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Information Sheet Clinical Genetics

Procedures for potentially inherited diseases

1. Take a family history

- What is known about the parents?
  Are mother and/or father affected by the same disease?
- What is known about littermates and potential additional full-siblings?
  How many animals in the litter? How many males, how many females?
  How many male / female animals are affected by the same disease?
  Were there stillborn puppies or puppies that died early and were not registered?
- Has the same disease already been observed in another relative or in an animal from the same breed?
- Is the affected animal a purebred animal? (inbreeding?)

2. If there is interest in a research project → comprehensive diagnostics (clinics, pathology) according to the phenotype (e.g. neurological, dermatological, etc.)

3. Sample acquisition

- **Mandatory:** For genetic investigations: genomic DNA
  Suitable material for the isolation of genomic DNA:
  EDTA blood, hairs with roots, native tissue samples
- **Optional:** For additional functional investigations: RNA and/or protein
  Tissue samples of the affected organ(s) in RNA-later (for RNA),
  native tissue samples at -80°C (for protein)

Genetic investigations with isolated cases are possible. However, the chances of success are much better, if DNA samples of complete families are obtained. These are the priorities (1 = high, 4 = low):

1 = affected animal
2 = parents of affected animal
3 = full siblings of affected animal
4 = other relatives or animals from the same breed

4. Requirements for samples submitted to the Institute of Genetics

- 1 – 5 ml EDTA blood (ideally ≥ 2 ml, if the animal is big enough)
- Filled and signed (by the owner) submission form
- Copy of the pedigree
- Copies of relevant clinical and diagnostic reports
  (the family history is part of the clinical report)

Link to submission form:
https://www.genetics.unibe.ch/research/sample_submission/index_eng.html