| *Gen-symbol* | *Genname* | Erkrankung(en) | OMIM | Gengröße (kb) |
|---|---|---|---|---|
| *ABCC6* | *ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 6* | Arterial calcification, generalized, of infancy, 2 | 614473 | 4,5 |
| | | Pseudoxanthoma elasticum | 264800 | |
| | | Pseudoxanthoma elasticum, forme fruste | 177850 | |
| *ACSL4* | *ACYL-CoA SYNTHETASE LONG CHAIN FAMILY, MEMBER 4* | Mental retardation, X-linked 63 | 300387 | 2,1 |
| *AFF2* | *AF4/FMR2 FAMILY, MEMBER 2 (FMR2)* | Mental retardation, X-linked, FRAXE type | 309548 | 3,9 |
| *AIFM1* | *APOPTOSIS-INDUCING FACTOR, MITOCHONDRION-ASSOCIATED, 1 (PDCD8)* | Combined oxidative phosphorylation deficiency 6 | 300816 | 1,8 |
| | | Cowchock syndrome | 310490 | |
| *AKT1* | *V-AKT MURINE THYMOMA VIRAL ONCOGENE HOMOLOG 1* | Proteus syndrome, somatic | 176920 | 1,4 |
| | | Cowden syndrome 6 | 615109 | |
| *AKT3* | *V-AKT MURINE THYMOMA VIRAL ONCOGENE HOMOLOG 3 (PKBG)* | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2 | 615937 | 1,4 |
| *AP1S2* | *ADAPTOR-RELATED PROTEIN COMPLEX 1, SIGMA-2 SUBUNIT* | Mental retardation, X-linked syndromic 5 | 304340 | 0,5 |
| *ARHGEF6* | *RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 6 (PIXA)* | Mental retardation, X-linked 46 | 300436 | 2,3 |
| *ARHGEF9* | *RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 9 (PEM2)* | Epileptic encephalopathy, early infantile, 8 | 300607 | 1,6 |
| *ARID1A* | *AT-RICH INTERACTION DOMAIN-CONTAINING PROTEIN 1A (SMARCF1)* | Coffin-Siris syndrome 2 | 614607 | 6,9 |
| *ARID1B* | *AT-RICH INTERACTION DOMAIN-CONTAINING PROTEIN 1B* | Coffin-Siris syndrome 1 | 135900 | 6,7 |
| *ARX* | *ARISTALESS-RELATED HOMEOBOX, X-LINKED* | Epileptic encephalopathy, early infantile, 1 | 308350 | 1,7 |
| | | Hydranencephaly with abnormal genitalia | 300215 | |
| | | Lissencephaly, X-linked 2 | 300215 | |
| | | Mental retardation, X-linked 29 and others | 300419 | |
| | | Partington syndrome | 309510 | |
| | | Proud syndrome | 300004 | |
| *ASPA* | *ASPARTOACYLASE* | Canavan disease | 271900 | 0,9 |
| *ATP6AP2* | *ATPase, H+ TRANSPORTING, LYSOSOMAL, ACCESSORY PROTEIN 2* | ?Mental retardation, X-linked, syndromic, Hedera type | 300423 | 1,1 |
| *ATP7A* | *ATPase, Cu(2+)-TRANSPORTING, ALPHA POLYPEPTIDE* | Menkes disease | 309400 | 4,5 |
| | | Occipital horn syndrome | 304150 | |
| | | Spinal muscular atrophy, distal, X-linked 3 | 300489 | |
| *ATRX* | *ATR-X GENE; HELICASE 2, X-LINKED (XH2)* | Alpha-thalassemia/mental retardation syndrome | 301040 | 7,5 |
| | | Mental retardation-hypotonic facies syndrome, X-linked | 309580 | |
| *BCOR* | *BCL6 COREPRESSOR* | Microphthalmia, syndromic 2 | 300166 | 5,2 |
| *BRAF* | *V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1* | Cardiofaciocutaneous syndrome | 115150 | 2,3 |
| | | LEOPARD syndrome 3 | 613707 | |
| | | Noonan syndrome 7 | 613706 | |

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|---|---|---|---|---|
| *BRWD3* | *BROMODOMAIN- AND WD REPEAT-CONTAINING PROTEIN 3* | Mental retardation, X-linked 93 | 300659 | 5,4 |
