

Entwicklungsverzögerung, Intelligenzminderung

Bardet-Biedl-Syndrom [GP003] (~ 22 kb)

ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, MKKS, MKS1, TTC8

umfassende Diagnostik [GP003XL]

ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

Cornelia-de-Lange-Syndrom (CdLS) [GP078] (~19 kb)

1. NIPBL, 2. SMC1A, SMC3, RAD21, HDAC8

Dravet-Syndrom [GP016] (~21 kb)

1. SCN1A, 2. GABRG2, SCN2A, SCN9A, STXBP1

Glykosylierungsstörungen, kongenitale (CDG) [GP079] (~22 kb)

1. PMM2, 2. MPI, ALG6, 3. ALG1, ALG12, ALG2, ALG8, ALG9, DOLK, DPAGT1, DPM1, MAGT1, MPDU1, RFT1 SRD5A3, TMEM165, TUSC3

Joubert-Syndrom [GP004] (~25 kb)

AHI1, CC2D2A, CEP290, NPHP RPGRIP1L, TMEM67

umfassende Diagnostik [GP004XL]

AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

Zellweger-Syndrom [GP035] (~18 kb)

1. PEX1, 2. PEX6, 3. ABCD3, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5