NIMGenetics
New Integrated Medical Genetics

PROCEDURES FOR OBTAINING, PREPARING AND SHIPPING SAMPLES FOR GENETIC DIAGNOSIS

Before the samples are obtained please fill out the informed consent form.

This document should be read jointly by the patient and the physician or genetic counselor to ensure the best understanding.

It is essential that this document contain the following:

- The patient´s personal data.
- Name and e-mail address of the requesting physician or GP to which the report will be submitted.
- Signature of the patient and the physician or GP requesting the test.

Before the samples are obtained, please consult the shipping conditions as well as the type and volume of sample required in the following tables.
Please use our courier service to ship the samples, available from 8 to 17.30h (Monday to Thursday) and 8 to 15.00h (Friday).

If the sample you wish to send us does not meet the minimum requirements, we recommend that you contact out Technical Department prior to sending the sample.

Tel. 918 047 760
info@NIMGenetics.com

POSTNATAL DIAGNOSTICS (excluding tumor samples)

All samples must be sent at ROOM TEMPERATURE

Type of technique	Type of sample	Quantity
DNA-based molecular diagnostics (sequencing of unique genes, MLPA, NGS panels and ExoNIM)	Peripheral blood in EDTA (tubes with purple top)*	1-2 tubes (5-10 ml)
	DNA at a concentration greater than 10 ng/ml, dissolved in water, low TE buffer (≤0.1 mM EDTA) or 10 mM Tris	200-1000 ng**
Molecular disgnostics based on RNA-sequencing	Peripheral blood in EDTA (tubes with purple top)	1-2 tubes (5-10 ml)
CGH-Arrays	Peripheral blood in EDTA (tubes with purple top)	1-2 tubes (5-10 ml)
	DNA	1-2 µg***

Oral mucosa swab smear
Metabolopathies
Dried blood on Whatman paper

*Blood extraction will be performed through venipuncture and the patient will not need any special draw requirements.
** Depends on the test requested. Please enquire.
*** Depends on the array platform requested. In those cases with limited DNA availability please enquire.

PRENATAL DIAGNOSTICS

Unless otherwise specified, samples should be shipped at room temperature within 24 h.

Type of technique	Type of sample*	Quantity
Karyotype	Amniotic fluid	10-20 ml
DNA-based molecular sequencing diagnostics		
	Amniotic fluid	10-20 ml
	Chorionic villus	Approx. 2 mm^3
	DNA dissolved in water, low TE buffer (≤0.1 mM EDTA) or 10mM Tris	200-1000 ng**
TrisoNIM	Peripheral blood ATTENTION! Special Delivery!! This procedure requires the sample to be collected in a STRECK tube and shipped inside the NIMTransporter facilitated by NIMGenetics	

Samples processed 72 h after extraction will not be accepted

1 tube (10 ml)

KaryoNIM Prenatal		
	Amniotic fluid (from week 16 onwards)	5-10 ml
	DNA	1000 ng
	Amniocyte culture	
	Chorionic villus	Approx. 2 mm^3
MLPA	Amniocyte culture	
QF-PCR	Amniotic fluid (from week 16 onwards)	5 ml

* In cases of limited sample availability please enquire.
** Depends on the test requested. Please enquire.

ONCOLOGICAL AND HEMATONCOLOGICAL DIAGNOSTICS

Unless otherwise specified samples should be shipped at room temperature

Type of technique	Type of sample*	Quantity

Karyotype /FISH for oncohematology

Bone marrow or peripheral blood in heparin (tubes with green top) 1 tube of bone marrow (0.5-3 ml) and/or 1 tube of peripheral blood (5 ml)

The maximum shipping time is 48 h. In exceptional cases, keep samples refrigerated. Ship on ice (4 ºC) within 16 h of extraction.

Molecular diagnostics

Bone marrow or peripheral blood in EDTA (tubes with purple top) 1 tube of bone marrow (0.5-3 ml) and/or 1 tube of peripheral blood (5 ml)

The maximum shipping time is 48 h. In exceptional cases, keep samples refrigerated. Ship on ice (4 ºC) within 16 h of extraction.