| *CASK* | *CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE (LIN2)* | FG syndrome 4 | 300422 | 2,8 |
| | | Mental retardation and microcephaly with pontine and cerebellar hypoplasia | 300749 | |
| | | Mental retardation, with or without nystagmus | 300422 | |
| *CBL* | *CAS-BR-M MURINE ECOTROPIC RETROVIRAL TRANSFORMING SEQUENCE HOMOLOG* | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia | 613563 | 2,7 |
| *CCND2* | *CYCLIN D2* | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 | 615938 | 0,8 |
| *CDKL5* | *CYCLIN-DEPENDENT KINASE-LIKE 5 (STK9)* | epileptic encephalopathy, early infantile, 2 | 300672 | 3,1 |
| *CDKN1C* | *CYCLIN-DEPENDENT KINASE INHIBITOR 1C (KIP2)* | Beckwith-Wiedemann syndrome | 130650 | 0,9 |
| | | IMAGE syndrome | 614732 | |
| *CHD7* | *CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 7* | Charge syndrome | 214800 | 9,0 |
| | | Hypogonadotropic hypogonadism 5 with or without anosmia | 612370 | |
| *CNTNAP2* | *CONTACTIN-ASSOCIATED PROTEIN-LIKE 2; Neurexin 4* | Cortical dysplasia-focal epilepsy syndrome | 610042 | 4,0 |
| | | Pitt-Hopkins like syndrome 1 | 610042 | |
| *CUL4B* | *CULLIN 4B* | Mental retardation, X-linked, syndromic 15 (Cabezas type) | 300354 | 2,7 |
| *DCX* | *DOUBLECORTIN* | Lissencephaly, X-linked | 300067 | 1,3 |
| | | Subcortical laminal heteropia, X-linked | 300067 | |
| *DIS3L2* | *DIS3 MITOTIC CONTROL, S. CEREVISIAE, HOMOLOG-LIKE 2* | Perlman syndrome | 267000 | 2,7 |
| *DKC1* | *DYSKERIN* | Dyskeratosis congenita, X-linked | 305000 | 1,5 |
| *DLG3* | *DISCS LARGE, DROSOPHILA, HOMOLOG OF, 3* | Mental retardation, X-linked 90 | 300850 | 2,5 |
| *DNMT3A* | *DNA METHYLTRANSFERASE 3A* | Tatton-Brown-Rahman syndrome | 615879 | 2,2 |
| *DYRK1A* | *DUAL-SPECIFICITY TYROSINE PHOSPHORYLATION-REGULATED KINASE 1A* | Mental retardation, autosomal dominant 7 | 614104 | 2,3 |
| *EHMT1* | *EUCHROMATIC HISTONE METHYLTRANSFERASE 1* | Kleefstra syndrome | 610253 | 2,4 |
| *EIF2B5* | *EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 5* | Leukoencephalopathy with vanishing white matter | 603896 | 2,2 |
| | | Ovarioleukodystrophy | 603896 | |
| *EZH2* | *ENHANCER OF ZESTE, DROSOPHILA, HOMOLOG 2, ENX1* | Weaver syndrome | 277590 | 2,2 |
| *FANCB* | *FANCB GENE* | Fanconi anemia, complementation group B | 300514 | 2,6 |
| *FGD1* | *FYVE, RhoGEF, AND PH DOMAIN-CONTAINING PROTEIN 1* | Aarskog-Scott syndrome | 305400 | 2,9 |
| | | Mental retardation, X-linked syndromic 16 | 305400 | |

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|---|---|---|---|---|
| *FLNA* | *FILAMIN A* | FG syndrome 2 | 300321 | 7,9 |
| | | Frontometaphyseal dysplasia | 305620 | |
| | | Heterotopia, periventricular | 300049 | |
| | | Melnick-Needles syndrome | 309350 | |
| | | Otopalatodigital syndrome, type I | 311300 | |
| | | Otopalatodigital syndrome, type II | 304120 | |
| | | Terminal osseous dysplasia | 300244 | |
| *FMR1* | *FMR1 GENE; FRAGILE X MENTAL RETARDATION PROTEIN; FMRP* | Fragile X syndrome | 300624 | 1,9 |
| | | Fragile X tremor/ataxia syndrome | 300623 | |
