

# Whole Exome Sequencing

## Gene package Intellectual disability, version 13, 30-9-2021



### Technical information

DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique 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/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. It is not excluded that pathogenic variants 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 types of sequence changes).



**Dept. Clinical Genetics**

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
A2ML1	610627	100	100	99.79	98.52
AARS1	601065	100	100	100	99.73
AASS	605113	100	100	99.77	98.39
ABAT	137150	93.95	93.95	93.01	83.82
ABCC9	601439	100	100	99.81	97.71
ABCD1	300371	100	93.68	88.97	73.59
ABCD4	603214	100	100	99.98	94.11
ABHD5	604780	100	100	92.98	91.88
ACAD8	604773	100	100	99.32	91.19
ACAD9	611103	100	100	100	99.82
ACO2	100850	100	100	100	100
ACOT9	300862	100	100	97.60	93.09
ACOX1	609751	100	100	100	98.50
ACSF3	614245	100	100	100	99.11
ACSL4	300157	100	100	99.61	91.09
ACTB	102630	100	100	100	99.59
ACTG1	102560	100	100	100	100
ACVR1	102576	100	100	100	99.36
ACY1	104620	100	100	100	95.35
ADA2	607575	100	100	100	100

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ADAM22	603709	100	100	99.75	97.03
ADAR	146920	98.45	98.45	98.45	98.30
ADAT3	615302	100	100	97.73	88.58
ADGRG1	604110	100	99.08	96.19	85.38
ADK	102750	100	93.49	93.49	85.72
ADNP	611386	100	100	100	100
ADSL	608222	98.31	98.31	98.31	98.31
AFF2	300806	100	99.90	97.45	88.64
AFF4	604417	100	100	100	98.30
AFG3L2	604581	93.67	91.50	91.50	90.44
AGA	613228	100	100	100	97.37
AGAP2	605476	100	96.93	92.38	83.92
AGO1	606228	100	100	100	98.00
AGO2	606229	98.58	98.58	98.58	96.21
AGPAT2	603100	100	100	100	94.13
AGTR2	300034	100	100	100	100
AHCY	180960	100	100	100	100
AHDC1	615790	98.59	96.81	96.14	95.11
AHI1	608894	100	100	100	99.14
AIFM1	300169	100	100	99.27	95.48
AIMP1	603605	100	100	99.15	84.04
AK1	103000	100	100	99.20	82.93
AKT1	164730	100	100	100	98.26
AKT3	611223	100	100	100	97.02
ALDH18A1	138250	100	100	100	98.26
ALDH3A2	609523	100	100	99.77	96.69
ALDH4A1	606811	100	98.94	95.95	93.87
ALDH5A1	610045	100	98.82	90.32	79.94
ALG1	605907	100	98.64	96.25	87.80
ALG11	613666	100	100	100	99.61
ALG12	607144	100	100	100	100
ALG13	300776	100	100	100	95.01
ALG2	607905	100	100	100	97.36
ALG3	608750	100	100	99.33	95.82
ALG6	604566	100	97.72	95.84	94.45
ALG8	608103	100	96.76	91.57	88.06
ALG9	606941	99.91	99.91	98.32	90.50
ALMS1	606844	99.93	99.38	98.73	95.61
ALX1	601527	100	100	100	95.32

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ALX4	605420	100	97.64	93.37	75.86
AMMECR1	300195	100	100	100	81.22
AMPD2	102771	100	100	98.55	90.09
AMT	238310	100	100	100	98.49
ANK3	600465	100	100	99.85	98.91
ANKEF1	No ID	100	100	100	99.44
ANKH	605145	100	100	100	98.72
ANKLE2	616062	98.57	94.60	92.49	89.95
ANKRD11	611192	100	99.77	97.95	94.26
ANO10	613726	100	100	100	97.41
ANTXR1	606410	98.59	97.25	95.93	88.60
AP1S1	603531	100	95.99	95.99	95.99
AP1S2	300629	100	99.91	91.29	75.69
AP3B1	603401	97.72	95.97	94.31	85.98
AP3B2	602166	100	98.95	82.50	74.57
AP4B1	607245	100	100	100	100
AP4E1	607244	100	100	100	97.99
AP4M1	602296	100	100	96.46	81.67
AP4S1	607243	100	100	100	100
APC2	612034	98.41	94.37	91.08	78.13
APTX	606350	100	100	100	98.27
ARFGEF2	605371	100	100	100	98.69
ARG1	608313	100	100	100	99.52
ARHGAP31	610911	100	100	100	99.93
ARHGAP4	300023	100	99.62	94.75	67.77
ARHGEF33	No ID	99.97	99.22	97.13	90.92
ARHGEF6	300267	100	100	100	95.85
ARHGEF9	300429	96.19	96.19	95.89	89.65
ARID1A	603024	97.55	93.37	90.49	85.63
ARID1B	614556	96.99	93.58	87.86	77.69
ARID2	609539	100	100	99.40	95.70
ARL13B	608922	100	99.19	97.91	85.75
ARL6	608845	100	100	100	95.53
ARNT2	606036	100	100	98.72	95.99
ARSA	607574	100	100	100	100
ARSL	300180	100	100	99.83	90.82
ARX	300382	83.87	74.50	64.03	41.41
ASAH1	613468	100	100	99.21	92.70
ASCL1	100790	100	100	100	86.06

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ASH1L	607999	100	100	100	99.32
ASL	608310	100	100	100	97.14
ASNS	108370	100	100	100	97.53
ASPA	608034	100	100	100	100
ASPM	605481	100	99.21	97.94	93.28
ASS1	603470	100	100	100	91.16
ASXL1	612990	98.42	98.42	98.42	97.45
ASXL2	612991	100	98.95	97.24	96.97
ASXL3	615115	100	99.87	99.44	97.67
ATAD3A	612316	95.68	89.87	86.81	82.45
ATAD3B	612317	95.41	90.06	87.19	83.50
ATCAY	608179	100	100	100	99.70
ATIC	601731	100	100	99.93	95.94
ATN1	607462	100	98.75	95.68	82.04
ATP1A2	182340	100	100	100	97.98
ATP1A3	182350	100	100	99.43	95.12
ATP2A2	108740	100	100	99.74	95.36
ATP6AP2	300556	100	100	98.50	86.79
ATP6VOA2	611716	100	100	99.23	94.78
ATP6V1B2	606939	100	100	98.37	94.21
ATP7A	300011	100	100	99.49	95.63
ATP8A2	605870	100	100	100	97.00
ATPAF2	608918	100	100	100	99.51
ATR	601215	100	100	99.66	95.63
ATRIP	606605	100	100	100	98.58
ATRX	300032	100	99.42	96.84	86.54
AUH	600529	100	100	100	100
AUTS2	607270	100	99.39	94.65	72.72
AVPR2	300538	100	100	97.67	85.94
B3GALNT2	610194	100	100	100	92.67
B3GALT6	615291	79.89	76.12	72.74	70.01
B3GLCT	610308	100	94.98	94.98	94.31
B4GALNT1	601873	100	97.80	95.61	87.91
B4GALT1	137060	100	100	99.73	78.37
B4GALT7	604327	98.64	93.64	93.64	85.79
B4GAT1	605517	100	100	100	100
BBS1	209901	100	100	100	99.32
BBS10	610148	100	100	99.55	98.14
BBS12	610683	100	100	100	100

