

Whole Exome Sequencing

Gene package Prenatal, version 1, 1-11-2020



Technical information

DNA was enriched using Agilent SureSelect DNA + 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/>). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. 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 types of sequence changes).



Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
A2ML1	610627	128	100	100	100
A4GALT	607922	341	100	100	100
AAAS	605378	175	100	100	100
AAGAB	614888	91	100	100	98
AARS1	601065	161	100	100	100
AARS2	612035	239	100	100	100
AASS	605113	87	100	98	93
ABAT	137150	142	100	100	100
ABCA1	600046	128	100	100	99
ABCA12	607800	91	100	97	93
ABCA3	601615	281	100	100	100
ABCA4	601691	157	100	99	97
ABCB11	603201	114	100	99	97
ABCB4	171060	98	98	96	91
ABCB6	605452	217	100	100	100
ABCB7	300135	64	100	94	83
ABCC2	601107	101	100	97	95
ABCC6	603234	201	100	100	99
ABCC8	600509	242	100	100	100
ABCC9	601439	77	100	99	94
ABCD1	300371	178	100	100	100
ABCD4	603214	169	100	100	100
ABCG2	603756	79	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCG5	605459	115	100	99	91
ABCG8	605460	188	100	100	100
ABHD12	613599	133	100	99	93
ABHD5	604780	116	100	96	92
ABL1	189980	215	100	100	100
ACACA	200350	97	99	99	97
ACAD8	604773	186	100	100	100
ACAD9	611103	186	100	100	100
ACADM	607008	51	93	80	68
ACADS	606885	349	100	100	100
ACADSB	600301	74	100	99	94
ACADVL	609575	238	100	100	100
ACAN	155760	234	93	92	92
ACAT1	607809	65	96	91	80
ACAT2	100678	156	100	100	100
ACBD5	616618	104	100	100	97
ACE	106180	209	100	100	100
ACO2	100850	270	100	100	100
ACOT9	300862	63	100	97	90
ACOX1	609751	126	100	100	98
ACP5	171640	280	100	100	100
ACSF3	614245	244	100	100	100
ACSL4	300157	47	97	86	72
ACSL6	604443	135	100	100	100
ACTA1	102610	234	100	100	100
ACTA2	102620	142	100	100	100
ACTB	102630	253	100	100	100
ACTC1	102540	173	100	100	100
ACTG1	102560	233	100	100	100
ACTG2	102545	169	100	100	100
ACTN1	102575	171	100	100	99
ACTN4	604638	258	100	100	100
ACVR1	102576	104	100	100	99
ACVR1B	601300	157	100	100	97
ACVR2B	602730	272	100	100	100
ACVRL1	601284	298	100	100	100
ACY1	104620	215	100	100	100
ADA	608958	161	100	100	100
ADA2	607575	157	100	100	100
ADAM10	602192	67	100	94	86
ADAM17	603639	98	99	94	88
ADAM22	603709	101	100	99	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ADAM9	602713	75	100	96	92
ADAMTS10	608990	267	100	100	100
ADAMTS13	604134	267	100	100	99
ADAMTS17	607511	177	98	96	94
ADAMTS18	607512	136	100	100	99
ADAMTS19	607513	87	99	96	90
ADAMTS2	604539	253	98	96	96
ADAMTSL2	612277	97	47	45	44
ADAMTSL4	610113	244	100	100	100
ADAR	146920	163	100	100	100
ADAT3	615302	319	100	100	100
ADCY5	600293	259	100	99	98
ADGRG1	604110	233	100	100	100
ADGRV1	602851	98	99	94	89
ADIPOQ	605441	195	100	100	100
ADK	102750	58	99	89	81
ADNP	611386	160	100	100	98
