Whole Exome Sequencing
Gene package Neuronal migration disorders, version 5, 30-7-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. Additionally, MPLA analysis was performed for several (fragments of) genes involved in neuronal migration disorders (SALSA P061 Lissencephaly; MRC Holland). It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x
ACTB	Baraitser-Winter syndrome 1, 243310?Dystonia, juvenile-onset, 607371	102630	NM_001101.3	208	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717	102560	NM_001199954.1	174	100	100	100
ADA2	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410	607575	NM_001282225.1	91	100	100	97
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	NM_001111.4	79	100	100	98
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	NM_005682.6	112	100	100	99
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500	164730	NM_005163.2	136	100	100	100
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	611223	NM_005465.4	59	100	97	82
ANKLE2	?Microcephaly 16, primary, autosomal recessive, 616681	616062	NM_015114.2	96	100	98	94
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	NM_001272071.1	40	100	87	62
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	NM_001278512.1	97	100	100	98
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066	607245	NM_006594.4	70	100	100	98
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450	607244	NM_007347.4	55	100	99	90

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AP4M1	Spastic paraplegia 50, autosomal recessive, 612936	602296	NM_004722.3	117	100	100	99
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067	607243	NM_007077.4	38	100	97	74
APC2	?Sotos syndrome 3, 617169	612034	NM_005883.2	116	100	97	94
ARFGEF2	Periventricular heterotopia with microcephaly, 608097	605371	NM_006420.2	74	100	99	92
ARNT2	?Webb-Dattani syndrome, 615926	606036	NM_014862.3	81	100	100	94
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	NM_139058.2	46	91	81	72
ASNS	Asparagine synthetase deficiency, 615574	108370	NM_133436.3	64	100	99	90
ASPM	Microcephaly 5, primary, autosomal recessive, 608716	605481	NM_018136.4	61	100	100	95
ASXL1	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286	612990	NM_015338.5	94	100	100	98
ATAD3A	Harel-Yoon syndrome, 617183	612316	NM_018188.3	128	99	98	97
ATAD3B	No OMIM phenotype	612317	NM_031921.5	130	99	98	95
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250	611716	NM_012463.3	70	100	99	94
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600	601215	NM_001184.3	79	100	97	85
ATRIP	No OMIM phenotype	606605	NM_130384.2	97	100	100	96
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181	610194	NM_152490.4	52	100	99	89
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	605517	NM_006876.2	160	100	100	100
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	NM_003688.3	44	100	94	67
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	123833	NM_001759.3	98	100	100	97
CDK5	?Lissencephaly 7 with cerebellar hypoplasia, 616342	123831	NM_004935.3	102	100	100	100
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804	608201	NM_018249.5	63	100	98	88
CDK6	?Microcephaly 12, primary, autosomal recessive, 616080	603368	NM_001145306.1	70	100	100	94
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676	609279	NM_018451.4	64	100	100	95
CEP135	Microcephaly 8, primary, autosomal recessive, 614673	611423	NM_025009.4	72	100	97	86
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823	613529	NM_001194998.1	57	100	97	89
CEP63	?Seckel syndrome 6, 614728	614724	NM_025180.3	61	100	97	84
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	NM_001083314.3	91	100	100	100
CIT	Microcephaly 17, primary, autosomal recessive, 617090	605629	NM_001206999.1	85	100	99	94
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	NM_006831.2	83	100	100	100
COL18A1	Knobloch syndrome, type 1, 267750	120328	NM_130445.2	138	100	100	98

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COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 Schizencephaly, 269160	120130	NM_001845.5	80	100	100	96
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 2, 614483	120090	NM_001846.3	97	100	100	98
CRADD	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499	603454	NM_003805.4	139	100	100	100
CRB2	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730	609720	NM_173689.6	120	100	100	100
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	NM_000100.3	75	100	100	100
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	NM_025099.5	90	100	100	99
CTNNA2	Cortical dysplasia, complex, with other brain malformations 9, 618174	114025	NM_001282597.2	68	100	100	96
CTNND2	No OMIM phenotype	604275	NM_001332.3	81	96	92	85
DAB1	Spinocerebellar ataxia 37, 615945	603448	NM_021080.3	61	100	100	95
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	128239	NM_001165928.3	136	100	100	100
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	NM_003737.2	118	100	100	100
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067	300121	NM_178153.2	48	100	97	87
DDX3X	Mental retardation, X-linked 102, 300958	300160	NM_001356.4	67	100	99	96
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	NM_001242896.1	76	100	99	94
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	NM_001363.3	43	100	97	79
DNMT3A	Acute myeloid leukemia, somatic, 601626 Tatton-Brown-Rahman syndrome, 615879	602769	NM_175629.2	111	100	100	99
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	NM_001376.4	86	100	100	96
EIF2AK3	Wolcott-Rallison syndrome, 226980	604032	NM_004836.6	62	100	99	91
EMG1	Bowen-Conradi syndrome, 211180	611531	NM_006331.7	63	100	100	100
EML1	Band heterotopia, 600348	602033	NM_001008707.1	64	100	99	91
EOMES	No OMIM phenotype	604615	NM_005442.3	84	100	100	100
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	NM_001983.3	69	100	100	93
ERCC2	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	NM_000400.3	91	100	99	97
ERCC5	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	133530	NM_000123.3	75	100	100	96