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BBS2	606151	100	100	100	100
BBS4	600374	100	100	100	95.74
BBS5	603650	100	100	100	97.62
BBS7	607590	100	100	99.45	96.44
BBS9	607968	99.97	99.59	98.64	93.69
BCAP31	300398	100	100	96.53	73.18
BCKDHA	608348	100	98.94	96.34	89.57
BCKDHB	248611	100	100	100	100
BCL11A	606557	100	100	100	99.52
BCL11B	606558	100	99.08	97.43	90.55
BCOR	300485	100	100	99.79	96.98
BCORL1	300688	100	99.39	96.66	84.83
BCS1L	603647	100	100	100	100
BICRA	605690	97.83	93.83	87.96	74.01
BLM	604610	100	99.64	98.63	94.07
BRAF	164757	95.04	94.33	93.05	87.97
BRAT1	614506	100	98.26	93.33	89.77
BRF1	604902	100	98.15	93.39	77.63
BRPF1	602410	100	100	99.95	98.39
BRWD3	300553	100	99.61	96.64	87.61
BSCL2	606158	100	100	98.45	95.32
BTD	609019	100	100	100	100
BUB1B	602860	100	99.13	96.32	95.21
c12orf4	616082	No coverage data			
C12orf57	615140	100	100	100	100
C1orf167		100	99.54	95.67	87.08
C2CD3	615944	100	100	100	98.84
CA2	611492	100	100	100	100
CA5A	114761	100	100	100	96.97
CA8	114815	100	100	100	98.45
CACNA1A	601011	99.96	98.59	94.57	79.29
CACNA1C	114205	100	100	99.73	97.26
CACNA2D1	114204	98.29	97.81	95.33	86.90
CACNG2	602911	100	100	100	99.34
CAD	114010	100	100	99.97	97.66
CAMK2A	114078	100	100	100	92.85
CAMK2B	607707	100	97.27	92.49	84.64
CAMTA1	611501	98.81	98.81	98.05	92.63
CAPN10	605286	100	99.69	97.81	88.98

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CASK	300172	100	99.04	96.53	85.71
CBL	165360	100	100	100	99.36
CBS	613381	100	100	100	100
CBY1	607757	100	100	100	100
CC2D1A	610055	100	99.80	98.03	86.89
CC2D2A	612013	100	99.87	98.46	91.02
CCBE1	612753	100	100	100	96.49
CCDC13	No ID	100	99.98	97.75	87.40
CCDC14	617147	100	100	99.46	97.23
CCDC174	616735	100	99.23	90.22	74.24
CCDC22	300859	100	99.01	94.15	79.00
CCDC78	614666	100	100	100	92.47
CCDC88C	611204	100	100	99.51	94.41
CCND2	123833	100	99.69	96.07	84.38
CDC5L	602868	100	100	99.54	92.11
CDH15	114019	100	98.49	94.36	77.94
CDK16	311550	100	100	99.33	94.06
CDK5	123831	100	100	100	94.89
CDK5RAP2	608201	100	99.50	98.31	94.35
CDK6	603368	100	100	100	94.36
CDKL5	300203	100	99.74	96.36	89.33
CDKN1C	600856	82.09	66.60	58.55	48.84
CDON	608707	100	100	99.41	96.44
CENPJ	609279	100	100	100	99.46
CEP104	616690	100	100	99.86	97.26
CEP135	611423	100	99.73	96.72	81.09
CEP152	613529	97.93	97.71	95.63	92.34
CEP290	610142	92.60	87.26	83.53	75.20
CEP41	610523	100	100	100	97.30
CEP63	614724	100	100	96.71	84.05
CEP85L	618865	98.08	94.49	90.83	86.10
CEP89	615470	100	98.13	94.16	85.23
CERT1	604677	100	100	100	94.94
CHAMP1	616327	100	100	100	100
CHD2	602119	100	99.22	96.72	90.17
CHD3	602120	95.74	95.72	94.54	89.90
CHD4	603277	100	100	99.90	96.26
CHD5	610771	99.70	97.43	93.55	82.60
CHD7	608892	100	100	99.90	98.10

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CHD8	610528	100	100	99.99	99.63
CHKB	612395	100	100	100	98.19
CHMP1A	164010	100	100	100	88.57
CHRNA4	118504	100	95.20	95.20	94.90
CIC	612082	100	100	99.81	92.41
CIT	605629	100	99.79	99.21	96.50
CKAP2L	616174	100	98.22	95.28	87.58
CLCN3	600580	100	100	100	98.64
CLCN4	302910	100	100	99	90.66
CLCNKB	602023	100	100	99.32	90.23
CLIC2	300138	100	100	100	97.74
CLIP1	179838	100	100	99.36	96.30
CLN3	607042	100	97.96	91.76	88.85
CLN5	608102	93.14	93.14	87.18	68.93
CLN6	606725	100	100	99.35	92.17
CLN8	607837	100	100	100	100
CLP1	608757	100	100	100	100
CLPB	616254	100	100	100	98.66
CLTC	118955	100	100	99.49	98.35
CNKS2	300724	100	99.49	96.30	80.03
CNNM2	607803	100	100	100	97.45
CNOT3	604910	99.42	96.15	90.73	82.07
CNTNAP2	604569	100	100	99.80	97.78
COASY	609855	100	100	99.80	94.52
COG1	606973	100	100	100	97.97
COG4	606976	100	100	100	98.45
COG5	606821	99.80	96.52	95.52	91.65
COG6	606977	100	100	100	94.13
COG7	606978	100	100	100	99.04
COG8	606979	92.70	92.27	90.10	86.93
COL11A1	120280	100	99.17	97.37	87.82
COL12A1	120320	100	99.75	98.55	92.77
COL18A1	120328	100	99.83	98.32	86.94
COL4A1	120130	98.87	97.12	94.05	85.47
COL4A2	120090	100	99.92	99.04	92.59
COLEC11	612502	100	100	99.10	93.47
COQ2	609825	100	100	98.08	85.92
COQ4	612898	100	94.33	84.65	64.60
COQ8A	606980	100	100	100	98.00

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COQ9	612837	100	96.08	91.80	87.87
COX10	602125	100	100	100	98.60
COX15	603646	100	100	99.63	94.46
COX6B1	124089	100	100	100	93.08
CPLANE1	614571	100	99.26	97.64	95.07
CPS1	608307	99.28	99.28	99.28	99.26
CRADD	603454	100	100	99.84	93.56
CRB2	609720	100	99.53	97.95	88.29
CRBN	609262	100	100	99.13	94.50
CREBBP	600140	100	99.23	97.31	92.60
CRLF1	604237	90.66	90.66	89.00	80.95
CRPPA	614631	100	99.97	97.01	87.99
CSNK2A1	115440	100	100	100	100
CSNK2B	115441	100	100	100	100
CSPP1	611654	100	97.28	95.13	90.44
CSTB	601145	100	100	90.82	75.71
CTBP1	602618	91.72	83.33	82.44	78.78
CTC1	613129	100	100	99.94	95.90
CTCF	604167	100	97.02	90.99	85.15
CTDP1	604927	99.95	94.29	88.29	81.86
CTNNB1	116806	100	100	100	100
CTNND1	601045	100	100	100	97.99
CTNND2	604275	94.56	92.55	90.44	84.97
CTSA	613111	100	99.83	99.83	94.72
CTSD	116840	99.58	93.79	93.79	91.28
CTTNBP2	609772	100	99.72	98.26	95.46
CUBN	602997	100	100	99.98	98.64
CUL3	603136	100	100	99.34	90.93
CUL4B	300304	100	99.56	96.66	87.12
CUX1	116896	99.14	96.52	93.60	85.32
CWF19L1	616120	100	100	100	94.90
CYB5R3	613213	100	100	97.18	86.07
CYP27A1	606530	100	100	100	100
CYP2U1	610670	99.02	95.24	92.84	88.42
D2HGDH	609186	100	100	98.62	95.35
DAB1	603448	100	100	100	100
DAG1	128239	100	100	99.82	97.58
DARS2	610956	100	100	100	97.36
DBT	248610	100	99.94	98.62	93.58



