

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
AAGAB	NGS, Sanger	945										
ABCA12	NGS, Sanger	7788										
ABCA4	NGS, Sanger	6822										
ABCC9	NGS	4647										
ABHD5	NGS, Sanger	1050										
ACTA2	NGS	1131										
ACTC1	NGS	1131										
ADAM9	NGS	2460										
ADAMTS2	NGS	3633										
ADAMTSL4	NGS	3291										
AIP1	NGS	1155										
AKAP9	NGS	11730										
AKT1	Sanger	1440										
ALDH3A2	NGS, Sanger	1527										
ALOX12B	NGS, Sanger	2106										
ALOXE3	NGS, Sanger	2604										
ANK2	NGS	11871										
AP1S1	NGS, Sanger	574										
APC	NGS	8529										
AQP2	Sanger	813										
AQP5	NGS, Sanger	795										
ARFGEF2	NGS, Sanger	5355										
ARHGAP31	NGS	4335										
ARX	Sanger	1686										
ASPM	NGS, Sanger	10431										
ATGL	Sanger	1512										
ATM	NGS	9168										
ATN1	Sanger	3570										
ATP2A2	NGS	3129										
AVP	Sanger	492										
AVPR2	Sanger	1113										
B3GALT6	NGS	987										
B4GALT7	NGS	981										
BARD1	NGS	2331										
BEST1	NGS	1812										
BLM	NGS	4251										
BRCA1	NGS, Sanger	5589										
BRCA2	NGS, Sanger	10254										
BRIP1	NGS	3747										
C16ORF57	NGS	795										
C7ORF11	NGS	540										
CACNA1C	NGS	6558										
CACNA1F	NGS	5931										
CACNA2D1	NGS	3273										
CACNB2	NGS	1839										
CALM1	NGS	453										
CALM2	NGS	450										
CARD14	NGS, Sanger	3015										
CASQ2	NGS	1200										
CAST	NGS	2250										
CAV3	NGS	456										
CCM2*	Sanger	1332										
CD151	NGS	762										
CDH1	NGS, Sanger	2646										
CDK5RAP2	Sanger	5679										
CDK6	Sanger	978										
CDKN2A	Sanger	396										
CDSN	NGS	1590										
CENPJ	Sanger	4014										
CEP135	Sanger	3420										
CEP152	Sanger	5130										
CEP290	NGS	7440										
CERKL	NGS	1677										
CERS3	NGS, Sanger	1152										
CFTR	Sanger	4440										
CHEK2	NGS, Sanger	1758										
CHM	NGS, Sanger	1962										
CHST14	NGS	1128										
CHST6	NGS	1188										
CHST8	NGS	1275										
CLDN1	NGS, Sanger	636										
CNGA1	NGS	2280										
CNGA3	NGS	2097										
CNGB3	NGS, Sanger	2430										
COG6	NGS	1971										
COL14A1	NGS	5388										

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
COL16A1	NGS	4815										
COL17A1	NGS	4491										
COL1A1	NGS	4392										
COL1A2	NGS	4098										
COL23A1	NGS	1623										
COL3A1	NGS	4398										
COL5A1	NGS	5514										
COL5A2	NGS	4497										
COL7A1	NGS	8832										
COL8A2	NGS	2112										
CRB1	NGS	4221										
CRX	NGS	897										
CSTA	NGS	294										
CTGF	NGS	1050										
CTNNA3	NGS	2688										
CTSC	NGS, Sanger	1392										
CYLD	Sanger	2868										
CYP4F22	NGS, Sanger	1596										
DCN	NGS	1080										
DCX	Sanger	1080										
DES	NGS	1413										
DJ1 (PARK7)	Sanger	1743										
DPP6	NGS	2598										
DSC2	NGS	2544										
DSC3	NGS	2688										
DSE	NGS	2874										
DSG1	NGS, Sanger	3150										
DSG2	NGS	3357										
DSG3	NGS	2997										