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ERCC6	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630	609413	NM_000124.2	74	100	99	94
ERMARD	?Periventricular nodular heterotopia 6, 615544	615532	NM_018341.2	59	100	99	90
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	NM_024582.4	78	100	100	98
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	NM_014845.5	53	100	99	88
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155	606596	NM_001039885.2	130	100	100	100
FKTN	Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588	607440	NM_006731.2	72	100	100	98
FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 ?FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	NM_001110556.1	98	100	100	99
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790	610865	NM_017791.2	128	100	100	96
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	616305	NM_018027.4	75	100	96	85
GNAQ	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300	600998	NM_002072.4	85	100	100	100
HNRNPK	Au-Kline syndrome, 616580	600712	NM_002140.4	42	95	80	60
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451	615316	NM_001010867.3	126	100	100	98
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	NM_016097.4	65	100	98	76
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	NM_022168.3	74	100	99	93

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INTS8	No OMIM phenotype	611351	NM_017864.3	44	100	94	74
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052	614631	NM_001101426.3	77	100	99	92
ITSN1	No OMIM phenotype	602442	NM_003024.2	59	100	96	85
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	NM_032801.4	61	100	100	93
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	NM_005886.2	133	100	100	100
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950	148760	NM_004523.3	63	100	98	86
KIF1BP	Goldberg-Shprintzen megacolon syndrome, 609460	609367	NM_015634.3	69	100	100	96
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411	602591	NM_001098511.2	72	100	97	85
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282	604593	NM_004522.2	59	100	98	87
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	NM_198525.2	104	99	96	93
KNL1	Microcephaly 4, primary, autosomal recessive, 604321	609173	NM_170589.4	51	100	98	93
KPTN	Mental retardation, autosomal recessive 41, 615637	615620	NM_007059.3	133	100	100	98
L1CAM	CRASH syndrome, 303350 Corpus callosum, partial agenesis of, 304100 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 MASA syndrome, 303350	308840	NM_000425.4	85	100	100	99
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	NM_005559.3	76	100	99	94
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855	156225	NM_000426.3	60	100	99	92
LAMB1	Lissencephaly 5, 615191	150240	NM_002291.2	84	100	99	95
LAMC1	No OMIM phenotype	150290	NM_002293.3	72	100	99	93
LAMC3	Cortical malformations, occipital, 614115	604349	NM_006059.3	121	100	100	99
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840	603590	NM_004737.4	86	100	100	98
LARP7	Alazami syndrome, 615071	612026	NM_001267039.1	56	100	97	85
MACF1	No OMIM phenotype	608271	NM_012090.5	57	100	97	89
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200	607117	NM_024596.4	76	94	94	88
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	603810	NM_004268.4	81	100	100	96
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219	603785	NM_003829.4	60	100	99	91
MYCN	Feingold syndrome 1, 164280	164840	NM_005378.5	153	100	100	100
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	NM_002485.4	59	100	97	81
NCAPD2	?Microcephaly 21, primary, autosomal recessive, 617983	615638	NM_014865.3	81	100	99	94
NCAPD3	Microcephaly 22, primary, autosomal recessive, 617984	609276	NM_015261.2	54	100	97	87

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NCAPH	?Microcephaly 23, primary, autosomal recessive, 617985	602332	NM_015341.4	61	100	99	92
NCAPH2	No OMIM phenotype	611230	NM_001185011.1	97	100	97	90
NDE1	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013	609449	NM_001143979.1	99	100	100	97
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	NM_001144964	62	100	98	91
NFIA	Brain malformations with or without urinary tract defects, 613735	600727	NM_001145512.1	72	100	100	97
NID1	No OMIM phenotype	131390	NM_002508.2	104	100	100	100
NIN	?Seckel syndrome 7, 614851	608684	NM_020921.3	72	100	98	90
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	NM_001077350.2	80	100	98	94
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	NM_015922.2	62	100	98	88
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	NM_002538.3	78	100	100	99
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	NM_000430.3	72	100	94	89
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	NM_001604.5	66	100	99	94
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	605925	NM_006031.5	121	100	100	98
PHC1	?Microcephaly 11, primary, autosomal recessive, 615414	602978	NM_004426.2	153	100	100	100
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	600286	NM_058004.3	111	100	99	95
PIK3CA	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000	171834	NM_006218.2	73	100	99	93
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	603157	NM_005027.3	85	96	92	90
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	605031	NM_014264.4	53	100	98	87
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	NM_007254.3	100	100	100	98