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DCAF17	612515	100	100	99.57	92.53
DCC	120470	100	100	100	98.17
DCHS1	603057	100	100	100	99.83
DCPS	610534	100	100	100	98.91
DCX	300121	100	100	97.92	83.70
DDC	107930	100	99.26	94.35	81.09
DDHD2	615003	100	100	100	99.41
DDX11	601150	100	100	98.12	85.83
DDX3X	300160	100	100	100	97.21
DEAF1	602635	95.14	87.42	83.98	79.33
DENND5A	617278	100	100	99.24	94.98
DEPDC5	614191	100	100	99.65	97.73
DHCR24	606418	100	100	100	97.63
DHCR7	602858	100	100	100	96.13
DHFR	126060	100	100	99.93	87.44
DHPS	600944	100	100	100	100
DHTKD1	614984	100	100	99.39	96.09
DHX30	616423	100	100	100	99.56
DIAPH1	602121	100	98.99	95.18	87.42
DIP2B	611379	98.86	97.81	97.81	97.52
DIS3L2	614184	100	100	100	92.79
DKC1	300126	100	99.10	97.39	86.54
DLD	238331	100	100	100	98.42
DLG3	300189	100	100	99.65	93.09
DLG4	602887	100	100	99.25	91.68
DMD	300377	100	100	98.85	92.14
DMPK	605377	100	100	98.36	88.21
DNAJC12	606060	100	100	100	97.91
DNAJC19	608977	100	100	100	100
DNM1	602377	99.13	94.20	92.24	88.77
DNMT3A	602769	100	100	94.09	74.73
DNMT3B	602900	100	100	99.60	94.18
DOCK6	614194	99.70	99.00	96.77	88.58
DOCK7	615730	100	98.84	97.99	95.35
DOCK8	611432	100	99.90	99.32	96.02
DOLK	610746	100	100	100	100
DONSON	611428	98.37	93.78	87.07	76.08
DPAGT1	191350	100	100	100	99.18
DPF2	601671	100	100	99.59	96.29

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DPH1	603527	100	100	100	94.88
DPM1	603503	100	100	100	98.96
DPP6	126141	95.41	95.41	95.41	94.36
DPYD	612779	100	100	100	100
DPYS	613326	100	100	99.77	97.44
DST	113810	100	100	100	99.32
DYM	607461	100	100	100	98.23
DYNC1H1	600112	100	100	99.80	97.54
DYRK1A	600855	100	100	100	97.83
EBP	300205	100	100	100	91.30
EDC3	609842	100	100	100	100
EDEM3	610214	99.42	95.93	93.90	89.78
EDNRB	131244	100	99.80	98.80	95.27
EDRF1	No ID	100	100	99.43	94.26
EED	605984	100	100	100	96.32
EEF1A2	602959	100	99.48	96.89	87.18
EFCAB1	No ID	100	100	98.21	88.11
EFTUD2	603892	100	100	100	97.84
EHMT1	607001	99.08	99.08	97.81	93.17
EIF2AK3	604032	100	97.98	94.32	90.79
EIF2S3	300161	100	100	100	97.37
EIF4A3	608546	100	99.46	95.69	83.91
EIF4G1	600495	100	99.47	98.70	94.28
ELAC2	605367	100	100	100	97.92
ELOVL4	605512	100	100	100	95.67
ELP2	616054	100	99.47	97.59	93.97
EMC1	616846	99.16	99.16	99.16	97.58
EMG1	611531	100	100	100	100
EML1	602033	99.86	97.06	97.06	96.49
EMX2	600035	100	99.26	93.26	78.68
ENTPD1	601752	100	100	100	99.49
EOMES	604615	100	100	96.53	83.60
EP300	602700	100	99.97	99.24	94.14
EPB41L1	602879	100	100	100	96.77
EPG5	615068	100	99.91	99.15	95.77
ERCC1	126380	100	99.34	95.56	68.39
ERCC2	126340	100	100	100	95.44
ERCC3	133510	100	100	100	97.49
ERCC5	133530	100	100	98.52	91.56

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ERCC6	609413	100	99.94	99.25	98.20
ERCC8	609412	100	100	100	98.99
ERF	611888	100	100	97.56	91.33
ERLIN2	611605	100	100	100	100
ERMARD	615532	100	100	98.43	94.19
ESCO2	609353	100	100	100	98.65
ETFB	130410	100	100	96.55	71.58
ETHE1	608451	100	95.23	88.80	87.64
EXOSC2	602238	100	100	100	99.06
EXOSC3	606489	100	100	100	100
EZH2	601573	100	100	99.96	97.82
FA2H	611026	100	95.14	87.98	73.33
FAM126A	610531	100	100	100	100
FAR1	616107	100	100	100	99.43
FAT2	604269	100	99.91	99.80	99.34
FAT4	612411	100	100	99.98	99.68
FBN1	134797	100	100	100	99.50
FBN2	612570	100	100	100	99.22
FBXL4	605654	100	100	100	98.13
FBXO11	607871	98.36	92.90	92.22	91.27
FBXO28	609100	100	100	94.98	79.74
FBXO31	609102	100	98.53	94.82	88.15
FGD1	300546	100	97.33	93.32	77.63
FGF12	601513	100	100	100	99.88
FGF14	601515	100	100	100	100
FGFR1	136350	100	100	100	98.89
FGFR2	176943	100	100	99.89	98.47
FGFR3	134934	100	99.64	98.10	91.14
FGL1	605776	100	100	95.96	87.64
FH	136850	100	97.46	94.80	91.21
FIBP	608296	100	100	100	92.80
FIG4	609390	100	100	100	99.32
FIGN	605295	100	100	100	100
FKRP	606596	100	100	100	98.44
FKTN	607440	100	100	99.52	93.95
FLNA	300017	99.98	98.83	97.47	92.13
FLVCR1	609144	100	100	98.87	92.92
FMN2	606373	99.93	94.63	87.43	71.08
FMR1	309550	100	99.91	96.49	87.66

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
FOXG1	164874	100	96.07	92.57	87.86
FOXP1	605515	100	100	100	95.16
FOXP2	605317	100	100	100	97.97
FRAS1	607830	100	100	99.39	94.71
FREM2	608945	100	100	99.98	99.09
FRMD4A	616305	100	99.95	99.14	95.91
FRMPD4	300838	77.87	76.76	75.92	73.00
FRRS1L	604574	100	91.27	83.66	68.54
FTCD	606806	97.87	92.56	92.56	91.59
FTO	610966	95.33	95.33	95.33	94.83
FTSJ1	300499	100	100	100	97.42
FUCA1	612280	100	100	100	99.84
GABBR2	607340	97.85	96.88	95.35	89.65
GABRA1	137160	100	100	100	97.29
GABRB1	137190	100	100	100	99.56
GABRB3	137192	100	100	100	100
GAD1	605363	100	100	100	100
GALC	606890	99.96	99.96	99.96	96.49
GALE	606953	100	100	99.76	97.07
GALT	606999	100	100	100	98.14
GAMT	601240	100	94.38	82.14	77.84
GAS6	600441	97.13	94.19	91.22	82.88
GATAD2B	614998	100	100	100	97.37
GATM	602360	100	100	100	100
GBA	606463	100	100	100	100
GCDH	608801	100	100	100	97.91
GCH1	600225	94.02	94.02	94.02	91.38
GCSH	238330	100	76.82	72.86	67.85
GDI1	300104	100	100	100	95.32
GFAP	137780	100	100	97.71	84.13
GFM2	606544	100	100	100	98.32
GJA1	121014	100	100	100	100
GJB1	304040	100	100	97.24	89.76
GJC2	608803	99.36	94.35	86.24	62.68
GK	300474	99.55	94.73	87.05	72.39
GLA	300644	100	100	100	97.62
GLB1	611458	100	100	100	98.35
GLDC	238300	100	100	99.72	96.67
GLI2	165230	100	98.72	96.47	88.37