| *FOXG1* | *FORKHEAD BOX G1* | Rett syndrome, congenital variant | 613454 | 1,5 |
| *FTSJ1* | *FTSJ HOMOLOG 1* | Mental retardation, X-linked 9 | 309549 | 1,0 |
| *GDI1* | *GDP DISSOCIATION INHIBITOR 1* | Mental retardation, X-linked 41 | 300849 | 1,3 |
| *GFAP* | *GLIAL FIBRILLARY ACIDIC PROTEIN* | Alexander disease | 203450 | 1,3 |
| *GK* | *GLYCEROL KINASE* | Glycerol kinase deficiency | 307030 | 1,6 |
| *GLI3* | *GLI-KRUPPEL FAMILY MEMBER 3* | Greig cephalopolysyndactyly syndrome | 175700 | 4,7 |
| | | Pallister-Hall syndrome | 146510 | |
| | | Polydactyly, postaxial, types A1 and B | 174200 | |
| | | Polydactyly, preaxial, type IV | 174700 | |
| *GNAQ* | *GUANINE NUCLEOTIDE-BINDING PROTEIN, Q POLYPEPTIDE* | Capillary malformations, congenital, 1, somatic, mosaic | 163000 | 1,1 |
| | | Sturge-Weber syndrome, somatic, mosaic | 185300 | |
| *GPC3* | *GLYPICAN 3* | Simpson-Golabi-Behmel syndrome, type 1 | 312870 | 1,7 |
| *GPSM2* | *G PROTEIN SIGNALING MODULATOR 2* | Chudley-McCullough syndrome | 604213 | 2,1 |
| *GRIA3* | *GLUTAMATE RECEPTOR, IONOTROPIC, AMPA 3* | Mental retardation, X-linked 94 | 300699 | 2,7 |
| *HCCS* | *HOLOCYTOCHROME C SYNTHASE* | Linear skin defects with multiple congenital anomalies 1 | 309801 | 0,8 |
| *HCFC1* | *HOST CELL FACTOR C1* | Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ) | 309541 | 6,1 |
| *HDAC8* | *HISTONE DEACETYLASE 8* | Cornelia de Lange syndrome 5 | 300882 | 1,1 |
| | | Wilson-Turner syndrome | 309585 | |
| *HEPACAM* | *HEPATOCYTE CELL ADHESION MOLECULE* | Megalencephalic leukoencephalopathy with subcortical cysts 2A | 613925 | 1,3 |
| | | Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation | 613926 | |
| *HERC2* | *HECT DOMAIN AND RCC1-LIKE DOMAIN 2* | Mental retardation, autosomal recessive 38 | 615516 | 14,5 |

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|---|---|---|---|---|
| *HPRT1* | *HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1* | Lesch-Nyhan syndrome | 300322 | 0,7 |
| *HRAS* | *V-HA-RAS HARVEY RAT SARCOMA VIRAL ONCOGENE HOMOLOG* | Costello syndrome | 218040 | 0,6 |
| | | Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic | 163200 | |
| *HSD17B10* | *17-BETA-HYDROXYSTEROID DEHYDROGENASE X* | 17-beta-hydroxysteroid dehydrogenase X deficiency | 300438 | 0,8 |
| | | ?Mental retardation, X-linked syndromic 10 | 300220 | |
| *HUWE1* | *HECT, UBA, AND WWE DOMAINS-CONTAINING PROTEIN 1* | Mental retardation, X-linked syndromic, Turner type | 300706 | 13,1 |
| *IDS* | *IDURONATE 2-SULFATASE* | Mucopolysaccharidosis II | 309900 | 1,7 |
| *IGBP1* | *IMMUNOGLOBULIN-BINDING PROTEIN 1* | Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia | 300472 | 1,0 |
| *IKBKG* | INHIBITOR OF KAPPA LIGHT POLYPEPTIDE GENE ENHANCER IN B CELLS, KINASE OF, GAMMA | Ectodermal dysplasia, hypohidrotic, with immune deficiency | 300291 | 1,3 |
| | | Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency | 300301 | |
| | | Immunodeficiency 33 | 300636 | |
| | | Incontinentia pigmenti | 308300 | |
