

# SAMPLE COLLECTION MANUAL



Not all sample types are suitable for performing all CGC Genetics tests.

SEE THE UPDATED TEST CATALOG AT  
<http://www.cgccgenetics.com/en/by-test-a-z>  
AND CHECK THE SPECIFIC REQUIREMENTS  
FOR THE TEST YOU WANT TO PERFORM.

#### DO NOT FORGET

1. Complete the CGC Genetics requisition forms, with personal data and clinical and family history, referring the type and reason for testing.
2. Seal the tube/container well with parafilm or adhesive in order to avoid loss and/or contamination of the sample. Label with patient's identification, containing at least two distinct identifiers. Shipping should be well conditioned and at room temperature.
3. If the sample collection is carried out on the eve or during the weekend, it is advisable to keep the sample at room temperature away from sources of cold or heat.

## MOLECULAR DIAGNOSTICS AND CLINICAL GENOMICS

The instructions below are organized by sample type and may differ for a specific test, so it is recommended to refer to the test instructions for the intended test at <http://www.cgccgenetics.com/en/by-test-a-z>.

Sample type	Methodology	Volume / Concentration	Container	Sample viability (days)
Peripheral blood	Sequencing (Sanger and NGS) Fragment analysis Deletion/duplication analysis	≥ 3mL	EDTA	5
	Transcript quantification	≥ 10mL	EDTA	2
			PAXgene	5
DNA	Postnatal: Sequencing (Sanger and NGS) Fragment analysis Deletion/duplication analysis	≥ 100µL (50ng/µL)	Eppendorf Safe-lock	n/a
	Prenatal: <sup>a</sup> Sequencing (Sanger) Fragment analysis Deletion/duplication analysis QF-PCR	≥ 20µL (25ng/µL)		
	Prenatal <sup>a</sup> (NGS)	≥ 50µL (20ng/µL)		
Bone marrow	Sanger sequencing	≥ 1mL	EDTA	2
	Transcript quantification	≥ 3mL	EDTA	2
	RT-PCR	≥ 1mL	PAXgene	5
Amniotic fluid <sup>a</sup>	Sequencing (Sanger) NGS (after cell culture) Fragment analysis Deletion/duplication analysis QF-PCR	≥ 5mL	Sterile tube with leak resistant screw cap (do NOT use urine collection tubes)	4
Chorionic villi <sup>a</sup>	Sequencing (Sanger e NGS) Fragment analysis Deletion/duplication analysis QF-PCR	≥ 30mg	Sterile tube with transport medium and with leak resistant screw cap (do NOT use urine collection tubes)	4
Fibroblasts cell culture <sup>a</sup>	Sequencing (Sanger e NGS) Fragment analysis Deletion/duplication analysis QF-PCR	2 confluent flasks	Flasks T25	2

<sup>a</sup> Maternal blood sample is necessary for exclusion of maternal cell contamination.

## CYTOGENETICS

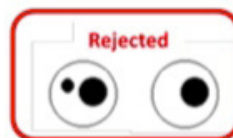
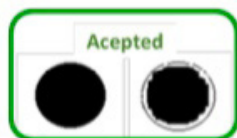
Methodology	Sample type	Volume / Concentration	Container	Sample viability (days)
Array CGH	Peripheral blood	≥ 3mL	EDTA (DNA analysis)	5
	DNA	≥ 100µL (50ng/µL)	Eppendorf Safe-lock	n/a
		≥ 20µL (25ng/µL) Prenatal Diagnosis		
	Amniotic fluid <sup>a</sup>	≥ 5mL	Sterile tube with leak resistant screw cap (do NOT use urine collection tubes)	4
	Chorionic villi <sup>a</sup>	≥ 30mg	Sterile tube with transport medium and with leak resistant screw cap (do NOT use urine collection tubes)	4
FISH	Fibroblasts cell culture	2 confluent flasks	Flasks T25	2
	Peripheral blood	≥ 3mL	Lithium heparin	5
	Bone Marrow	≥ 1mL		5
	Amniotic fluid <sup>a</sup>	≥ 5mL	Sterile tube with leak resistant screw cap (do NOT use urine collection tubes)	4
	Chorionic villi <sup>a</sup>	≥ 30mg	Sterile tube with transport medium and with leak resistant screw cap (do NOT use urine collection tubes)	4
	Fibroblasts cell culture	2 confluent flasks	Flasks T25	2
	Suspension of stimulated fixed cells	1 tube per culture	Sterile tube with leak resistant screw cap	n/a
	Suspension of unstimulated fixed cells (oncology)			4
	Slides, not colored	2 slides per probe	Slide container	4
	Fixed cell culture slides (uncolored)	3 slides per culture		4
Karyotype	Fresh tissue	≥ 1cm <sup>2</sup>	In saline solution and ampicillin or amoxicillin (1g/L)	4
	Tissue, block	≥ 1cm <sup>2</sup>	Block processed in buffered formaldehyde	n/a