ADRB2	109690	208	100	100	98
ADSL	608222	178	100	100	100
AFF2	300806	70	100	97	90
AFF4	604417	119	100	98	94
AFG3L2	604581	107	96	96	93
AGA	613228	83	100	98	90
AGAP2	605476	215	100	100	100
AGBL1	615496	134	100	100	98
AGBL2	617345	93	95	94	90
AGK	610345	97	100	97	97
AGL	610860	68	98	91	83
AGO1	606228	134	100	100	100
AGO2	606229	184	99	97	93
AGPAT2	603100	227	100	100	100
AGPS	603051	64	96	94	85
AGRN	103320	290	100	100	100
AGT	106150	275	100	100	100
AGTPBP1	606830	63	95	86	73
AGTR1	106165	97	100	100	100
AGTR2	300034	75	100	100	99
AGXT	604285	248	100	100	100
AGXT2	612471	118	100	100	97
AHCY	180960	268	100	100	100
AHDC1	615790	285	100	100	100
AHI1	608894	65	96	86	77

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
AHNAK2	No ID	241	100	100	100
AICDA	605257	191	100	100	100
AIFM1	300169	89	100	99	96
AIMP1	603605	70	99	88	81
AIP	605555	251	100	100	100
AIPL1	604392	227	100	100	100
AIRE	607358	248	100	100	100
AK1	103000	147	100	97	94
AK2	103020	99	100	100	98
AKAP9	604001	61	96	85	74
AKR1C2	600450	131	91	91	91
AKR1C4	600451	127	100	100	100
AKR1D1	604741	87	100	100	97
AKT1	164730	284	100	100	100
AKT2	164731	240	100	100	100
AKT3	611223	53	100	90	75
ALAD	125270	217	100	100	100
ALAS2	301300	102	100	100	100
ALB	103600	61	100	97	88
ALDH18A1	138250	169	100	100	99
ALDH1A3	600463	129	100	100	99
ALDH2	100650	208	100	100	100
ALDH3A2	609523	90	100	100	97
ALDH4A1	606811	315	100	100	100
ALDH5A1	610045	121	100	100	98
ALDH6A1	603178	116	100	100	99
ALDH7A1	107323	105	100	100	95
ALDOA	103850	278	100	100	100
ALDOB	612724	127	100	100	100
ALG1	605907	126	100	100	99
ALG11	613666	109	100	100	95
ALG12	607144	224	100	100	100
ALG13	300776	67	98	92	81
ALG14	612866	108	100	100	100
ALG2	607905	173	100	100	100
ALG3	608750	213	100	100	100
ALG6	604566	49	90	81	67
ALG8	608103	62	93	86	79
ALG9	606941	78	97	95	92
ALMS1	606844	116	100	99	96
ALOX12B	603741	262	100	100	100
ALOXE3	607206	206	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ALPK1	607347	122	100	99	96
ALPL	171760	323	100	100	100
ALS2	606352	120	100	98	96
ALX1	601527	188	100	100	94
ALX3	606014	202	100	98	96
ALX4	605420	206	100	99	97
AMACR	604489	129	100	100	99
AMELX	300391	95	98	83	79
AMER1	300647	240	100	100	100
AMH	600957	179	100	100	100
AMHR2	600956	209	100	100	100
AMMECR1	300195	93	100	100	100
AMN	605799	200	100	100	100
AMPD1	102770	90	100	99	95
AMPD2	102771	219	100	100	100
AMT	238310	225	100	100	100
ANG	105850	136	100	100	100
ANGPTL3	604774	54	100	90	71
ANK1	612641	237	100	100	98
ANK2	106410	115	100	99	98
ANK3	600465	118	100	99	98
ANKEF1	No ID	86	100	99	96
ANKH	605145	171	100	100	100
ANKK1	608774	270	100	100	100
ANKLE2	616062	152	99	97	92
ANKRD1	609599	83	87	86	85
ANKRD11	611192	247	100	99	97
ANKRD26	610855	43	74	58	48
ANKS6	615370	196	99	93	92
ANO10	613726	82	98	93	85