| *IL1RAPL1* | *INTERLEUKIN 1 RECEPTOR ACCESSORY PROTEIN-LIKE 1* | Mental retardation, X-linked 21/34 | 300143 | 2,1 |
| *IQSEC2* | *IQ MOTIF- AND SEC7 DOMAIN-CONTAINING PROTEIN 2* | Mental retardation, X-linked 1/78 | 309530 | 2,9 |
| *KDM5C* | *LYSINE-SPECIFIC DEMETHYLASE 5C* | Mental retardation, X-linked, syndromic, Claes-Jensen type | 300534 | 4,7 |
| *KDM6A* | *LYSINE-SPECIFIC DEMETHYLASE 6A* | Kabuki syndrome 2 | 300867 | 4,2 |
| *KIAA0196* | *KIAA0196 GENE; Strumpellin* | Ritscher-Schinzel syndrome 1 | 220210 | 3,5 |
| | | Spastic paraplegia 8, autosomal dominant | 603563 | |
| *KIAA2022* | *KIAA2022 GENE* | Mental retardation, X-linked 98 | 300912 | 4,6 |
| *KIF7* | KINESIN FAMILY MEMBER 7 | Acrocallosal syndrome | 200990 | 4,0 |
| | | Joubert syndrome 12 | 200990 | |
| | | ?Hydrolethalus syndrome 2 | 614120 | |
| *KMT2D* | *LYSINE-SPECIFIC METHYLTRANSFERASE 2D* | Kabuki syndrome 1 | 147920 | 16,6 |
| *KPTN* | *KAPTIN* | Mental retardation, autosomal recessive 41 | 615637 | 1,3 |
| *KRAS* | *V-KI-RAS2 KIRSTEN RAT SARCOMA VIRAL ONCOGENE HOMOLOG* | Cardiofaciocutaneous syndrome 2 | 615278 | 0,6 |
| | | Noonan syndrome 3 | 609942 | |
| | | RAS-associated autoimmune leukoproliferative disorder | 614470 | |
| | | Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic | 163200 | |

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|---|---|---|---|---|
| *L1CAM* | *L1 CELL ADHESION MOLECULE; MIC5* | Corpus callosum, partial agenesis of | 304100 | 3,8 |
| | | CRASH syndrome | 303350 | |
| | | Hydrocephalus due to aqueductal stenosis | 307000 | |
| | | Hydrocephalus with congenital idiopathic intestinal pseudoobstruction | 307000 | |
| | | Hydrocephalus with Hirschsprung disease | 307000 | |
| | | MASA syndrome | 303350 | |
| *LAMP2* | *LYSOSOME-ASSOCIATED MEMBRANE PROTEIN 2* | Danon disease | 300257 | 1,2 |
| *MAGT1* | *MAGNESIUM TRANSPORTER 1* | Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia | 300853 | 1,1 |
| *MAOA* | *MONOAMINE OXIDASE A* | Brunner syndrome | 300615 | 1,6 |
| *MAP2K1* | *MITOGEN-ACTIVATED PROTEIN KINASE KINASE 1* | Cardiofaciocutaneous syndrome 3 | 615279 | 1,2 |
| *MAP2K2* | *MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2* | Cardiofaciocutaneous syndrome 4 | 615280 | 1,2 |
| *MBD5* | *METHYL-CpG-BINDING DOMAIN PROTEIN 5* | Mental retardation, autosomal dominant 1 | 156200 | 4,5 |
| *MBTPS2* | *MEMBRANE-BOUND TRANSCRIPTION FACTOR PROTEASE, SITE 2* | ?Olmsted syndrome, X-linked | 300918 | 1,6 |
| | | IFAP syndrome with or without BRESHECK syndrome | 308205 | |
| | | Keratosis follicularis spinulosa decalvans, X-linked | 308800 | |
| *MECP2* | *METHYL-CpG-BINDING PROTEIN 2* | Encephalopathy, neonatal severe | 300673 | 1,5 |
| | | Mental retardation, X-linked, syndromic 13 | 300055 | |
| | | Rett syndrome | 312750 | |
| | | Mental retardation, X-linked syndromic, Lubs type | 300260 | |
| *MED12* | *MEDIATOR COMPLEX SUBUNIT 12* | Lujan-Fryns syndrome | 309520 | 6,5 |
| | | Ohdo syndrome, X-linked | 300895 | |
| | | Opitz-Kaveggia syndrome | 305450 | |