<sup>a</sup> Maternal blood sample is necessary for exclusion of maternal cell contamination.

## PRENATAL SCREENING

### HARVEST IN FILTER PAPER

1. Gently massage the finger (ring or middle). Warming the finger causes vasodilation.
2. Select the fingertip as the location for puncture. Clean the area with alcohol and dry with sterile gauze
3. Lancet with a sterile scalpel (supplied by CGC Genetics)
4. Put the finger down to increase blood flow. Let the second drop form. You do not have to press the finger too hard.
5. With the finger facing down, touch each circle to a drop of blood. Blood should cover at least 75% of both sides of each of the five circles. Apply one drop to each circle, and just ONE drop.
6. Allow the blood drops to dry at room temperature (at least 60 minutes) in the horizontal position. Protect from direct sunlight or heat. Do not overlap filter paper during drying.
7. After drying, place the filter paper in the small envelope provided and then in a largest envelope along with the request form (provided by CGC Genetics)
8. Store the sample in a cool and dry place until transport.





# TOMORROW

non invasive prenatal test

Before blood sampling, please read carefully the following instructions.

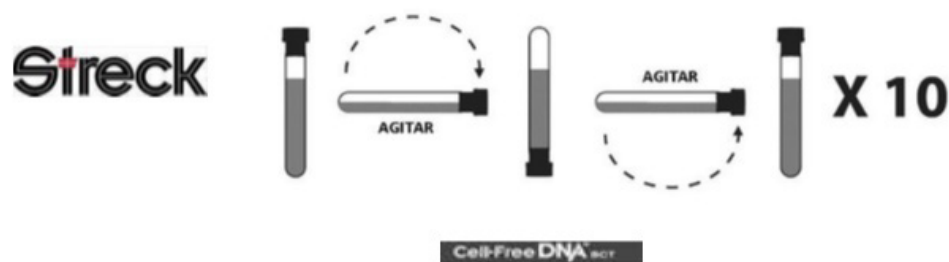
## PROCEDURE:

1. Collect the blood sample into a **Streck** tube (ref.: Cell-Free DNA BCT® CE).
2. Identify the Streck Tube with **at least 2 identifications**, for example, patient's name and date of birth;
3. Clean the region of sampling and harvest by venipuncture between **7–10 mL** of blood into the Streck tube;
4. Immediately after sampling, carefully rotate the tube 8-10 times (inadequate or late mixture may affect the test result);
5. Sample should be stored and shipped at room temperature (20-25°C, 68-77°F);
6. Make sure delivery arrives **within 4 days after blood sampling** and that is addressed to:

**CGC Genetics/Centro de Genética Clínica**

*Rua Sá da Bandeira, 706 - 1º*

*4000-432 Porto, Portugal*



## ATTENTION, THE TEST CANNOT BE PERFORMED IF :

- Blood sample volume is below 7 mL;
- The tube is not identified appropriately;
- The Requisition Form **TOMORROW** is not properly completed and signed by the health professional;
- Patient Informed Consent is not signed by the patient.

To find answers to the most frequent questions, please visit [www.tomorrowtest.com](http://www.tomorrowtest.com)

For additional information, please send us an email to [customercare@cgcgenetics.com](mailto:customercare@cgcgenetics.com) or call + 351 223 389 900, from Monday to Friday 09:00-18:00 (GMT/UTC + 00:00).