



GUIDELINE AND PROTOCOL FOR THE COLLECTION OF BIOLOGICAL MATERIAL FOR GENETIC ANALYSIS OF REPRODUCTIVE LOSSES

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Genetic analysis of reproductive losses is performed using the following molecular genetic methods:

- QF-PCR (quantitative fluorescent polymerase chain reaction),
- MLPA (multiplex ligation-dependent probe amplification),
- Sanger sequencing.

Biological material suitable for genetic analysis (listed in order of decreasing suitability):

1. Native (unfixed) biological material:

- A 1 x 1 cm excision from the musculus quadriceps femoris or from other fetal tissue (placenta, chorionic villi sampled from the placenta, a section of the umbilical cord, part of the fetal body, etc.).
- After collection, the excision may be rinsed free of excess blood with physiological saline or PBS and dried on sterile gauze / cellulose wadding / a laboratory wipe.
- Storage after collection at the collaborating clinical site: short-term (up to 24 hours) at 4 °C; long-term (up to two weeks) at -20 °C.

2. Formalin-fixed material:

- If the biological material has been placed in formalin, it must be removed from the formalin without delay, washed with physiological saline or PBS, and dried on sterile gauze / cellulose wadding / a laboratory wipe.
- Storage after collection at the collaborating clinical site: at 4 °C, with expedited processing of the collected tissue, given the effect of formalin on the tissue and on DNA integrity.

3. Formalin-fixed paraffin-embedded material (FFPE).

- In specific situations, where no other source of biological material is available, the analysis may also be performed from FFPE samples.





- Storage after collection at the collaborating clinical site: FFPE specimens may be stored long-term at room temperature, with no need for refrigeration or freezing.

Options for obtaining biological material from the mother of the (biological) reproductive loss:

1. Placental excision / whole placenta:

- in the case of native (unfixed) biological material, decidua may be sampled (for storage, see page 1, point 1).

2. Maternal peripheral blood for molecular genetic testing is collected into a Vacuette tube with EDTA:

- Peripheral blood collection does not need to be performed in a fasting state. Immediately after collection, the blood tube must be mixed thoroughly to prevent clot formation.
- Volume: 2 x 2 ml of blood.
- Storage: short-term (1-2 days) at room temperature, with no need for refrigeration or freezing. Long-term at -20 °C (the sample may be thawed only immediately before DNA isolation and must be kept frozen until then).

The procedure for processing biological material collected from reproductive losses is set out in the document „(GIFD) — Biobank Protocol“.

