral blood in EDTA** (purple lid) or **pre-extracted DNA**.

Sample delivery requisites

Sample delivery technique requisites:

- To carry out studies using sequencing panels, the minimum quantity of DNA is 100 ng and the quality should be DNA: ratio Abs 260/280 $\geq 1,8$ 1,8260/230 ≥ 2 .
- **Obtaining DNA** from 10-16 ml of peripheral blood collected in **EDTA (purple lid)**. It should be sent at room temperature and must arrive within **24 hours** of extraction. In the event that the sample cannot be sent the same day as the extraction, keep the blood refrigerated at 4°C until the next day when it can be sent at **room temperature**. A delay in sending or conserving it at an inappropriate temperature can lengthen the processing time and compromise the sample.
- If you are going to send **extracted DNA**, please check beforehand because the quantity we require will depend on the molecular study requested. It is preferable to send extracted DNA in **international sample deliveries**. In the event that the DNA received does not meet the minimum quality and integration requirements, the sender will be notified.
- **Obtaining RNA** from 4-8 ml of peripheral blood collected in EDTA (purple lid). The sample should be sent **within 24 hours** and it should be refrigerated in a box at 4°C with ice packs. However, so the sample does not freeze, direct contact with the ice packs should be avoided by simply wrapping the samples in a cloth.

NOTE:

In the case of the Facioscapulohumeral Muscular Dystrophy (FSHD) molecular study, it is preferable to send 10-20 ml of peripheral blood in EDTA, as a minimum quantity of 30 micrograms of DNA is required, preferably concentrated between 400-500 ng/μl. If extracted DNA is delivered for this molecular diagnosis, please kindly follow these instructions. If possible, deliver 30 DNA micrograms in case it is needed to repeat the study or run complementary techniques.

In the case of **prenatal studies** (amniotic liquid or chorionic villus), please make an appointment at **least** 15-30 days in advance. Sample conditions will depend on the molecular study requested.

Additionally, it is advisable to remember that:

- All tubes should be correctly labelled and sent with the corresponding request document, filled in as appropriate. This should include detailed instructions for the study and other family background information that might help interpret the results in a more appropriate context. This is a very important point of the Facioscapulohumeral Muscular Dystrophy type 1 (FSHD1) molecular study since knowing the family background can help on the diagnosis for better.
- The patient (or legal representative) must have signed the informed consent sheet to undergo this genetic test. This sheet should be sent with the analysis application.

In order to make an enquiry and order diagnostics services, kindly contact the head manager of the Molecular Diagnostics Platform.

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The Molecular Diagnostics Platform reserves the right to reject any request or sample that does not meet the set requirements and promises to notify the requesting party about any incident related to identifying, handling or processing the sample or request sheet (loss, accident, etc.).