

PR 031551	PROTOCOL FOR SENDING CHORIONIC VILLI MITOCHONDRIAL ENZYME DIAGNOSTICS	VALID UNTIL : 01 JAN. 2020
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Protocol for sending chorionic villi for prenatal diagnostics of the respiratory chain enzymes and pyruvate dehydrogenase

Prerequisites for prenatal diagnostics by enzyme activity measurements

Please contact us in order to discuss the possibilities for prenatal testing of your patients. In general, before prenatal testing at the enzyme level can be offered, the examination of the respiratory chain/PDHc in the index patient should have been completed (preferably in muscle and in cultured fibroblasts) and mtDNA mutations should have been excluded.

Duration of pregnancy

The chorionic villi biopsy should be taken between the 11th and 12th week of pregnancy.

Material (requirements)

- For the analysis of enzyme activities **we require approx. 30-40 mg of native chorionic villi**, collected in phosphate buffered saline (PBS), pH 7.4. Furthermore, we require 10 ml of EDTA-blood collected from the mother.
- The chorionic villi should be **free from maternal contamination**.
- The native villi should be collected by centrifugation in a 1.5ml Eppendorf tube (10 minutes; 4°C, 600g). The supernatant has to be discarded, the pelleted material contain the so-called "dry villi".
- Quick-freeze the "dry villi" in liquid nitrogen. Ship the frozen villi in a package containing a sufficient amount of dry ice (8-10 kg) to our laboratory. We also require 10 ml of EDTA-blood collected from the mother as reference material for testing of the villi for maternal contamination by microsatellite marker analysis. The **blood should be kept at room temperature** during shipment, please do not freeze! Please make sure that the tube is protected from possible damage during shipment.

Information about the mother of the patient

- Name of the mother
- Date of birth of the mother
- Registration number or reference number
- Doctor
- Department, hospital
- Insurance information
- Medical indication
- Medication
- Patients permission regarding the above mentioned point 5

Turn around time

The turn around time is 2 weeks.

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Address

Sent the material by courier service to:

Radboudumc
Radboud Center for Mitochondrial Medicine
Translational Metabolic Laboratory – Mitochondrial Enzyme Diagnostics group

815 Radboudumc Diagnostic Laboratory
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More information:

Should you require more information, please contact us as follows:

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