

## Haut- und Bindegeweberkrankungen

**Albinismus, okulokutaner [GP168] (~10 kb)**

*MC1R, OCA2, SLC24A5, SLC45A2 (MATP), TYR, TYRP1*

**Aneurysmen, Aortenfehlbildungen (TAAD) [GP037] (nach EBM 11448)**

1. *COL3A1*, 2. *ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, NOTCH1, SKI, SLC2A10, SMAD3, SMAD6, TGFB1, TGFB2, TGFB3, TGFB1, TGFB2*

**Ehlers-Danlos-Syndrom (EDS) [GP176] (~25 kb ohne *TNXB*)**

*ADAMTS2, COL5A1, COL5A2, COL1A1, COL1A2, PLOD1, TNXB (12.7kb) (COL3A1 nach EBM)*

**Marfan-Syndrom [GP044] (EBM11445, 11446, 11448)**

1. *FBN1*, 2. *TGFB1, TGFB2*  
ggfs. *ACTA2, FBN1, MYH11, MYLK, SMAD3, TGFB2*

**Cutis Laxa [GP161] (~25 kb)**

*ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1*

**Epidermolysis bullosa (EB) [GP041XL, 28,8kb]**

*COL17A1, COL7A1, KRT14, KRT5, LAMA3, LAMB3, LAMC2*

**Ichthyose, autosomal rezessiv (ARCI) [GP043] (~21 kb)**

1. *TGM1* 2. *ABCA12, ABHD5, ALOX12B, ALOXE3, CERS3, CYP4F22, NIPAL4, PNPLA1*

**Genodermatosen, umfassende Diagnostik [GP163XL]**

*AAGAB, ABCA12, ABHD5, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, CERS3, CLDN1, CYLD, CYP4F22, DSG, DSG1, DSP, ENPP1, SLC27A4 (FATP4), GJA1, GJB3, GJB4, IL36RN, KRT1, KRT10, KRT14, KRT16, KRT16, KRT17, KRT5, KRT9, LOR, MBTPS2, NIPAL4, PNPLA1, POMP, PORCN, RASA1, RECQL4, SLURP1, SPINK5, STS, TGM1, TGM5, TRPV3*

**Dyskeratosis congenita (DC, DKC) [GP038] (~16 kb)**

*CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, WRAP53*

**Ektodermale Dysplasie (ED) [GP040XL]**

*ANTXR1, APCDD1, AXIN2, BANF1, BCS1L, CDH3, CDSN, CTSC, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, FGF10, FGFR2, FGFR3, GJA1, GJB6, GRHL2, GTF2H5, HR, IFT122, IFT43, KRT14, KRT74, LIPH, LPAR6, MPLKIP, MSX1, NFKBIA, OFD1, PIGL, PKP1, PORCN, PVRL1, PVRL4, SHOC2, SOX18, ST14, TP63, TRPS1, TWIST2, UBR1, WDR19, WDR35, WNT10A*

**Stickler-Syndrom [GP013] (~22 kb)**

*COL11A1, COL2A1, COL9A2, COL9A3, COL11A2, COL9A1*

**Fanconi Anämie [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*