| *MEF2C* | *MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE C* | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations | 613443 | 1,4 |
| *MID1* | *MIDLINE 1* | Opitz GBBB syndrome, type I | 300000 | 2,0 |
| *MLC1* | *MLC1 GENE* | Megalencephalic leukoencephalopathy with subcortical cysts | 604004 | 1,1 |
| *NAA10* | *N-ALPHA-ACETYLTRANSFERASE 10, NatA CATALYTIC SUBUNIT* | ?Microphthalmia, syndromic 1 | 309800 | 0,7 |
| | | Ogden syndrome | 300855 | |
| *NDP* | *NDP GENE; Norrin* | Exudative vitreoretinopathy 2, X-linked | 305390 | 0,4 |
| | | Norrie disease | 310600 | |
| *NDUFA1* | *NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 1* | Mitochondrial complex I deficiency | 252010 | 0,2 |

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|---|---|---|---|---|
| *NF1* | *NEUROFIBROMIN 1* | Neurofibromatosis, type 1 | 162200 | 8,5 |
| | | Neurofibromatosis-Noonan syndrome | 601321 | |
| | | Watson syndrome | 193520 | |
| *NFIX* | *NUCLEAR FACTOR I/X* | Marshall-Smith syndrome | 602535 | 1,3 |
| | | Sotos syndrome 2 | 614753 | |
| *NHS* | *NHS GENE* | Nance-Horan syndrome | 302350 | 4,9 |
| *NIPBL* | *NIPPED-B-LIKE* | Cornelia-de-Lange syndrome 1 | 122470 | 8,4 |
| *NLGN4X* | *NEUROLIGIN 4* | Mental retardation, X-linked | 300495 | 2,5 |
| *NRAS* | *NEUROBLASTOMA RAS VIRAL ONCOGENE HOMOLOG* | Noonan syndrome 6 | 613224 | 0,6 |
| | | Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic | 163200 | |
| *NRXN1* | *NEUREXIN I* | Pitt-Hopkins-like syndrome 2 | 614325 | 1,3 |
| *NSD1* | *NUCLEAR RECEPTOR-BINDING Su-var, ENHANCER OF ZESTE, AND TRITHORAX DOMAIN PROTEIN 1* | Sotos syndrome 1 | 117550 | 8,1 |
| | | Beckwith-Wiedemann syndrome | 130650 | |
| *NSDHL* | *NAD(P)H STEROID DEHYDROGENASE-LIKE PROTEIN* | CHILD syndrome | 308050 | 1,1 |
| | | CK syndrome | 300831 | |
| *OCRL* | *OCRL GENE* | Dent disease 2 | 300555 | 2,7 |
| | | Lowe syndrome | 309000 | |
| *OFD1* | *OFD1 GENE* | Joubert syndrome 10 | 300804 | 3,0 |
| | | Orofaciodigital syndrome I | 311200 | |
| | | Simpson-Golabi-Behmel syndrome, type 2 | 300209 | |
| *OPHN1* | *OLIGOPHRENIN 1* | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance | 300486 | 2,4 |
| *OTC* | *ORNITHINE CARBAMOYLTRANSFERASE* | Ornithine transcarbamylase deficiency | 311250 | 1,1 |
| *PAK3* | *p21 PROTEIN-ACTIVATED KINASE 3* | Mental retardation, X-linked 30/47 | 300558 | 1,6 |
| *PCDH19* | *PROTOCADHERIN 19* | Epileptic encephalopathy, early infantile, 9 | 300088 | 3,3 |
| *PDHA1* | *PYRUVATE DEHYDROGENASE, ALPHA-1* | Pyruvate dehydrogenase E1-alpha deficiency | 312170 | 1,2 |
| *PGK1* | *PHOSPHOGLYCERATE KINASE 1* | Phosphoglycerate kinase 1 deficiency | 300653 | 1,3 |
| *PHF6* | *PHD FINGER PROTEIN 6* | Borjeson-Forssman-Lehmann syndrome | 301900 | 1,1 |
| *PHF8* | *PHD FINGER PROTEIN 8* | Mental retardation syndrome, X-linked, Siderius type | 300263 | 3,1 |
| *PIK3CA* | *PHOSPHATIDYLINOSITOL 3-KINASE, CATALYTIC, ALPHA* | Cowden syndrome 5 | 615108 | 3,2 |
| *PIK3R2* | *PHOSPHATIDYLINOSITOL 3-KINASE, REGULATORY SUBUNIT 2* | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 | 603387 | 2,2 |