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POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	NM_018082.5	66	100	98	88
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	NM_017739.3	80	100	100	98
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	NM_007171.3	96	100	100	100
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158	607439	NM_013382.5	78	100	100	95
PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481	617413	NM_021222.2	64	100	98	90
PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 {Meningioma}, 607174 PTEN hamartoma tumor syndrome {Prostate cancer, somatic}, 176807 Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950	601728	NM_000314.4	103	88	79	76
PTF1A	Pancreatic agenesis 2, 615935 Pancreatic and cerebellar agenesis, 609069	607194	NM_178161.2	127	100	100	93
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	NM_013328.3	109	100	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	NM_005051.2	92	100	100	98
RAB18	Warburg micro syndrome 3, 614222	602207	NM_021252.4	74	100	100	91
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	NM_001172435.1	55	100	100	93
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	NM_012414.3	57	100	98	86
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	NM_005732.3	83	100	100	96
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	NM_020320.4	57	100	97	85
RBBP8	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744	604124	NM_002894.2	49	100	98	81
RBM10	TARP syndrome, 311900	300080	NM_001204468.1	69	100	94	87
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	NM_173054.2	64	100	99	92

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RHEB	No OMIM phenotype	601293	NM_005614.3	24	86	54	29
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	NM_006397.2	98	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	NM_024570.3	54	100	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	NM_032193.3	279	100	100	100
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951	612944	NM_003730.4	100	100	100	94
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651	601428	NR_023343.1	No covera	0	0	0
RTEL1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	NM_001283009.1	132	100	100	99
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833	610436	NM_173630.3	59	100	98	88
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	NM_014254.2	91	100	98	87
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	NM_015474.3	58	100	97	80
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	NM_033517.1	106	98	91	84
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	NM_001324336.1	51	100	98	89
SLC25A19	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	606521	NM_001126122.1	99	100	100	99
SMPD4	No OMIM phenotype	610457	NM_017951.4	108	100	100	97
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	604202	NM_004782.3	142	100	100	100
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	NM_014467.2	48	100	98	87
STAMBP	Microcephaly-capillary malformation syndrome, 614261	606247	NM_006463.4	64	100	100	93
STIL	Microcephaly 7, primary, autosomal recessive, 612703	181590	NM_001048166.1	56	100	99	94
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	608626	NM_001003787.2	91	100	100	97
TBC1D20	Warburg micro syndrome 4, 615663	611663	NM_144628.3	67	100	93	93
TBC1D24	DOORS syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	NM_001199107.1	147	100	100	99
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000	612655	NM_016495.5	48	100	97	84
TMTC3	Lissencephaly 8, 617255	617218	NM_181783.3	63	100	98	89
TMX2	No OMIM phenotype	616715	NM_015959.3	61	100	93	86
TRAIP	Seckel syndrome 9, 616777	605958	NM_005879.2	82	100	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	NM_016381.5	217	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	NM_000368.4	153	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	NM_000548.3	189	100	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	NM_207346.2	103	99	96	96
TUBA1A	Lissencephaly 3, 611603	602529	NM_006009.3	101	100	100	100
TUBA8	Cortical dysplasia, complex, with other brain malformations 8, 613180	605742	NM_018943.2	105	100	100	100
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 Symmetric circumferential skin creases, congenital, 1, 156610	191130	NM_178014.3	174	100	99	96
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	NM_001069.2	242	100	99	95
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, 610031	612850	NM_178012.4	250	100	100	100
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638	602661	NM_006086.3	245	100	100	95
TUBB4A	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	NM_006087.3	216	100	100	99
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	NM_001070.4	168	100	100	100
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335	609610	NM_001286414.2	65	100	98	90
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	610053	NM_020461.3	147	100	100	99
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	NM_003383.4	59	100	100	93
VPS13B	Cohen syndrome, 216550	607817	NM_017890.4	63	100	98	91
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	NM_003384.2	46	100	97	84
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563	610657	NM_014846.3	52	100	98	86
WDR4	No OMIM phenotype	605924	NM_033661.4	103	100	100	100
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317	613583	NM_001083961.1	129	100	100	99
WDR73	Galloway-Mowat syndrome 1, 251300	616144	NM_032856.3	140	100	100	95
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, nonsyndromic, autosomal recessive 3, 617967	614218	NM_001163809.1	146	100	100	100
YWHAE	No OMIM phenotype	605066	NM_006761.4	66	100	100	91
ZIC1	Craniosynostosis 6, 616602	600470	NM_003412.3	217	100	100	100
ZIC2	Holoprosencephaly 5, 609637	603073	NM_007129.3	140	96	94	92
ZIC4	No OMIM phenotype	608948	NM_001168378.1	123	100	100	99

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x

- The statistics above are based on a set of 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x