



Step 2: *HHT Genetic Test Sample Requirements*

Sample Requirements

Proband:

- Blood or DNA from blood.

Pre/peri-natal sample options:

- Cultured amniocytes
- Direct amniotic fluid
- DNA extracted from amniocytes
- CVS tissue
- DNA extracted from CVS
- Cord blood

Maternal blood or DNA from blood (required for all pre/perinatal samples, to be submitted at same time or prior to procedure).

Genetically related family member for known mutation:

- Blood or DNA from blood;
- Or buccal swab (select cases only, contact lab directly to confirm if appropriate sample).

If genetic testing has been performed at a lab other than Impact Genetics, please provide copy of report.

Sample Preparation Instructions

Blood samples for DNA:

- 10 mls in yellow-topped ACD tubes **or** lavender-topped EDTA tubes (for infants 2-5 mls in pediatric or small tubes) at room temperature, to be received within 5 days after draw.

DNA from blood:

- DNA concentration—100 nanograms/microliter.
- DNA quantity—150 microliters.

Blood samples for RNA (only if requested):

- 10 mls in lavender-topped EDTA tubes (for infants 2-5 mls in pediatric or small tubes) on 4°C cool packs, to be received within 48 hours after draw.

Buccal swab:

- Kit available (select cases only contact lab directly to request); follow included instructions.

Amniocytes:

- Two T25 flasks of cultured amniotic cells **or** DNA extracted from amniotic cells at room temperature (100 nanograms/microliter: minimum 20 microliters).

Direct amnio:

- As much volume as possible in conical tube at room temperature, to be received within 48 hours after draw.

CVS:

- Send CVS tissue in sterile tissue-culture medium at room temperature **or** DNA extracted from CVS tissue at room temperature (100 nanograms/microliter: minimum 20 microliters).

Sample Identification

- Label each sample **with at least two** patient identifiers (e.g. name and date of birth).