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
GLI3	165240	100	100	100	99.98
GLYCTK	610516	100	100	100	98.88
GM2A	613109	100	100	100	100
GMPPA	615495	100	100	100	94.10
GMPPB	615320	100	100	100	99.85
GNAI1	139310	100	100	98.77	88.42
GNAO1	139311	100	100	100	100
GNAQ	600998	100	100	100	98.11
GNAS	139320	100	100	100	98.21
GNB1	139380	100	100	100	98.60
GNB2	139390	100	99.46	94.04	82.83
GNB5	604447	100	100	100	98.04
GNPAT	602744	100	100	99.79	95.83
GNPTAB	607840	100	100	99.81	98.21
GNS	607664	100	98.09	96.36	93.21
GPC3	300037	100	100	98.72	90.20
GPC4	300168	100	100	100	98.02
GPHN	603930	97.10	97.10	96.67	93.02
GPR101	300393	100	100	100	100
GPT2	138210	91.98	85.30	85.30	82.31
GRIA3	305915	100	100	100	96.88
GRID2	602368	100	100	100	98.71
GRIK2	138244	100	100	99.97	97.09
GRIK5	600283	100	100	99.25	94.21
GRIN1	138249	100	100	98.83	93.09
GRIN2A	138253	100	100	100	99.40
GRIN2B	138252	100	100	100	99.30
GRIN3B	606651	93.09	86.36	78.15	63.03
GRIP1	604597	100	100	100	98.20
GRM1	604473	100	100	100	99.47
GSE1	616886	100	99.57	95.90	82.90
GSS	601002	100	100	97.62	92.03
GTF2H5	608780	100	100	100	100
GTPBP3	608536	100	100	98.41	90.36
GUCY2F	300041	100	100	100	95.47
GUSB	611499	100	100	100	98.65
H1-4	142220	100	100	100	85.38
H19	103280	No coverage data			
HACE1	610876	100	100	98.41	92.50

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
HAX1	605998	100	100	100	99.54
HCCS	300056	100	100	100	97.78
HCFC1	300019	100	100	99.56	94.75
HCN1	602780	100	100	100	98.62
HDAC4	605314	99.74	95.85	94.63	84.31
HDAC6	300272	100	100	100	97.58
HDAC8	300269	100	100	98.92	85.02
HECTD1	618649	99.97	98.88	97.36	95.24
HECW2	617245	100	100	99.48	97.55
HEPACAM	611642	100	96.69	79.94	76.95
HERC1	605109	100	100	99.96	98.40
HERC2	605837	98.08	95.57	93.40	87.87
HESX1	601802	100	100	93.54	82.44
HEXA	606869	100	100	100	99.26
HEXB	606873	100	97.69	94.61	88.91
HID1	605752	100	99.96	98.38	84.95
HIVEP2	143054	100	100	100	100
HLCS	609018	86.97	86.97	86.97	86.62
HMGCL	613898	100	100	100	100
HNMT	605238	100	100	100	100
HNRNPH2	300610	100	100	100	100
HNRNPK	600712	100	100	98.82	93.82
HNRNPU	602869	100	98.62	95.50	87.79
HOXA1	142955	100	100	100	100
HPD	609695	100	100	100	98.87
HPRT1	308000	97.24	94.54	88.61	78.00
HRAS	190020	100	100	100	100
HSD17B10	300256	100	100	100	94.24
HSPA9	600548	100	100	99.73	97.50
HSPD1	118190	100	100	99.54	97.11
HUWE1	300697	100	99.63	98.20	89.54
IARS1	600709	100	100	100	99.63
IBA57	615316	100	100	100	100
IDS	300823	100	99.97	95.71	84.45
IDUA	252800	99.58	89.68	81.91	70.12
IER3IP1	609382	100	100	100	97.55
IFIH1	606951	100	100	100	99.12
IFT172	607386	100	100	99.99	98.15
IFT81	605489	100	100	99.16	89.36

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
IGBP1	300139	100	100	96.65	86.72
IGF1	147440	100	100	100	96.47
IGF1R	147370	100	100	98.94	96.84
IKBKKG	300248	38.62	26.61	25.50	20.50
IL1RAPL1	300206	100	100	99.48	95.56
IMPA1	602064	100	100	93.34	93.34
INPP5B	147264	100	100	100	99.34
INPP5E	613037	99.81	97.70	95.03	87.08
INPP5K	607875	100	100	100	95.86
INTS1	611345	99.43	96.56	93.16	82.70
INTS8	611351	99.91	98.40	95.50	87.06
IQSEC2	300522	98.08	94.51	87.89	61.40
ISG15	147571	100	100	100	95.89
ITGA7	600536	100	99.94	98.73	92.68
ITPR1	147265	100	100	99.68	97.29
ITPR2	600144	99.99	98.60	97.71	93.01
ITSN1	602442	100	100	99.40	94.28
IVD	607036	100	100	100	98.26
JAG1	601920	100	98.32	97.58	97.57
JAM3	606871	100	100	100	99.41
JMJD1C	604503	100	99.93	99.38	96.21
KALRN	604605	100	100	99.83	96.99
KANK1	607704	100	100	100	99.96
KANSL1	612452	100	100	99.55	96.31
KAT6A	601408	100	100	99.78	98.40
KAT6B	605880	100	99.58	98.78	94.01
KATNB1	602703	100	100	100	99.55
KCNA2	176262	100	100	100	100
KCNA4	176266	100	100	100	100
KCNB1	600397	100	100	100	100
KCNC3	176264	87.07	80.48	73.69	64.23
KCNH1	603305	100	100	100	99.00
KCNJ10	602208	100	100	100	100
KCNJ11	600937	100	100	100	100
KCNJ6	600877	100	100	100	100
KCNJ8	600935	100	100	100	100
KCNK9	605874	100	100	100	100
KCNMA1	600150	100	100	93.77	72.83
KCNQ1OT1	604115	No coverage data			

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
KCNQ2	602235	99.77	97.41	92.06	84.22
KCNQ3	602232	99.66	96.84	95.34	91.05
KCNQ5	607357	100	97.40	94.23	90.49
KCNT1	608167	99.91	97.05	91.61	79.87
KCTD7	611725	100	100	100	98.84
KDM1A	609132	100	98.15	94.57	87.24
KDM4B	609765	100	99.50	98.29	94.51
KDM5B	605393	100	100	99.88	98.07
KDM5C	314690	100	100	99.48	94.03
KDM6A	300128	100	99.78	97.98	86.21
KDM6B	611577	99.69	96.75	92.60	82.54
KDSR	136440	100	100	99.50	89.80
KIAA0586	610178	100	100	98.11	89.90
KIAA1109	611565	100	100	99.80	96.84
KIDINS220	615759	100	99.97	99.00	95.71
KIF11	148760	100	99.50	96.77	89.01
KIF1A	601255	100	100	99.63	94.52
KIF2A	602591	100	100	98.28	88.42
KIF4A	300521	100	100	99.73	90.90
KIF5C	604593	100	98.83	95.34	87.27
KIF7	611254	99.84	96.33	91.96	81.11
KIFBP	609367	100	100	100	94.81
KIRREL3	607761	100	100	99.70	95.46
KLHL15	300980	100	100	100	100
KMT2A	159555	99.92	99.22	98.31	95.93
KMT2B	606834	95.64	94.85	93.53	89.22
KMT2C	606833	99.45	98.99	97.89	94.42
KMT2D	602113	100	100	99.94	98.49
KMT2E	608444	100	99.93	98.42	92.53
KMT5B	610881	100	100	100	96.81
KNL1	609173	99.29	99.28	98.97	98.08
KPTN	615620	100	100	97.19	85.14
KRAS	190070	100	100	100	100
KRBOX4	300585	100	100	100	100
L1CAM	308840	100	100	99.08	86.61
L2HGDH	609584	100	100	98.93	90.06
LAMA1	150320	100	100	99.99	98.15
LAMA2	156225	100	100	99.89	97.74
LAMC1	150290	100	99.74	98.81	95.38