DSG4	NGS	3180										
DSP	NGS, Sanger	8616										
DST	NGS	16611										
DYT1 (TOR1A)	NGS	999										
DYT11 (SGCE)	NGS	1416										
DYT12 (ATP1A3)	NGS	3081										
DYT23 (CIZ1)	NGS	2865										
DYT24 (ANO3)	NGS	2946										
DYT25 (GNAL)	NGS	1377										
DYT4 (TUBB4A)	NGS	1335										
DYT5a (GCH1)	NGS	753										
DYT5b (TH)	NGS	1587										
DYT6 (THAP1)	NGS	642										
EBP	NGS	963										
EDA	NGS	1176										
EDAR	NGS	4113										
EFEMP2	NGS	1329										
ELOVL4	NGS, Sanger	945										
ENPP1	NGS, Sanger	2775										
EPCAM	NGS	-										
ERCC2	NGS	2283										
ERCC3	NGS	2349										
EXPH5	NGS	5967										
EYS	NGS	9498										
FAH	Sanger	1257										
FAM175A	NGS	1227										
FBLN5	NGS	1467										
FBN1	NGS	8613										
FBN2	NGS	8739										
FERMT1	NGS	2034										
FKBP14	NGS	633										
FKRP	Sanger	1485										
FKTN	Sanger	1383										
FLCN	Sanger	1737										
FLG	NGS	12186										
FLII	NGS	3807										
FLNA	NGS, Sanger	7917										
FOXG1	Sanger	1467										
GBA	NGS, Sanger	1608										
GCH1	Sanger	750										
GJA1	NGS, Sanger	1149										
GJA5	NGS	1077										
GJB2	NGS, Sanger	681										
GJB3	NGS, Sanger	813										
GJB4	NGS, Sanger	801										
GJB6	NGS, Sanger	786										
GLMN	Sanger	1782										

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
GNA11	NGS	1080										
GNAQ	NGS, Sanger	1080										
GNAT1	NGS	1050										
GNAT2	NGS	1062										
GPD1L	NGS	1056										
GPR56	Sanger	2079										
GRHL2	NGS	1875										
GRIP1	NGS	3231										
GSN	NGS	2349										
GTF2H5	NGS	213										
GUCY2D	NGS	3312										
HCN4	NGS	3612										
HOXB1	NGS, Sanger	903										
HPD	Sanger	1062										
HTT	FA											
IL36RN	NGS, Sanger	468										
ILK	NGS	1356										
IMPDH1	NGS	1770										
IMPG1	NGS	2391										
ITGA2	NGS	3543										
ITGA3	NGS	3153										
ITGA5	NGS	3147										
ITGA6	NGS	3219										
ITGB1	NGS	2397										
ITGB4	NGS	5466										
JUP	NGS	2238										
KANK2	NGS	2553										
KCNA5	NGS	1842										
KCND3	NGS	1968										
KCNE1	NGS	390										
KCNE2	NGS	372										
KCNE3	NGS	312										
KCNE5	NGS	429										
KCNJ2	NGS	1284										
KCNJ5	NGS	1260										
KCNJ8	NGS	1275										
KCNQ1	NGS	2031										
KCNV2	NGS	1638										
KLHL24	NGS	1803										
KLHL7	NGS	1761										
KRT1	NGS, Sanger	1935										
KRT10	NGS, Sanger	1755										
KRT12	NGS	1485										
KRT13	NGS, Sanger	1377										
KRT14	NGS, Sanger	1416										
KRT16	NGS, Sanger	1419										
KRT17	NGS, Sanger	1296										
KRT2	NGS, Sanger	1920										
KRT3	NGS	1887										
KRT4	NGS, Sanger	1785										
KRT5	NGS, Sanger	1773										
KRT6A	NGS, Sanger	1692										
KRT6B	NGS	1692										
KRT6C	NGS	1692										
KRT81	NGS, Sanger	1518										
KRT83	NGS, Sanger	1482										
KRT86	NGS, Sanger	1461										
KRT9	NGS, Sanger	1872										
LAMA3	NGS	9999										
LAMA5	NGS	11088										