ANO3	610110	61	100	98	90
ANO5	608662	61	94	87	75
ANO6	608663	76	99	94	87
ANOS1	300836	85	100	98	94
ANTXR1	606410	116	99	98	95
ANTXR2	608041	77	100	99	91
AP1S1	603531	171	100	100	100
AP1S2	300629	33	95	74	53
AP2S1	602242	209	100	100	100
AP3B1	603401	58	93	79	66
AP3B2	602166	182	100	100	100
AP4B1	607245	130	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
AP4E1	607244	66	97	87	75
AP4M1	602296	141	100	100	100
AP4S1	607243	50	100	96	78
AP5Z1	613653	253	100	100	100
APC2	612034	235	100	99	97
APCDD1	607479	256	100	100	100
APOA1	107680	213	100	100	100
APOA2	107670	138	100	100	100
APOA5	606368	469	100	100	100
APOB	107730	309	100	100	99
APOC2	608083	138	100	100	100
APOC3	107720	161	100	100	100
APOE	107741	151	100	100	100
APRT	102600	203	100	100	100
APTX	606350	166	100	98	90
AQP2	107777	250	100	100	100
AQP5	600442	169	100	100	100
AR	313700	134	100	99	96
ARFGEF2	605371	114	100	97	93
ARG1	608313	89	100	99	95
ARHGAP26	605370	114	100	100	97
ARHGAP31	610911	186	100	100	100
ARHGAP4	300023	151	100	100	100
ARHGEF10	608136	162	100	100	99
ARHGEF12	604763	96	99	96	93
ARHGEF33	No ID	101	100	96	90
ARHGEF6	300267	51	99	91	78
ARHGEF9	300429	93	100	100	99
ARID1A	603024	182	100	98	97
ARID1B	614556	166	100	100	98
ARID2	609539	109	100	96	92
ARL13B	608922	56	100	90	72
ARL2BP	615407	81	100	98	90
ARL6	608845	44	98	88	79
ODAD2	615408	81	92	85	78
ARNT	126110	99	100	100	98
ARNT2	606036	174	100	100	100
ARSA	607574	380	100	100	100
ARSB	611542	130	100	100	98
ARSL	300180	101	100	100	100
ARX	300382	100	92	89	84
ASAH1	613468	73	98	92	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ASB10	615054	345	100	100	100
ASCC1	614215	101	100	97	95
ASCL1	100790	249	100	100	100
ASH1L	607999	115	100	100	99
ASL	608310	294	100	100	100
GET3	601913	201	100	100	100
ASNS	108370	82	100	98	91
ASPA	608034	103	100	98	94
ASPM	605481	64	94	87	75
ASPSCR1	606236	202	100	100	100
ASS1	603470	215	100	98	96
ASXL1	612990	187	100	98	97
ASXL2	612991	150	97	97	97
ASXL3	615115	119	100	97	94
ATAD3A	612316	313	98	97	94
ATAD3B	612317	336	98	95	90
ATCAY	608179	200	100	100	100
ATIC	601731	84	100	99	94
ATL1	606439	101	98	98	93
ATL3	609369	83	96	96	95
ATM	607585	61	96	86	75
ATN1	607462	189	100	100	100
ATP13A2	610513	228	100	100	100
ATP1A2	182340	189	100	100	100
ATP1A3	182350	221	100	100	100
ATP2A1	108730	235	100	100	100
ATP2A2	108740	119	100	99	95
ATP2C1	604384	70	100	95	87
ATP5F1C	108729	92	95	95	94
ATP5F1E	606153	95	100	100	100
ATP6AP1	300197	134	100	100	99
ATP6AP2	300556	54	100	93	81
ATP6V0A2	611716	120	100	97	91
ATP6V0A4	605239	104	100	99	98
ATP6V1B1	192132	206	100	100	100
ATP6V1B2	606939	94	100	99	98
ATP7A	300011	59	99	94	81
ATP7B	606882	195	100	100	100
ATP8A2	605870	125	98	96	94
ATP8B1	602397	84	99	92	86
ATPAF2	608918	175	100	100	97
ATR	601215	76	98	92	83

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ATRIP	606605	189	100	98	92