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LAMC3	604349	100	99.45	98.02	88.98
LAMP2	309060	100	100	99.04	84.25
LARGE1	603590	100	100	99.45	96.92
LARP7	612026	100	99.19	94.94	79.55
LAS1L	300964	100	100	100	97.16
LIAS	607031	100	99.33	92.76	92.76
LIG4	601837	96.16	96.16	96.16	96.16
LINS1	610350	100	100	100	100
LMAN2L	609552	100	100	100	96.26
LONP1	605490	100	98.80	96.85	90.50
LRP2	600073	100	99.89	99.21	97.47
LRPPRC	607544	100	99.82	98.76	95.24
LZTFL1	606568	100	97.97	88.39	76.98
MAB21L2	604357	100	100	100	99.91
MACF1	608271	100	100	99.71	97.26
MACROD2	611567	89.26	89.26	89.26	87.45
MAF	177075	84.75	78.90	75.82	70.62
MAGEC3	300469	100	99.26	96.72	87.46
MAGEL2	605283	100	100	99.73	94.20
MAGT1	300715	100	97.92	91.47	86.70
MAN1B1	604346	100	100	100	98.54
MAN2B1	609458	100	99.90	97.81	88.61
MANBA	609489	100	100	99.97	99.16
MAOA	309850	100	100	100	93.35
MAP2K1	176872	100	100	100	98.21
MAP2K2	601263	100	100	98.06	86.80
MAPRE2	605789	100	100	100	100
MASP1	600521	100	100	100	99.30
MAST1	612256	100	99.97	98.40	89.81
MAT1A	610550	100	100	100	100
MBD5	611472	100	100	99.78	98.89
MBOAT7	606048	100	98.91	95.40	86.31
MBTPS2	300294	100	99.80	96.51	91.78
MCCC1	609010	100	100	100	94.60
MCCC2	609014	100	100	97.68	91.01
MCF2L	609499	100	100	98.56	93.29
MCM3AP	603294	100	99.82	98.82	94.78
MCOLN1	605248	100	100	98.79	91.56
MCOLN3	607400	100	100	100	99.32

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
MCPH1	607117	100	97.13	94.14	89.45
MDH2	154100	100	100	100	99.14
MECP2	300005	100	100	100	88.50
MECR	608205	99.41	93.32	86.43	83.31
MED12	300188	100	99.82	97.42	89.88
MED13	603808	100	99.86	99.35	97.16
MED13L	608771	100	99.89	98.72	96.49
MED17	603810	100	100	99.45	92.64
MED23	605042	100	100	99.69	96.56
MED25	610197	100	99.89	98.03	86.70
MEF2C	600662	100	100	100	93.96
MEIS2	601740	100	100	99.34	97.31
METTTL23	615262	100	100	100	100
MFSD2A	614397	100	100	100	94.76
MFSD8	611124	100	100	100	98.06
MGAT2	602616	100	100	100	100
MICU1	605084	100	100	100	99.74
MID1	300552	100	100	100	95.23
MID2	300204	100	100	99.77	94.30
MKKS	604896	100	100	100	100
MKS1	609883	100	99.80	98.92	94.23
MLC1	605908	100	100	100	92.34
MLYCD	606761	99.49	93.89	86.45	72.96
MMAA	607481	100	100	100	96.51
MMADHC	611935	100	100	99.17	85.36
MMUT	609058	100	100	100	98.63
MOCS1	603707	100	100	96.80	93.26
MOCS2	603708	100	100	100	100
MOGS	601336	100	100	100	98.42
MORC2	616661	100	100	100	99.50
MPDU1	604041	100	100	100	90.24
MPDZ	603785	98.36	98.36	98.36	97.32
MPLKIP	609188	100	94.97	89.77	75.48
MRPL3	607118	100	100	100	100
MRPS22	605810	100	100	100	92.14
MSI1	603328	94.22	89.75	81.73	62.41
MSL2	614802	100	100	100	100
MTFMT	611766	100	100	99.44	93.47
MTHFR	607093	100	100	100	97.21

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
MTHFS	604197	100	100	100	98.06
MTMR9	606260	100	100	99.62	95.37
MTOR	601231	100	99.87	99.52	97.16
MTR	156570	100	100	99.46	97.37
MTRFR	613541	No coverage data			
MTRR	602568	100	100	100	99.62
MVK	251170	100	100	100	97.30
MYCN	164840	100	100	97.68	91.49
MYH9	160775	100	98.68	93.13	77.91
MYO5A	160777	98.09	96.39	94.93	92.09
MYT1L	613084	100	100	100	98.08
NAA10	300013	100	99.83	90.58	57.17
NAA15	608000	100	100	100	95.71
NACC1	610672	100	100	98.27	94.35
NAGA	104170	100	100	100	91.86
NAGLU	609701	100	94.55	91.12	82.97
NALCN	611549	100	100	100	98.05
NANS	605202	100	100	100	97.28
NARS2	612803	99.46	93.76	93.76	92.04
NAT10	609221	100	99.15	97.14	96.53
NBEA	604889	99.00	98.87	97.98	93.94
NBN	602667	100	100	100	97.35
NCOR2	600848	99.82	96.70	88.51	68.93
NDE1	609449	100	100	96.78	82.83
NDP	300658	100	100	100	100
NDST1	600853	100	100	99.66	96.79
NDUFA1	300078	100	100	100	100
NDUFA11	612638	100	100	100	86.78
NDUFA12	614530	100	100	100	97.57
NDUFA2	602137	100	100	100	100
NDUFAF3	612911	100	100	98.93	90.03
NDUFAF5	612360	100	95.12	95.12	91.79
NDUFS1	157655	100	100	100	99.52
NDUFS2	602985	100	100	100	93.11
NDUFS3	603846	100	100	100	100
NDUFS4	602694	100	100	100	100
NDUFS6	603848	100	100	99.89	85.62
NDUFS7	601825	100	96.18	92.87	79.60
NDUFS8	602141	100	100	98	90.13

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
NDUFV1	161015	100	100	99.91	96.29
NDUFV2	600532	100	100	97.41	90.85
NECAP1	611623	100	100	100	100
NECTIN1	600644	100	100	98.62	90.54
NEDD4L	606384	100	99.80	97.43	93.46
NEU1	608272	100	100	100	99.71
NEXMIF	300524	100	100	100	100
NF1	613113	100	99.58	98.32	93.20
NFATC1	600489	100	99.83	98.40	95.15
NFE2L2	600492	100	100	100	96.61
NFIA	600727	100	95.90	95.14	88.28
NFIX	164005	100	100	98.51	88.56
NGLY1	610661	100	100	97.64	91.78
NHS	300457	100	96.81	92.89	87.99
NID1	131390	100	100	99.81	94.35
NIN	608684	100	100	99.22	96.48
NIPBL	608667	100	99.98	99.38	96.10
NKX2-1	600635	100	100	99.29	74.39
NLGN3	300336	100	99.27	98.17	90.46
NLGN4X	300427	100	100	100	100
NLRP3	606416	100	100	100	99.67
NONO	300084	100	99.91	97.71	87.17
NOVA2	601991	93.41	85.25	79.88	65.84
NPC1	607623	100	99.98	99.07	97.09
NPC2	601015	100	100	100	100
NPHP1	607100	96.88	96.88	96.88	94.79
NPRL3	600928	100	100	99.80	87.95
NR2F1	132890	100	97.40	92.36	82.84
NRAS	164790	100	100	100	99.85
NRXN1	600565	100	100	100	98.88
NSD1	606681	100	99.93	99.58	97.67
NSDHL	300275	100	100	100	96.78
NSUN2	610916	100	97.69	94.44	90.92
NTRK1	191315	100	99.76	96.50	84.79
NUP62	605815	100	100	100	99.21
OAT	613349	100	95.40	92.47	87.93
OCLN	602876	82.90	72.78	70.85	58.74
OCRL	300535	98.15	98.15	98.15	97.93
ODC1	165640	100	100	100	98.96