LAMB1	NGS	5361										
LAMB3	NGS	3516										
LAMC1	NGS	4827										
LAMC2	NGS	3579										
LARGE1	Sanger	2268										
LCA5	NGS	2091										
LDB3	NGS	2196										
LIPN	NGS	1194										
LIS1	Sanger	1230										
LIS1 (PAFAH1B1)	NGS	1233										
LIS2 (RELN)	NGS	10383										
LIS3 (TUBA1A)	NGS	1356										
LIS4 (NDE1)	NGS	1008										
LISX1 (DCX)	NGS	1326										
LISX2 (ARX)	NGS	1689										
LMNA	NGS	1955										

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
LOR	NGS, Sanger	939										
LRIT3	NGS	2037										
LRRK2	Sanger	7581										
MAT2A	NGS	1185										
MBTPS2	NGS, Sanger	1560										
MCPH1	Sanger	2565										
MED12	NGS	6531										
MEN1	NGS	1845										
MFAP5	NGS	519										
MFN2	NGS, Sanger	2274										
MLH1	NGS	2268										
MMP1	NGS	1410										
MPZ	Sanger	747										
MRE11A	NGS	2124										
MSH2	NGS	2802										
MSH6	NGS	4080										
MT-ND1	Sanger	954										
MT-ND4	Sanger	1377										
MT-ND4L	Sanger	294										
MT-ND6	Sanger	522										
MT-TS1	NGS	207										
MUTYH	NGS	1605										
MYBPC3	NGS, Sanger	3825										
MYH11	NGS	5937										
MYH7	NGS, Sanger	5808										
MYL2	NGS	459										
MYLK	NGS	5742										
NBN	NGS	2262										
NCSTN	Sanger	2130										
NID1	NGS	3744										
NID2	NGS	4128										
NIPAL4	NGS, Sanger	1401										
NKX2-5	NGS	975										
NLRP1	NGS	4125										
NMNAT1	NGS	837										
NOS1AP	NGS	1521										
NPPA	NGS	459										
NR2E3	NGS	1233										
NSDHL	NGS, Sanger	1122										
NYX	NGS	1443										
OAT	NGS	1317										
OFD1	Sanger	3036										
OPA1	NGS, Sanger	3048										
OPA3	NGS, Sanger	543										
PALB2	NGS, Sanger	3558										
PARK1/4 (SNCA)	NGS	423										
PARK17 (VPS35)	NGS	2391										
PARK18 (EIF4G1)	NGS	4821										
PARK19 (DNAJC6)	NGS	2913										
PARK2 (PRKN)	NGS	1398										
PARK20 (SYNJ1)	NGS	4053										
PARK6 (PINK1)	NGS	1746										
PARK7 (DJ1)	NGS	570										
PARK8 (LRRK2)	NGS	7584										
PDCD10*	Sanger	636										
PDE6A	NGS	2583										
PDE6B	NGS	2565										
PDE6C	NGS	2577										
PDE6H	NGS	249										
PHC1	Sanger	3012										
PHYH	NGS	1017										
PIK3CA	Sanger	3204										
PIKFYVE	NGS	6297										
PINK1	Sanger	1743										
PKP1	NGS	2241										
PKP2	NGS	2514										
PLEC	NGS	14052										
PLN	NGS	159										
PLOD1	NGS	2181										
PMP22	Sanger	483										
PMS2	NGS	2586										
PNKP	Sanger	1563										
PNPLA1	NGS, Sanger	1602										
POFUT1	Sanger	1164										
POGLUT1	Sanger	1176										
POMGNT1	Sanger	2244										

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
POMP	NGS, Sanger	426										
POMT1	Sanger	2175										
POMT2	Sanger	2250										
PORCN	Sanger	1365										
PRDM16	NGS	3828										
PRKAG2	NGS	1710										
PRKG1	NGS	2058										
PRKN	Sanger	1395										
PROM1	NGS, Sanger	2598										
PRPF31	NGS, Sanger	1500										
PRPF8	NGS, Sanger	7008										