ATRX	300032	37	92	74	52
ATXN1	601556	263	100	100	100
ATXN10	611150	85	99	98	95
ATXN2	601517	92	91	82	75
ATXN3	607047	75	93	92	83
ATXN7	607640	116	99	96	94
ATXN8OS	603680	No coverage data			
AUH	600529	81	100	98	91
AURKC	603495	145	100	100	100
AUTS2	607270	204	100	99	98
AVP	192340	144	100	100	100
AVPR2	300538	182	100	100	100
AXIN1	603816	218	100	100	99
AXIN2	604025	193	100	100	100
B2M	109700	131	100	100	100
B3GALNT1	603094	109	100	100	99
B3GALNT2	610194	101	100	100	99
B3GALT6	615291	283	83	80	78
B3GAT3	606374	243	100	100	100
B3GLCT	610308	64	97	84	73
B4GALNT1	601873	269	100	100	100
B4GALT1	137060	136	100	100	100
B4GALT7	604327	254	100	100	100
B4GAT1	605517	291	100	100	100
B9D1	614144	285	100	100	100
B9D2	611951	336	100	100	100
BAAT	602938	243	100	100	99
BAG3	603883	212	100	100	100
BANF1	603811	209	100	100	100
BAP1	603089	213	100	100	100
BAX	600040	222	100	100	97
BBS1	209901	225	100	100	100
BBS10	610148	100	100	100	99
BBS12	610683	118	100	100	98
BBS2	606151	117	100	99	94
BBS4	600374	100	100	100	93
BBS5	603650	52	91	66	47
BBS7	607590	68	95	82	75
BBS9	607968	73	99	94	88
BCAP31	300398	82	100	100	95
BCHE	177400	89	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
BCKDHA	608348	221	100	100	100
BCKDHB	248611	137	100	100	99
BCKDK	614901	220	100	100	100
BCL10	603517	82	96	85	77
BCL11A	606557	271	100	100	100
BCL11B	606558	242	100	100	99
BCL2	151430	191	100	100	100
BCL7A	601406	121	100	100	100
BCO1	605748	129	100	100	100
BCOR	300485	120	100	100	98
BCORL1	300688	135	100	100	99
BCR	151410	223	100	100	98
BCS1L	603647	263	100	100	100
BDNF	113505	158	100	100	100
BEAN1	612051	201	100	100	100
BEST1	607854	214	100	100	100
BFSP1	603307	138	100	100	100
BFSP2	603212	268	100	100	100
BGN	301870	186	100	100	100
BHMT	602888	99	100	97	90
BICD2	609797	245	100	100	100
BIN1	601248	195	100	100	100
BLK	191305	167	100	100	100
BLM	604610	64	99	90	76
BLNK	604515	89	95	93	88
BLOC1S3	609762	141	100	100	100
BLOC1S6	604310	65	100	78	57
BLVRA	109750	138	100	100	100
BMP1	112264	301	100	100	100
BMP15	300247	111	100	100	100
BMP2	112261	211	100	99	98
BMP4	112262	224	100	100	100
BMPER	608699	116	100	100	99
BMPR1A	601299	103	100	100	96
BMPR1B	603248	79	100	98	92
BMPR2	600799	102	100	100	98
BOLA3	613183	63	100	98	94
BPGM	613896	117	100	100	99
BRAF	164757	84	100	99	94
BRAT1	614506	243	100	100	100
BRF1	604902	149	100	100	98
BRIP1	605882	73	97	91	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
BRPF1	602410	242	100	100	100
BRWD3	300553	47	98	89	77
BSCL2	606158	178	100	100	100
BSND	606412	267	100	100	100
BTD	609019	195	100	100	100
BTK	300300	77	100	99	95
BUB1	602452	100	99	97	93
BUB1B	602860	99	98	95	89
c12orf4	616082	57	94	84	74
C12orf57	615140	308	100	100	100
C12orf65	613541	100	100	94	58
CDIN1	615626	98	100	100	100
