

Tumor (Keimbahn)

Mammakarzinom (Brustkrebs) [GP148] (nach EBM 11440)

1. Stufe: *BRCA1, BRCA2*,
2. Stufe: *CHEK2, PALB2, RAD51C*
3. Stufe: **umfassende Diagnostik [GP148XL]**
ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, FAM175A, MEN1, MRE11A, NBN, PALB2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53

Ovarialkarzinom (Eierstockkrebs) [GP151XL]

- BARD1, BRCA1, BRCA2, BRIP1, MLH1, MRE11A, MSH2, NBN, PALB2, PMS2, RAD50, RAD51C, RAD51D, TP53*

Fanconie Anämi [GP042] (~24 kb)

- BRCA2, FANCA, FANCC, FANCG, FANCD2, FANCE*

umfassende Diagnostik [GP042XL]

- BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4*

Gastrointestinaler Stromatumor (GIST) [GP143] (~19 kb)

- KIT, MAX, MEN1, NF1, PRKAR1A, SDHB, SDHC, SDHD, SMARCB1, TMEM127*

Gorlin-Goltz-Syndrom [GP144] (~10 kb)

- PTCH1, PTCH2, SUFU*

Hereditäres Nicht-Polypöses Kolorektales Karzinom (HNPCC) / Lynch-Syndrom [GP145] (nach EBM 11431 od. 11432)

- MLH1, MSH2, MSH6, PMS2, EPCAM (nur Deletionen)*

Kolonkarzinom [GP146XL]

- APC, BMPR1A, BUB1B, MET, CHEK2, MLH1, MSH3, MSH6, MUTYH, NBN, PMS1, PMS2, PTEN, SMAD4, STK11, TP53*

Magenkarzinom [GP147] (~23 kb)

- BMPR1A, CDH1, CHEK2, MLH1, MSH2, MSH6, PMS2, STK11, TP53*

Melanom, familiär [GP149] (~9,5 kb)

- CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, RB1*

Nierenkarzinom [GP150XL]

- CHEK2, EPCAM (nur Deletionen), FH, FLCN, GPC3, HRPT2, MET, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1*

Pankreaskarzinom (inkl. chronischer Pankreatitis) [GP152XL]

- APC, ATM, BRCA1, BRCA2, CDK4, CDKN2A, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK1, STK11, TP53*

Phäochromozytom-Paragangliom-Syndrom [GP153] (~19 kb)

- MAX, MEN1, NF1, PRKAR1A, RET, SDHB, SDHC, SDHD, TMEM127, VHL*

Polyposis Coli [GP157XL]

- APC, BMPR1A, CHEK2, FLCN, MSH3, MUTYH, NTHL1, POLD, POLE, PTEN, SMAD4, STK11*

Schilddrüsenkarzinom [GP154] (~6 kb)

- PTEN, RET, SDHB, SDHC, SDHD*

Umfassende Diagnostik erblicher Tumorerkrankungen [GP155XL]

- APC, AR, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BTNL2, BUB1B, CDH1, CDK4, CDKN2A, CHEK2, ELAC2, FH, FLCN, GPC3, HOXB13, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MRE11A, MSH2, MSH6, MSR1, MUTYH, NBN, NF1, PALB2, PMS1, PMS2, PRKAR1A, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, RNASEL, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, TMEM127, TP53, TSC1, TSC2, VHL, WT1, ZFH3*