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
OFD1	300170	100	99.66	98.24	86.94
OGT	300255	100	100	100	98.45
OPHN1	300127	100	100	100	95.34
ORC1	601902	100	100	100	99.40
OTC	300461	100	100	99.52	93.62
P2RX6	608077	100	100	100	98.85
PACS1	607492	98.03	93.40	88.90	88.48
PACS2	610423	99.27	96.39	95.14	89.31
PAFAH1B1	601545	100	97.90	93.36	93.36
PAH	612349	100	100	100	100
PAK3	300142	100	100	99.32	83.57
PANK2	606157	100	100	100	99.06
PANX1	608420	100	100	100	96.70
PAX1	167411	100	95.30	80.70	68.74
PAX6	607108	100	100	100	100
PAX8	167415	100	98.92	96.97	88.76
PBX1	176310	100	98.91	92.04	79.97
PC	608786	100	100	99.95	96.50
PCCA	232000	100	99.02	95.93	90.63
PCCB	232050	100	100	100	98.60
PCDH19	300460	100	100	100	98.04
PCGF2	600346	100	97.77	94.47	87.46
PCLO	604918	99.92	99.76	99.57	98.68
PCNT	605925	100	99.96	99.08	93.59
PDE4D	600129	100	100	100	100
PDHA1	300502	100	100	97.74	86.32
PDP1	605993	100	100	100	100
PDSS1	607429	100	100	100	100
PDSS2	610564	100	100	97.94	93.22
PEPD	613230	100	100	99.80	91.29
PET100	614770	100	100	100	100
PEX1	602136	100	99.27	96.71	92.61
PEX10	602859	100	100	100	95.75
PEX11B	603867	100	100	100	90.14
PEX12	601758	100	100	100	100
PEX13	601789	100	100	100	96.74
PEX16	603360	100	100	96.58	70.67
PEX19	600279	100	100	98.19	86.96
PEX2	170993	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
PEX26	608666	100	100	100	97.05
PEX3	603164	100	100	100	98.79
PEX5	600414	100	100	100	95.40
PEX6	601498	100	99.44	96.19	88.40
PEX7	601757	100	100	98.16	87.17
PGAP1	611655	100	98.06	92.79	82.03
PGAP2	615187	100	100	100	98.69
PGAP3	611801	100	100	100	100
PGK1	311800	100	100	100	95.68
PHACTR1	608723	80.56	80.56	76.94	65
PHC1	602978	100	100	97.83	89.49
PHF21A	608325	100	99.96	97.88	87.88
PHF23	612910	100	100	100	95.87
PHF6	300414	100	98.24	90.39	61.14
PHF8	300560	100	100	98.08	90.76
PHGDH	606879	100	100	100	96.57
PHIP	612870	99.20	97.21	94.90	89.74
PI4KA	600286	100	99.27	96.68	91.65
PIGA	311770	100	100	96.12	95.35
PIGC	601730	100	100	100	100
PIGG	616918	100	100	99.78	96.62
PIGL	605947	100	100	100	100
PIGN	606097	100	99.81	95.39	79.08
PIGO	614730	100	99.96	98.18	93.15
PIGT	610272	100	100	99.14	91.41
PIGV	610274	100	100	100	100
PIGW	610275	100	100	100	100
PIGY	610662	100	100	100	100
PIK3CA	171834	100	100	100	98.85
PIK3R1	171833	100	100	100	98.58
PIK3R2	603157	93.42	90.78	89.90	85.23
PLA2G6	603604	100	100	99.74	96.69
PLCB1	607120	100	100	99.53	94.06
PLCH1	612835	100	100	100	98.32
PLK4	605031	100	100	100	97.35
PLP1	300401	100	100	97.94	87.28
PLPBP	604436	100	100	89.39	87.92
PLXNA3	300022	100	98.75	96.20	85.57
PLXND1	604282	99.21	98.08	96.81	89.51

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PMM2	601785	100	100	100	98.16
PMPCA	613036	100	100	99.05	93.22
PNKP	605610	100	100	98.86	93.95
PNP	164050	100	100	100	100
PNPO	603287	100	100	100	99.51
POC1A	614783	100	100	100	97.36
POGZ	614787	100	100	99.96	97.23
POLA1	312040	100	99.72	96.89	86.82
POLG	174763	100	100	99.06	93.20
POLR2B	180661	100	100	99.95	98.92
POLR3A	614258	100	100	100	99.38
POLR3B	614366	100	100	100	97.68
POLRMT	601778	100	97.15	94.62	83.53
POMGNT1	606822	100	100	100	99.44
POMGNT2	614828	100	100	100	100
POMK	615247	100	100	100	100
POMT1	607423	100	100	99.50	95.82
POMT2	607439	100	100	99.96	97.88
PORCN	300651	100	97.56	90.53	77.48
POU1F1	173110	100	91.84	91.79	87.61
POU3F3	602480	71.68	63.12	56.09	44.67
PPFIA4	603145	100	100	98.77	89.54
PPIL1	601301	100	100	100	100
PPM1D	605100	100	100	100	100
PPOX	600923	100	100	100	94.97
PPP1CB	600590	100	100	100	98.60
PPP1R15B	613257	100	100	100	100
PPP2CA	176915	100	100	100	97.83
PPP2R1A	605983	100	100	99.78	96.20
PPP2R5B	601644	100	100	99.00	84.78
PPP2R5C	601645	97.72	97.72	95.16	93.24
PPP2R5D	601646	100	100	100	97.46
PPP3CA	114105	100	100	100	98.62
PPT1	600722	100	100	100	96.43
PQBP1	300463	100	100	100	94.75
PRIM1	176635	100	100	99.41	96.55
PRKAR1A	188830	100	100	100	100
PRKAR1B	176911	100	100	99.66	93.04
PRKCE	176975	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PRMT7	610087	95.92	95.92	95.92	94.62
PRODH	606810	99.13	95.96	93.53	87.26
CDC40	613799	100	98.33	94.26	88.54
PRPS1	311850	100	100	100	98.76
PRR12	616633	99.92	97.82	92.93	79.57
PRSS12	606709	100	100	100	98.44
PSAP	176801	100	100	100	95.80
PSAT1	610936	100	100	100	95.58
PSMD12	604450	100	100	100	100
PSPH	172480	100	98.77	93.61	73.16
PTCH1	601309	96.46	96.46	96.46	96.46
PTCHD1	300828	100	100	99.08	95.83
PTDSS1	612792	100	100	100	99.11
PTEN	601728	100	100	100	100
PTF1A	607194	100	97.85	85.01	50.63
PTGR1	601274	99.91	98.63	96.96	94.26
PTPN11	176876	98.37	98.37	98.37	98.37
PTRH2	608625	100	100	100	100
PTRHD1	617342	100	100	100	100
PTS	612719	100	100	100	100
PUF60	604819	100	100	98.20	90.80
PUM1	607204	100	100	100	98.87
PURA	600473	100	94.88	90.47	87.32
PUS1	608109	100	100	98.93	91.01
PUS3	616283	100	100	100	100
PUS7	616261	100	100	100	99.91
PYCR1	179035	100	100	100	96.80
PYCR2	616406	100	100	98.48	86.83
QARS1	603727	100	100	100	98.82
QDPR	612676	100	100	98.97	85.67
RAB18	602207	100	92.49	88.09	78.21
RAB27A	603868	100	100	100	100
RAB39B	300774	100	100	100	100
RAB3GAP1	602536	100	100	100	99.51
RAB3GAP2	609275	100	99.48	98.16	95.87
RAB40AL	300405	100	100	100	100
RAC1	602048	92.88	92.88	92.88	92.76
RAD21	606462	100	100	99.58	96.96
RAD50	604040	100	99.35	95.53	82.44



HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
RAF1	164760	100	100	99.87	97.21
RAI1	607642	100	100	100	98.89
RALA	179550	100	95.77	84.03	71.70
RARB	180220	100	100	100	97.77
RARS2	611524	100	100	99.95	97.60
RBBP8	604124	100	100	99.41	95.13
RBFOX1	605104	100	100	99.91	97.30
RBM10	300080	99.99	97.66	94.82	80.06
RBM28	612074	100	100	100	99.64
RBPJ	147183	100	100	98.87	96.74
RCBTB1	607867	100	100	100	100
RELN	600514	100	100	99.94	98.49
RERE	605226	99.03	97.22	95.47	83.04
REV3L	602776	99.15	98.01	98.01	97.08
RFT1	611908	100	99.50	98.38	90.04
RHEB	601293	91.22	89.89	89.89	75.14
RHOBTB2	607352	100	100	100	99.10
RIT1	609591	100	100	100	100
RLIM	300379	100	100	100	99.48
RMND1	614917	100	100	100	94.86
RMRP	157660	No coverage data			
RNASEH2A	606034	100	100	100	95.93
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
RNASET2	612944	100	100	96.89	86.08
RNF113A	300951	100	100	100	99.38
RNF125	610432	100	100	100	94.61
RNF135	611358	91.87	86.45	75.97	51.69
ROGDI	614574	100	98.24	94.45	83.30
RORA	600825	100	100	100	99.06
RPGRIP1L	610937	99.64	97.81	94.99	91.37
RPL10	312173	100	100	100	100
RPS6KA3	300075	100	99.68	96.65	84.98
RSPRY1	616585	100	100	100	100
RTKL1	608833	100	100	99.83	95.32
RTTN	610436	100	100	99.96	96.53
RUBCN	613516	100	98.91	97.44	92.41
RUSC2	611053	100	100	99.33	93.90
RXYLT1	605862	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
SALL1	602218	100	100	100	98.69
SAMHD1	606754	100	100	100	100
SATB1	602075	100	100	99.77	94.66
SATB2	608148	100	99.50	98.04	90.79
SBDS	607444	100	100	100	100
SC5D	602286	100	100	100	100
SCAF4	616023	99.10	97.47	95.80	91.32
SCAMP5	613766	100	100	97.03	80.42
SCARB2	602257	100	100	100	99.04
SCN1A	182389	100	100	100	99.87
SCN1B	600235	93.06	93.06	93.06	92.71
SCN2A	182390	100	100	99.88	98.13
SCN3A	182391	100	100	100	99.27
SCN8A	600702	100	100	99.94	98.39
SCO1	603644	100	100	100	100
SCO2	604272	100	100	100	100
SDHA	600857	100	96.38	96.38	95.00
SEMA3E	608166	100	98.59	95.80	92.61
SEPSECS	613009	100	100	100	99.45
SERAC1	614725	100	100	100	98.51
SETBP1	611060	100	100	100	99.87
SETD1A	611052	99.20	97.46	95.33	91.28
SETD1B	611055	98.11	97.15	95.36	85.27
SETD2	612778	100	99.35	97.90	96.38
SETD5	615743	100	100	99.89	97.17
SF1	601516	83.42	79.66	74.39	64.95
SGSH	605270	93.52	93.52	93.52	93.52
SH3KBP1	300374	100	99.02	97.43	90.71
SHANK2	603290	97.94	97.94	97.15	92.70
SHANK3	606230	93.53	88.94	82.01	68.36
SHH	600725	100	100	100	100
SHOC2	602775	100	100	100	98.74
SIK1	605705	100	100	100	99.83
SIL1	608005	100	100	98.91	93.28
SIN3A	607776	100	100	99.38	97.80
SIX3	603714	100	100	97.64	93.34
SKI	164780	97.53	92.04	86.04	74.94
SLC12A6	604878	100	100	99.59	97.19
SLC13A5	608305	100	100	100	97.38

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
SLC16A2	300095	100	97.44	89.35	79.62
SLC17A5	604322	93.58	93.58	93.58	91.22
SLC19A3	606152	99.94	99.94	99.94	99.94
SLC1A1	133550	100	100	100	99.53
SLC1A2	600300	100	100	99.77	94.70
SLC1A4	600229	100	100	100	99.62
SLC25A12	603667	100	100	100	99.62
SLC25A15	603861	100	100	100	100
SLC25A19	606521	100	100	99.91	97.04
SLC25A22	609302	100	100	100	98.96
SLC2A1	138140	100	97.73	97.73	97.73
SLC33A1	603690	100	100	100	98.25
SLC35A1	605634	100	98.77	96.30	95.28
SLC35A2	314375	100	100	86.85	83.56
SLC35A3	605632	100	100	100	98.88
SLC35C1	605881	100	100	100	100
SLC39A12	608734	100	100	99.63	97.17
SLC39A8	608732	100	100	100	97.08
SLC4A4	603345	100	100	100	98.67
SLC6A1	137165	100	95.11	88.30	81.92
SLC6A17	610299	100	99.33	97.17	82.22
SLC6A3	126455	100	100	99.32	95.75
SLC6A8	300036	99.75	89.01	70.12	47.02
SLC7A7	603593	100	100	100	99.30
SLC9A6	300231	100	99.88	98.12	92.22
SLC9A7	300368	100	96.45	92.47	71.76
SMAD4	600993	100	100	100	99.36
SMARCA2	600014	100	100	100	100
SMARCA4	603254	100	100	99.63	94.75
SMARCB1	601607	100	100	100	98.78
SMARCC2	601734	100	100	99.77	92.80
SMARCE1	603111	100	100	100	98.12
SMC1A	300040	100	100	100	93.45
SMC3	606062	100	99.68	96.35	87.44
SMOC1	608488	100	97.99	93.27	88.67
SMPD1	607608	100	100	100	98.48
SMPD4	610457	100	100	99.33	90.56
SMS	300105	100	100	98.75	94.42
SNAP25	600322	100	100	100	98.02

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
SNAP29	604202	100	100	100	99.43
SNIP1	608241	100	100	98.94	95.70
SNRNP70	180740	100	100	100	95.41
SNRPN	182279	100	100	100	92.43
SNX14	616105	99.91	94.90	87.30	71.21
SOBP	613667	97.99	95.62	92.77	82.86
SON	182465	99.99	99.92	99.27	97.01
SOS1	182530	100	98.47	97.56	94.16
SOS2	601247	99.48	98.09	95.96	88.50
SOX10	602229	100	100	98.32	91.53
SOX11	600898	100	100	97.02	83.88
SOX2	184429	100	100	99.23	93.41
SOX3	313430	100	99.04	90.83	63.55
SOX5	604975	100	100	100	97.68
SPAST	604277	100	100	98.24	88.12
SPATA5	613940	100	100	100	98.88
SPECC1L	614140	100	100	100	100
SPEN	613484	100	100	100	99.73
SPG11	610844	100	100	99.63	96.70
SPOCK1	602264	100	99.31	92.06	88.40
SPRED1	609291	100	100	100	100
SPTAN1	182810	100	100	99.64	96.79
SPTBN1	182790	100	100	99.74	97.38
SPTBN2	604985	100	98.56	95.52	87.30
SRCAP	611421	100	100	99.70	96.68
SRD5A3	611715	100	100	98.10	93.45
SRPX2	300642	100	100	99.06	93.98
SSR4	300090	100	100	100	98.12
ST3GAL3	606494	100	100	100	100
ST3GAL5	604402	100	100	100	99.47
STAG1	604358	100	97.88	97.88	95.20
STAMBP	606247	100	100	100	99.70
STEEP1	301012	No coverage data			
STIL	181590	100	100	100	98.97
STRA6	610745	100	100	100	97.57
STRADA	608626	100	100	100	99.22
STT3A	601134	100	100	100	100
STT3B	608605	100	96.96	94.32	89.01
STX1B	601485	100	98.59	95.30	88.39

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
STXBP1	602926	100	100	100	97.53
SUCLA2	603921	100	100	100	99.44
SUCLG1	611224	100	100	100	98.77
SUCO	619434	99.57	96.83	94.36	90.02
SUMF1	607939	100	100	100	100
SUOX	606887	100	100	100	98.55
SURF1	185620	100	100	100	99.93
SUZ12	606245	96.53	84.13	78.01	69.20
SYN1	313440	100	94.78	81.77	64.80
SYNCRIP	616686	100	100	100	99.98
SYNE1	608441	100	99.79	98.83	94.14
SYNGAP1	603384	98.03	98.03	97.55	92.81
SYP	313475	100	94.38	86.40	64.54
SYT1	185605	100	100	99.16	92.67
SYT14	610949	100	99.93	98.64	95.33
SZT2	615463	100	99.68	98.79	94.25
TAF1	313650	100	99.89	99.48	96.46
TAF2	604912	100	100	99.73	94.79
TANC2	615047	100	100	99.76	97.45
TAT	613018	100	100	100	100
TBC1D20	611663	94.89	93.43	93.43	93.43
TBC1D24	613577	100	100	99.45	96.51
TBC1D7	612655	100	100	100	97.66
TBCD	604649	98.98	95.41	94.07	87.84
TBCE	604934	100	100	99.18	95.59
TBCK	616899	99.84	97.44	94.78	87.39
TBL1XR1	608628	100	100	98.79	94.48
TBP	600075	100	100	100	100
TBR1	604616	100	100	100	100
TBX1	602054	89.95	75.58	63.04	42.43
TCF12	600480	100	100	100	99.68
TCF20	603107	100	100	100	100
TCF4	602272	100	99.49	98.62	94.81
TCN2	613441	100	100	100	96.44
TCTN3	613847	100	100	100	98.86
TDP2	605764	100	100	100	94.13
TECPR2	615000	100	100	100	98.75
TECR	610057	100	99.27	88.20	67.21
TELO2	611140	99.93	98.40	95.65	83.00