FISH on paraffin-embedded tissue

Paraffin block and one H&E-stained section identifying the tumor area*

FISH on imprints from fresh tissue

Cut open the fresh tissue and softly touch the cut surface several times using the central part of the slide. Let the slide air dry and then place on a covered container to prevent damage.

OncoNIM Seq50

Paraffin block and one H&E-stained section identifying the tumor area

DNA*

Attention!!

Samples must have a DNA concentration greater than 10 ng/µl, dissolved in water, low TE buffer (≤0.1 mM EDTA) or 10 mM Tris 500 ng*

Fresh tissue frozen at -20ºC

Ship on dry ice within a maximum of 18 h 25-50 ng (2-3 mm^3)

CGH Arrays

Peripheral blood in EDTA (tubes with purple top) 1-2 tubes (5-10 ml)

DNA 1-2 µg **

Paraffin block and one H&E-stained section identifying the tumor area*

*In those cases where the H&E sections are not available, the selection of the tumoral area will be performed at NIMGenetics

SAMPLE REJECTION CRITERIA

Samples will be inspected at reception to ensure that they meet the requirements specified in this document.

If anomalies are detected, NIMGenetics will contact the sender, either to complete the information missing or to request a new sample if the one received needs to be discarded.

The most common reasons for sample rejection are the following:

- Patient with no identification (Name/Identification number)
- Inappropriate sample for the requested study
- Date and time of extraction not specified
- Sample origin not specified
- Request form illegible or incorrectly completed
- Samples of fresh tissue/fixed or frozen
- Samples not shipped under the required conditions
- Tubes/containers broken
- Evident external contamination
- Coagulated peripheral blood
- Insufficient volume. If the volume is not sufficient for several tests, the clinician will be asked to specify their priority

Annex 1

INSTRUCTIONS FOR COLLECTION AND SHIPPING OF SAMPLES TO BE USED IN THE PLATFORM KaryoNIM 60k Postnatal ACCORDING TO NIMGENETICS QUALITY PROTOCOLS

<u>Obtaining peripheral blood by venipucture</u>

Peripheral blood extraction must be performed by venipucture. Since the test analyses the DNA, which is not affected by metabolic parameters, there is no need for the patient to fast.

The type of tube recommended to obtain the sample is a sterile tube with EDTA as an anticoagulant. Venipucture is a standard procedure to obtain peripheral blood and does not require special instructions, although it is recommended to

invert the tube several times after extraction to facilitate the mixing of the blood with the anticoagulant agent.

DNA extraction can be performed using different methods, including automated techniques (Magnacube, Qiacube, etc), glass fiber columns, phenol- choroform, or salt extraction (check with NIMGenetics if in doubt regarding any of the procedures). The DNA obtained must be of high quality and quantity; the following values, analyzed by spectrophotometry, are recommended

Concentration >50 ng/μl (at least 2 μg of shipped material)
A260/280 1.8-2.2
A260/230 >1.8

If samples do not meet these requirements please contact MIMGenetics

PROTOCOL FOR TRANSPORTATION AND SHIPPING OF SAMPLES-NIMGENETICS GENOMICS

Samples to be used in genetic studies must be processed in an environment as sterile as possible and that preserves cellular viability. In general, samples must be identified with the same number as that registered in the attached request form.

For shipping, samples must be placed in containers to avoid tube rupture, such as bubble envelopes or preferibly containers for biological hazards. Shipping conditions for each type of samples are specified below.

In the event that you wish to use NIMGenetics Logistic systems, contact 910847760 and arrange collection. A collection schedule must be specified allowing a 2 h bracket from phone call to package pick-up.

1.- Peripheral blood

Blood extracted must be collected in EDTA-tubes (or in heparin-lithium).

Tubes must be identified with the same number as that registered in the attached request form and must be sent to the lab as soon as possible (recommended 2-4 h) at room temperature. In exceptional cases, if transport needs to be postponed until the following day, samples must be kept at 4ºC (NOT FROZEN) until delivery to the courier service.

2.- Isolated nucleic acids

Purified DNA can be delivered at room temperature, 4 ºC or frozen. In the latter case, thermal rupture containers and dry ice must be used.

Once the samples are received, they will be registered and appropriately stored until analysis. For longer storage times, DNA must be kept at -20ºC.