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|---|---|---|---|---|
| *PLP1* | *PROTEOLIPID PROTEIN 1* | Pelizaeus-Merzbacher disease | 312080 | 0,8 |
| | | Spastic paraplegia 2, X-linked | 312920 | |
| *PORCN* | *PORCUPINE, DROSOPHILA, HOMOLOG OF* | Focal dermal hypoplasia | 305600 | 1,4 |
| *PQBP1* | *POLYGLUTAMINE-BINDING PROTEIN 1* | Renpenning syndrome | 309500 | 0,8 |
| *PRPS1* | *PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE I* | Arts syndrome | 301835 | 1,0 |
| | | Gout, PRPS-related | 300661 | |
| | | Phosphoribosylpyrophosphate synthetase superactivity | 300661 | |
| *PTCH1* | *PATCHED, DROSOPHILA, HOMOLOG OF, 1* | Basal cell nevus syndrome | 109400 | 3,9 |
| | | Holoprosencephaly 7 | 610828 | |
| *PTEN* | *PHOSPHATASE AND TENSIN HOMOLOG* | Bannayan-Riley-Ruvalcaba syndrome | 153480 | 1,2 |
| | | Cowden syndrome 1 | 158350 | |
| | | Lhermitte-Duclos syndrome | 158350 | |
| | | Macrocephaly/autism syndrome | 605309 | |
| | | VATER association with macrocephaly and ventriculomegaly | 276950 | |
| *PTPN11* | *PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 11* | LEOPARD syndrome 1 | 151100 | 1,4 |
| | | Metachondromatosis | 156250 | |
| | | Noonan syndrome 1 | 163950 | |
| *RAB39B* | *RAS-ASSOCIATED PROTEIN RAB39B* | ?Waisman syndrome | 311510 | 0,6 |
| | | Mental retardation, X-linked 72 | 300271 | |
| *RAD21* | *RAD21, S. POMBE, HOMOLOG OF* | Cornelia de Lange syndrome 4 | 614701 | 1,9 |
| *RAF1* | *V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1* | LEOPARD syndrome 2 | 611554 | 1,9 |
| | | Noonan syndrome 5 | 611553 | |
| *RBM10* | *RNA-BINDING MOTIF PROTEIN 10* | TARP syndrome | 311900 | 2,8 |
| *RNF135* | *RING FINGER PROTEIN 135* | Macrocephaly, macrosomia, facial dysmorphism syndrome | 614192 | 1,3 |
| *RPS6KA3* | *RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 3* | Coffin-Lowry syndrome | 303600 | 2,2 |
| | | Mental retardation, X-linked 19 | 300844 | |
| *SETD2* | *SET DOMAIN-CONTAINING PROTEIN 2; HUNTINGTIN-INTERACTING PROTEIN B* | Luscan-Lumish syndrome | 616831 | 7,7 |
| *SHOC2* | *SUPPRESSOR OF CLEAR, C. ELEGANS, HOMOLOG OF* | Noonan-like syndrome with loose anagen hair | 607721 | 1,6 |
| *SHROOM4* | *SHROOM FAMILY MEMBER 4* | ?Stocco dos Santos X-linked mental retardation syndrome | 300434 | 4,5 |
| *SLC16A2* | *SOLUTE CARRIER FAMILY 16 (MONOCARBOXYLIC ACID TRANSPORTER), MEMBER 2* | Allan-Herndon-Dudley syndrome | 300523 | 1,6 |

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|---|---|---|---|---|
| *SLC6A8* | *SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, CREATINE), MEMBER 8* | Cerebral creatine deficiency syndrome 1 | 300352 | 1,9 |
| *SLC9A6* | *SOLUTE CARRIER FAMILY 9, MEMBER 6* | Mental retardation, X-linked syndromic, Christianson type | 300243 | 2,0 |
| *SMARCA2* | *SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, MEMBER 2* | Nicolaides-Baraitser syndrome | 601358 | 4,8 |
| *SMARCA4* | *SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, MEMBER 4* | Coffin-Siris syndrome 4 | 614609 | 4,9 |