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
TFAP2A	107580	100	100	100	94.92
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	97.70
TGIF1	602630	100	100	100	100
TH	191290	100	98.55	94.77	85.86
THG1L	618802	100	100	100	100
THOC2	300395	99.25	94.90	88.74	73.36
THOC6	615403	100	100	100	92.83
THRB	190160	100	100	100	99.22
TIMM8A	300356	100	100	100	90.18
TINF2	604319	100	100	100	100
TLK2	608439	100	100	99.95	95.65
TM4SF20	615404	100	100	100	100
TMCO1	614123	100	100	100	89.40
TMEM165	614726	100	100	100	96.98
TMEM231	614949	100	100	100	97.47
TMEM237	614423	100	100	96.66	94.14
TMEM240	616101	100	100	96.66	87.15
TMEM67	609884	100	100	99.45	90.94
TMEM70	612418	100	100	96.90	88.99
TMLHE	300777	88.67	86.74	85.14	77.16
TMPRSS7	No ID	100	100	100	99.06
TMTC3	617218	100	100	98.93	89.87
TMX2	616715	100	100	100	100
TNIK	610005	100	99.24	96.75	90.90
TNPO2	603002	100	100	100	98.97
TOE1	613931	100	100	100	100
TOGARAM1	617618	100	100	99.18	95.78
TPI1	190450	100	97.24	94.44	90.63
TPO	606765	100	99.71	97.63	91.41
TPP1	607998	100	100	98.00	93.71
TRAIP	605958	100	100	100	100
TRAPPC11	614138	100	99.97	98.09	95.05
TRAPPC6B	610397	100	100	100	81.40
TRAPPC9	611966	98.96	98.96	98.37	94.01
TREX1	606609	100	100	100	100
TRIM32	602290	100	100	100	100
TRIM8	606125	100	100	100	99.44
TRIO	601893	98.72	98.26	97.75	95.72

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
TRIP12	604506	100	100	99.84	97.89
TRMT1	611669	100	99.30	96.36	83.41
TRMT10A	616013	100	100	100	99.91
TRPM3	608961	100	100	97.74	92.71
TSC1	605284	100	100	100	99.95
TSC2	191092	100	100	100	97.97
TSEN15	608756	100	100	100	76
TSEN54	608755	96.30	95.77	94.33	85.79
TSHB	188540	100	100	100	100
TSPAN7	300096	100	100	100	94.93
TTC19	613814	100	100	100	98.52
TTC37	614589	100	100	100	98.33
TTC8	608132	100	100	100	100
TTI2	614426	100	100	100	100
TUBA1A	602529	100	100	100	100
TUBA8	605742	98.41	98.41	98.41	98.41
TUBB	191130	100	100	100	98.37
TUBB2A	615101	100	99.61	94.56	75.58
TUBB2B	612850	100	100	99.61	81.13
TUBB3	602661	100	99.30	94.62	94.62
TUBB4A	602662	100	94.98	87.57	87.57
TUBG1	191135	100	100	100	100
TUBGCP2	617817	100	100	99.51	93.63
TUBGCP4	609610	100	100	99.79	97.71
TUBGCP6	610053	100	100	100	98.75
TUSC3	601385	100	100	100	99.68
TWIST1	601622	100	89.22	81.55	65.97
TWNK	606075	100	100	100	100
UBA5	610552	99.66	94.59	90.70	76.65
UBE2A	312180	100	100	100	93.92
UBE3A	601623	100	100	100	97.29
UBE3B	608047	100	100	100	100
UBR1	605981	100	100	99.80	95.09
UGDH	603370	100	100	100	97.88
UGP2	191760	100	100	100	99.56
UNC80	612636	98.01	97.96	97.31	93.09
UPB1	606673	100	100	100	100
UPF3B	300298	97.30	86.07	78.61	64.50
UQCC2	614461	100	100	100	99.89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
UQCRQ	612080	100	100	100	100
UROC1	613012	100	100	100	96.69
USP18	607057	95.21	95.21	95.21	93.58
USP27X	300975	100	100	100	100
USP7	602519	97.48	97.48	97.48	97.10
USP9X	300072	100	99.95	99.10	92.29
UTRN	128240	100	100	99.88	96.51
VAMP2	185881	100	100	92.51	92.51
VLDLR	192977	100	100	97.02	96.50
VPS11	608549	100	100	100	98.53
VPS13B	607817	99.37	98.22	97.00	90.85
VPS13C	608879	98.55	97.73	96.94	92.88
VPS16	608550	100	100	98.52	91.21
VPS37A	609927	100	99.29	97.66	89.72
VPS41	605485	100	100	97.55	97.55
VPS50	616465	99.30	95.86	93.86	85.60
VPS53	615850	100	100	100	97.84
VRK1	602168	100	100	100	96.88
VWA3B	614884	100	100	99.11	96.43
WAC	615049	99.37	97.25	96.62	91.18
WASHC4	615748	100	99.84	97.87	91.83
WASHC5	610657	100	100	100	98.17
WBP11	618083	100	98.23	95.61	88.43
WDR13	300512	100	100	100	97.22
WDR19	608151	100	100	99.54	96.20
WDR26	617424	91.96	86.99	85.27	82.42
WDR4	605924	100	100	97.65	84.66
WDR45	300526	100	100	100	97.31
WDR62	613583	100	100	99.32	94.62
WDR73	616144	100	100	100	100
WDR81	614218	100	99.68	98.13	91.95
WFS1	606201	100	100	99.45	97.03
WWOX	605131	100	100	100	87.63
XIAP	300079	100	100	99.35	89.29
XPA	611153	100	99.95	94.99	79.45
XPC	613208	100	100	99.08	89.94
XPNPEP3	613553	100	100	100	99.51
XYLT1	608124	98.57	94.72	91.00	86.04
YAP1	606608	100	98.30	95.04	88.85



HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
YIF1B	619109	100	100	99.60	89.08
YME1L1	607472	100	99.23	96.25	84.87
YWHAE	605066	100	98.31	90.45	84.52
YY1	600013	100	100	95.49	83.31
ZBTB16	176797	100	100	100	100
ZBTB18	608433	100	100	100	99.72
ZBTB20	606025	100	100	99.46	97.24
ZBTB24	614064	100	100	100	100
ZC3H14	613279	100	100	99.25	92.96
ZC4H2	300897	100	100	100	94.95
ZDHHC15	300576	100	94.07	76.48	76.48
ZDHHC9	300646	100	100	95.28	71.54
ZEB2	605802	100	100	100	100
ZFYVE26	612012	100	100	99.51	95.91
ZIC1	600470	100	100	98.72	93.11
ZIC2	603073	92.27	89.10	87.56	84.00
ZIC4	608948	100	100	100	100
ZMYM2	602221	100	100	99.91	97.21
ZMYND11	608668	100	100	100	100
ZNF101	603983	100	100	100	100
ZNF292	616213	100	100	100	99.57
ZNF407	615894	100	100	100	100
ZNF41	314995	100	100	100	99.78
ZNF462	617371	100	99.88	99.51	98.09
ZNF526	614387	100	100	100	100
ZNF592	613624	100	100	100	99.62
ZNF674	300573	100	100	100	100
ZNF711	314990	100	100	99.61	93.55
ZNF81	314998	71.70	71.70	68.30	54.26
ZSWIM6	615951	95.13	92.52	89.92	84.84

- OMIM release used: 23-9-2021

- The statistics above are based on a set of 150 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ( $\pm 10bp$  flanking introns) that is covered at least 10x, 20x, 30x or 50x