

Familial Tumor Predispositions

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Familial Adenomatous Polyposis (FAP/MAP/PPAP) ⁺	<i>APC</i> (CDS complete + MLPA) <i>MUTYH</i> (CDS complete + MLPA) <i>POLD1</i> (CDS complete) <i>POLE</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Hereditary Non-Polyposis Colorectal Cancer (HNPCC) ⁺	<i>EPCAM</i> (CDS complete) <i>MLH1</i> (CDS complete + MLPA) <i>MSH2</i> (CDS complete + MLPA) <i>MSH6</i> (CDS complete + MLPA) <i>PMS2</i> (CDS complete + MLPA)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Juvenile Polyposis Syndrome (JPS) ⁺	<i>BMPR1A</i> (CDS complete) <i>SMAD4</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Hereditary Breast and Ovarian Cancer Syndrome (HBOC)	<i>ATM</i> (CDS komplett) ⁺ <i>BRCA1</i> (CDS komplett + MLPA) <i>BRCA2</i> (CDS komplett + MLPA) <i>BRIP1</i> (CDS komplett) ⁺ <i>CDH1</i> (CDS komplett) ⁺ <i>CHEK2</i> (CDS komplett) <i>NBN</i> (CDS komplett) ⁺ <i>PALB2</i> (CDS komplett) <i>PTEN</i> (CDS komplett) ⁺ <i>RAD51C</i> (CDS komplett) <i>RAD51D</i> (CDS komplett) ⁺ <i>STK11</i> (CDS komplett) ⁺ <i>TP53</i> (CDS komplett) ⁺	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA

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Noonan Syndrome ⁺	<i>BRAF</i> (CDS komplett) <i>KRAS</i> (CDS komplett) <i>PTPN11</i> (CDS komplett) <i>RAF1</i> (CDS komplett) <i>RIT1</i> (CDS komplett) <i>SOS1</i> (CDS komplett) <i>SPRED1</i> (CDS komplett)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Tuberous Sclerosis Complex (TSC) ⁺	<i>TSC1</i> (CDS complete) <i>TSC2</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Von Hippel-Lindau-Syndrom (VHL) ⁺	<i>VHL</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS
Wilms Tumor (Nephroblastoma) ⁺	<i>WT1</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS

Note:

According to the Genetic Diagnostics Act, every examination request for human genetic diagnostics must be accompanied by a declaration of consent from the patient or his legal representative.

Forms for this as well as request forms for the analyses are available in the download area of our homepage.