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

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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