C19orf12	614297	235	100	100	100
C1GALT1C1	300611	166	100	100	100
C1orf167	No ID	218	100	100	100
C1QA	120550	288	100	100	100
C1QB	120570	230	100	100	100
C1QC	120575	262	100	100	100
C1QTNF5	608752	352	100	100	100
C1S	120580	126	100	100	97
C2	613927	174	100	100	100
C2CD3	615944	132	100	100	99
C3	120700	210	100	100	100
C4A	120810	50	25	21	19
C4B	120820	60	25	23	22
C5	120900	70	92	83	78
C6	217050	96	100	100	99
C7	217070	107	100	99	93
C8A	120950	141	100	100	100
C8B	120960	125	100	98	97
C8orf37	614477	124	100	93	85
C9	120940	78	100	96	93
C9orf72	614260	88	100	100	97
CA12	603263	158	98	97	97
CA2	611492	116	100	100	100
CA4	114760	236	100	100	100
CA5A	114761	147	100	100	100
CA8	114815	95	100	100	94
CABP2	607314	162	100	100	100
CABP4	608965	258	100	100	100
CACNA1A	601011	139	100	100	99
CACNA1C	114205	179	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CACNA1D	114206	148	100	99	96
CACNA1F	300110	121	100	100	99
CACNA1G	604065	219	100	100	100
CACNA1S	114208	217	100	100	100
CACNA2D1	114204	61	91	79	70
CACNA2D4	608171	207	100	100	100
CACNB2	600003	127	100	97	92
CACNB4	601949	176	100	100	99
CACNG2	602911	166	100	100	98
CAD	114010	218	100	100	100
CALCOCO1	No ID	145	100	100	98
CALM1	114180	77	100	100	100
CALR	109091	166	100	100	100
CALR3	611414	118	99	91	91
CAMK2A	114078	199	100	100	100
CAMK2B	607707	187	100	100	100
CAMTA1	611501	210	100	98	95
CANT1	613165	318	100	100	100
CAPN10	605286	256	100	100	100
CAPN3	114240	135	100	100	100
CAPN5	602537	227	100	100	100
CARD11	607210	214	100	100	100
CARD14	607211	291	100	100	100
CARD9	607212	265	100	100	100
CARS1	123859	184	100	100	96
CASK	300172	60	96	86	70
CASP10	601762	115	100	100	98
CASP8	601763	117	100	97	92
CASQ2	114251	102	100	98	96
CASR	601199	210	100	100	100
CAT	115500	120	100	100	100
CATSPER1	606389	195	100	100	100
CAV1	601047	134	100	100	100
CAV3	601253	178	100	100	100
CAVIN1	603198	225	100	100	100
CBL	165360	130	100	100	99
CBLIF	609342	No coverage data			
CBS	613381	430	100	100	100
CBX2	602770	220	100	99	97
CC2D1A	610055	228	100	100	100
CC2D2A	612013	88	98	93	86
CCBE1	612753	163	95	95	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CCDC103	614677	311	100	100	100
ODAD1	615038	213	100	100	100
CCDC115	613734	184	100	100	100
CCDC13	No ID	150	100	100	99
CCDC14	617147	98	94	90	82
CCDC174	616735	102	87	78	71
CCDC22	300859	158	100	100	100
CCDC39	613798	44	80	58	47
CCDC40	613799	166	100	100	100
CCDC50	611051	94	100	96	89
CCDC65	611088	89	100	98	93
CCDC78	614666	215	100	100	98
CCDC8	614145	247	100	100	100
CCDC88C	611204	187	100	100	98
CCM2	607929	228	100	100	100
CCN6	603400	110	100	99	97
CCNB1	123836	94	100	100	100
CCND2	123833	152	100	100	100
CCNQ	300708	126	81	81	81
CCT5	610150	135	100	99	92
CD151	602243	200	100	100	100
CD19	107265	226	100	100	99
CD247	186780	180	100	100	100
