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Technical information of the Institut Central des Hôpitaux in Sion (ICH), Switzerland:

- **Genetic testing for hereditary cancer predisposition syndromes**
- **Testing for BRCA pathogenic variants in tumour tissues**

Genetic testing for hereditary cancer predisposition syndromes

- Sequencing of the 27 genes listed on orpha.net with Multiplicom's BRCA Hereditary Cancer MASTR Plus kit and FAP MASTR kit on a MiSeq instrument (Illumina). Data analysis with SophiaGenetics' DDM. Confirmation of identified pathogenic variants by Sanger sequencing.
- Detection of copy number variants (CNVs) by MLPA with MRC Holland's kits in selected genes (case-specific selection of genes). In addition, CNV detection with NGS in all requested genes with Multiplicom's MASTR Reporter software.
- Detection of familial pathogenic variants (cascade testing) by Sanger sequencing.

Testing for BRCA pathogenic variants in tumour tissues

- DNA extraction from FFPE tissues in collaboration with the Service d'Histocytopathologie (ICH). Sequencing of *BRCA1* and *BRCA2* with Multiplicom's BRCA MASTR Plus Dx kit on a MiSeq instrument (Illumina). Data analysis with SophiaGenetics' DDM.
- CNV detection: currently not possible in FFPE tissues. We recommend testing a peripheral blood sample by MLPA to exclude the presence of constitutional CNVs.

Do not hesitate to contact Dr. phil nat. Thomas von Känel (FAMH in Medical Genetics) in case of questions: thomas.vonkaenel@hopitalvs.ch / ++41 27 603 48 50.