| *SMARCB1* | *SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY B, MEMBER 1* | Coffin-Siris syndrome 3 | 614608 | 1,2 |
| *SMARCE1* | *SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY E, MEMBER 1* | Coffin-Siris syndrome 5 | 616938 | 1,2 |
| *SMC1A* | *STRUCTURAL MAINTENANCE OF CHROMOSOMES 1A* | Cornelia de Lange syndrome 2 | 300590 | 3,7 |
| *SMC3* | *STRUCTURAL MAINTENANCE OF CHROMOSOMES 3* | Cornelia de Lange syndrome 3 | 610759 | 3,7 |
| *SMS* | *SPERMINE SYNTHASE* | Mental retardation, X-linked, Snyder-Robinson type | 309583 | 1,1 |
| *SOS1* | *SON OF SEVENLESS, DROSOPHILA, HOMOLOG 1* | Noonan syndrome 4 | 610733 | 4,0 |
| *SOX3* | *SRY-BOX 3* | Mental retardation, X-linked, with isolated growth hormone deficiency | 300123 | 1,3 |
| | | Panhypopituitarism, X-linked | 312000 | |
| *SPRED1* | *SPROUTY-RELATED EVH1 DOMAIN-CONTAINING PROTEIN 1* | Legius syndrome (NF1-like) | 611431 | 1,3 |
| *SRPX2* | *SUSHI REPEAT-CONTAINING PROTEIN, X-LINKED, 2* | ?Rolandic epilepsy, mental retardation, and speech dyspraxia | 300643 | 1,4 |
| *SYN1* | *SYNAPSIN I* | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 300491 | 2,1 |
| *SYNGAP1* | *SYNAPTIC RAS-GTPase-ACTIVATING PROTEIN 1* | Mental retardation, autosomal dominant 5 | 612621 | 4,0 |
| *SYP* | *SYNAPTOPHYSIN* | Mental retardation, X-linked 96 | 300802 | 0,9 |
| *TBC1D7* | *TBC1 DOMAIN FAMILY, MEMBER 7* | Macrocephaly/megalencephaly syndrome, autosomal recessive | 248000 | 0,9 |
| *TCF4* | *TRANSCRIPTION FACTOR 4* | Pitt-Hopkins syndrome | 610954 | 2,0 |
| *THOC2* | *THO COMPLEX, SUBUNIT 2* | Mental retardation, X-linked 12/35 | 300957 | 4,8 |
| *TIMM8A* | *TRANSLOCASE OF INNER MITOCHONDRIAL MEMBRANE 8, YEAST, HOMOLOG OF, A* | Mohr-Tranebjaerg syndrome | 304700 | 0,3 |
| *TSC1* | *TSC1 GENE* | Tuberous sclerosis-1 | 191100 | 3,3 |
| | | Focal cortical dysplasia, Taylor balloon cell type | 607341 | |
| *TSC2* | *TSC2 GENE* | Tuberous sclerosis-2 | 613254 | 5,4 |
| *TSPAN7* | *TETRASPANIN 7* | Mental retardation, X-linked 58 | 300210 | 0,8 |

| *Gen-symbol* | *Genname* | Erkrankung(en) | OMIM | Gengröße (kb) |
|---|---|---|---|---|
| *UBE2A* | *UBIQUITIN-CONJUGATING ENZYME E2A* | Mental retardation, X-linked syndromic, Nascimento-type | 300860 | 0,5 |
| *UBE3A* | *UBIQUITIN-PROTEIN LIGASE E3A* | Angelman syndrome | 105830 | 2,6 |
| *UPF3B* | *UPF3, YEAST, HOMOLOG OF, B* | Mental retardation, X-linked, syndromic 14 | 300676 | 1,4 |
| *WDR45* | *WD REPEAT-CONTAINING PROTEIN 45* | Neurodegeneration with brain iron accululation 5 | 300894 | 1,1 |
| *ZDHHC15* | *ZINC FINGER DHHC DOMAIN-CONTAINING PROTEIN 15* | ?Mental retardation, X-linked 91 | 300577 | 1,0 |
| *ZDHHC9* | *ZINC FINGER DHHC DOMAIN-CONTAINING PROTEIN 9* | Mental retardation, X-linked syndromic, Raymond type | 300799 | 1,1 |
| *ZEB2* | *ZINC FINGER E BOX-BINDING HOMEOBOX 2* | Mowat-Wilson syndrome | 235730 | 3,6 |
| *ZNF711* | *ZINC FINGER PROTEIN 711* | Mental retardation, X-linked 97 | 300803 | 2,3 |
| *ZNF81* | *ZINC FINGER PROTEIN 81* | Mental retardation, X-linked 45 | 300498 | 2,0 |