

## PROCEDURES FOR OBTAINING, PREPARING, AND SENDING SAMPLES FOR GENETIC DIAGNOSIS

**The informed consent must be completed prior to the sample collection.**

This document must be read jointly by the patient and their physician or genetic counsellor for better understanding.

Such document must include the following information:

- The patient's personal data.
- Name and e-mail address of the requesting physician or medical practitioner to whom the report shall be sent.
- Signature of the patient and the physician or medical practitioner requesting the test.

**Prior to the sample collection, please check the shipping conditions as well as the volume and type of sample required to carry out the study in the charts below.**

To send samples, use our courier services available from 8 a. m. to 5.30 p.m. from Monday to Thursday and on Friday from 8 a. m. to 3 p. m.

If the sample does not meet the minimum requirements, please contact our Technical Department before sending it.

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## POSTNATAL DIAGNOSIS (except for tumour samples)

All samples should be sent at **ROOM TEMPERATURE**

Type of technique	Type of sample	Quantity
DNA-based molecular diagnostics (single gene sequencing, MLPA, NGS panels, and exomes)	Peripheral blood in EDTA (tubes with purple cap). *	1-2 tubes (5-10 ml)
	DNA at a concentration above 10ngr/ul, dissolved in water, low TE buffer ( $\leq 0.1\text{mM}$ EDTA or 10mM TRIS)	200-1000 ng**
Molecular diagnostics based on RNA sequencing	Peripheral blood in EDTA (tubes with purple cap).	1-2 tubes (5-10 ml)
CGH-Arrays	Peripheral blood in EDTA (tubes with purple cap).	1-2 tubes (5-10 ml)
	DNA	1-2 $\mu\text{g}^{***}$
	Smear of buccal mucosa in swab	-
Metabolopathies	Dried blood on Whatman paper	-

\* Blood shall be drawn by venipuncture, and no special preparation or fasting shall be required by the patient.

\*\* According to the requested test. Check.

\*\*\*According to the requested array platform. In cases with limited DNA availability, please check.

## PRENATAL DIAGNOSIS

Unless otherwise specified in special cases, samples should be sent at **ROOM TEMPERATURE WITHIN 24 HOURS AFTER SAMPLE COLLECTION**

Type of technique	Type of sample*	Quantity
Karyotype	Amniotic fluid	10-20 ml
Molecular diagnostics based on DNA sequencing	Amniotic fluid	10-20 ml
	Chorionic villus	Aprox. 2mm <sup>3</sup>
	DNA dissolved in water, Low TE buffer ( $\leq 0.1$ mM EDTA) or 10 mM TRIS	200-1000ng***
	Peripheral blood in EDTA from parents (tubes with purple cap). ** Along with any type of prenatal sample.	1-2 tubes (5-10 ml)
TrisoNIM <sup>®</sup>	<p><b>Peripheral Blood</b> <b>NOTE: SPECIAL SHIPPING!</b></p> <p>This procedure requires that the sample be collected <b>in a STRECK TUBE</b> and sent in the NIMTransporter container provided by NIMGenetics</p> <p>Samples sent after 72 hours have passed between extraction and processing shall not be accepted.</p>	1 tube (10 ml)
TrisoNIM <sup>®</sup> NEOSEQ	<p><b>Peripheral Blood</b> <b>NOTE: SPECIAL SHIPPING!</b></p> <p>This procedure requires that the sample be collected <b>in two STRECK TUBE</b> and sent in the NIMTransporter container specific for NEOSEQ provided by NIMGenetics</p> <p>Samples sent after 72 hours have passed between extraction and processing shall not be accepted.</p>	2 Tubes de 10 ml por paciente
KaryoNIM <sup>®</sup> Prenatal	Amniotic fluid (as of the 16th week)	5-10 ml
	DNA	1000ngrs
	Amniocyte culture	-
	Chorionic villus	Aprox. 2mm <sup>3</sup>
MLPA	Amniocyte culture	-
QF-PCR	Amniotic fluid (as of the 16th week)	5 ml

\* In cases with limited availability of material, please check.

\*\* Blood shall be drawn by venipuncture, and no special preparation or fasting shall be required by the patient.

\*\*\* According to the requested test. Check.

## ONCOLOGICAL AND ONCOHEMATOLOGICAL DIAGNOSIS

Unless otherwise specified in special cases, samples should be sent at **ROOM TEMPERATURE**

Type of technique	Type of sample	Quantity
Karyotype/FISH oncohematology	Bone marrow or peripheral blood in heparin (tubes with green cap). Maximum shipping time 48 hours. <b>In exceptional cases</b> , keep the sample refrigerated. Delivery on ice (4° C) within 16 hours after extraction.	1 tubo de MO (0,5-3ml) y/o 1 tubo de SP (5ml)
Molecular diagnostics	Bone marrow or peripheral blood in EDTA (tubes with purple cap). Maximum shipping time: 48 hours. <b>In exceptional cases</b> , keep the sample refrigerated. Delivery on ice (4° C) within 16 hours after extraction.	1 tubo de MO (0,5-3ml) y/o 1 tubo de SP (5ml)
FISH in Paraffin Blocks	Paraffin block and H&E stained section, identifying the tumour area*.	
FISH on fresh imprints	Cut the fresh tissue and touch the cut surface gently several times with the centre of the slide. Let the slide dry and put it in a covered container to avoid damages on the sample.	
OncoNIM® Seq50	<b>Paraffin block and H&amp;E stained section, identifying the tumour area*.</b>	
	<b>DNA</b> <b>Attention!</b> Sent samples must have a concentration above 10ngr/ul, dissolved in water, low TE buffer (<0.1mM EDTA or 10mM TRIS)	500 ng*
	Fresh tissue frozen at -20 <C. <b>Shipped on dry ice within a maximum period of 18 hours</b>	25-50ng (2-3 mm <sup>3</sup> )
CGH-Arrays	Peripheral blood in EDTA (tubes with purple cap).	1-2 tubos (5-10 ml)
	<b>DNA</b>	1-2 µg**
	Paraffin block and H&E stained section, identifying the tumour area*.	

\* In those cases in which the H&E stained section is not available and the tumour area is identified, the selection of said area to be studied shall be carried out at NIMGenetics.

\*\*According to the requested array platform. In those cases with limited availability of DNA, please check.

## CRITERIA FOR SAMPLE REJECTION

Samples shall be checked on arrival to ensure that they meet the requirements specified in this document.

If any anomaly is detected, NIMGenetics shall contact the sender either to amend the lack of information or to discard the sample and request a new one.

The most common causes of sample rejection are the following ones:

- Unidentified Patient (Name/Identification No.)
- Inappropriate sample for the required study.
- Unspecified day and time of sample extraction.
- Unspecified sample origin.
- Unreadable or incorrectly filled out request.
- Samples of fresh tissue received are attached or frozen.
- Samples not sent according to required terms.
- Broken tubes / containers.
- Obvious external contamination.
- Clotted peripheral blood.
- Insufficient volume. If the quantity sent is insufficient for the total number of samples requested, the physician shall be asked to specify the priority of the samples.

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