PRPH2	NGS, Sanger	1041										
PSEN1	Sanger	1404										
PSENN	Sanger	306										
PTEN	NGS, Sanger	1209										
RAB18	NGS, Sanger	546										
RAB3GAP1	NGS, Sanger	2943										
RAB3GAP2	NGS, Sanger	4179										
RAD50	NGS	3936										
RAD51C	NGS, Sanger	1128										
RAD51D	NGS, Sanger	984										
RANGRF	NGS	558										
RASA1	Sanger	2610										
RDH5	NGS	957										
RECQL4	Sanger	3624										
RHBDF2	NGS	2481										
RHO	NGS, Sanger	1047										
RP1	NGS, Sanger	6471										
RP2	NGS	1053										
RPE65	NGS	1602										
RPGR	NGS	3459										
RPGRIP1	NGS	3861										
RSP01	NGS	789										
RYR2	NGS	14904										
SCA1	FA											
SCA17	FA											
SCA2	FA											
SCA3	FA											
SCA6	FA											
SCA7	FA											
SCN10A	NGS	5871										
SCN1B	NGS	807										
SCN2B	NGS	648										
SCN3B	NGS	648										
SCN4B	NGS	687										
SCN5A	NGS	6051										
SDR9C7	NGS	942										
SERPINB7	NGS, Sanger	1140										
SGCE	Sanger	1311										
SKI	NGS	2184										
SLC27A4	NGS, Sanger	1932										
SLC2A10	NGS	1623										
SLC39A13	NGS	1113										
SLC4A11	NGS	2676										
SLMAP	NGS	2487										
SLURP1	NGS, Sanger	309										
SMAD3	NGS	1275										
SNAP29	NGS	777										
SNCA	Sanger	336										
SNTA1	NGS	1518										
SOX18	NGS	1152										
SOX7	NGS	1167										
SOX9	Sanger	1527										
SPINK5	NGS, Sanger	3285										
SPR	NGS, Sanger	783										
SRD5A3	NGS	954										
ST14	NGS	2568										
STIL	Sanger	3861										
STK11	NGS, Sanger	1299										
STS	NGS, Sanger	1752										
SUMF1	NGS	1281										
TACSTD2	NGS	972										
TAT	Sanger	1362										
TAZ	NGS	876										
TBR2 (EOMES)	NGS	2118										
TEK	Sanger	3372										

Gen	Methode	Größe bp	Auge	Haut g	Haut k	Kardio	Tumor	PPK	EB	FAP	NEURO	MARFAN
TGFB2	NGS	1326										
TGFB3	NGS	1239										
TGFB1	NGS	2049										
TGFBR1	NGS	1521										
TGFBR2	NGS	1776										
TGM1	NGS, Sanger	2454										
TGM5	NGS, Sanger	2163										
TH	Sanger	1584										
THAP1	Sanger	639										
TMEM43	NGS	1203										
TNNI3	NGS, Sanger	633										
TNNT2	NGS, Sanger	876										
TNXB	NGS	12732										
TOR1A	Sanger	996										
TP53	NGS, Sanger	1023										
TP63	NGS	2043										
TPM1	NGS	822										
TRDN	NGS	2190										
TRPM1	NGS	4926										
TRPM4	NGS	3645										
TRPV3	NGS, Sanger	2376										
TUBA1A	Sanger	1353										
TUBA8	Sanger	1347										
TUBB2B	Sanger	1335										
TUBB3	NGS, Sanger	1134										
TUBB3 (CDCBM1)	NGS	1353										
UBIAD1	NGS	1017										
UPF3B	NGS	1449										
USH2A	NGS	15609										
VEGFR (KDR)	Sanger	4068										
VEGFR1 (FLT1)	Sanger	4014										
VEGFR3 (FLT4)	Sanger	4089										
VHL	Sanger	642										
VLDLR	NGS, Sanger	2619										
VPS33B	NGS	1854										
VPS35	Sanger	2388										
WDR62	NGS, Sanger	4569										
WNT10A	NGS, Sanger	1254										
XRCC2	NGS	840										
ZDHHC9	NGS	1092										
ZEB1	NGS	3378										