CD27	186711	205	100	100	100
CD2AP	604241	41	96	84	67
CD320	606475	320	100	100	100
CD36	173510	62	95	88	83
CD3D	186790	150	100	100	100
CD3E	186830	103	100	95	90
CD3G	186740	143	100	100	100
CD4	186940	187	100	100	100
CD40	109535	158	100	100	100
CD40LG	300386	61	100	92	77
CD59	107271	136	100	100	100
CD79A	112205	222	100	99	96
CD79B	147245	197	100	100	100
CD81	186845	187	100	100	100
CD8A	186910	221	100	100	100
CD96	606037	96	100	100	99
CDAN1	607465	159	100	100	100
CDC45	603465	172	100	100	100
CDC5L	602868	68	99	91	81

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CDC6	602627	186	100	100	98
CDC73	607393	53	100	92	76
CDH1	192090	120	100	100	98
CDH3	114019	235	100	100	100
CDH23	605516	257	100	100	100
CDH15	114021	173	100	100	100
CDHR1	609502	176	100	100	100
CDK16	311550	117	100	100	100
CDK5	123831	223	100	100	100
CDK5RAP2	608201	113	99	94	90
CDK6	603368	137	98	94	93
CDKL5	300203	74	97	92	86
CDKN1B	600778	257	100	100	100
CDKN1C	600856	156	92	87	83
CDKN2A	600160	272	100	100	100
CDON	608707	123	99	97	95
CDSN	602593	213	100	100	100
CDT1	605525	222	100	100	98
CEACAM16	614591	211	100	100	100
CEBPA	116897	177	100	100	94
CEBPE	600749	294	100	100	100
CEL	114840	254	97	95	91
CENPJ	609279	97	97	93	89
CEP104	616690	94	100	99	95
CEP120	613446	84	99	94	89
CEP135	611423	42	92	74	57
CEP152	613529	79	93	86	81
CEP164	614848	149	100	100	100
CEP19	615586	97	100	100	100
CEP290	610142	35	74	58	45
CEP41	610523	83	100	94	91
CEP57	607951	58	99	89	75
CEP63	614724	50	97	81	65
CEP89	615470	103	95	89	87
CERKL	608381	82	100	94	81
CERS3	615276	66	100	100	92
CES1	114835	132	77	77	77
CETP	118470	174	100	100	100
CFAP298	615494	113	100	91	87
CFAP410	603191	250	100	100	100
CFAP53	614759	78	89	80	74
CFC1	605194	59	33	33	30

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CFD	134350	226	100	100	100
CFH	134370	78	95	89	81
CFHR5	608593	65	98	90	80
CFI	217030	78	96	92	89
CFL2	601443	39	86	79	75
CFP	300383	156	100	100	100
CFTR	602421	124	98	94	88
CHAMP1	616327	157	100	100	100
CHAT	118490	172	100	100	99
CHD2	602119	82	98	92	84
CHD3	602120	152	98	96	95
CHD4	603277	129	100	99	97
CHD7	608892	167	100	99	97
CHD8	610528	185	100	100	99
CHKB	612395	373	100	100	100
CHM	300390	47	87	79	69
CHMP1A	164010	202	100	100	100
CHMP2B	609512	44	85	75	54
CHMP4B	610897	116	100	100	100
CHN1	118423	72	100	95	88
CHRD1	300350	73	100	98	90
CHRM3	118494	196	100	100	100
CHRNA1	100690	121	100	100	100
CHRNA2	118502	314	100	100	100
CHRNA4	118504	386	100	100	99
CHRN1	100710	155	100	100	99
CHRN2	118507	245	100	100	100
CHRN3	100720	163	100	100	100
CHRN4	100725	237	100	100	100
CHRN5	100730	180	100	100	100
CHST14	608429	278	100	100	98
CHST3	603799	345	100	100	100
CHST6	605294	533	100	100	100
CHSY1	608183	141	97	94	94
CHUK	600664	76	99	96	87
CIB2	605564	185	100	100	100
CIC	612082	232	100	100	100
CIITA	600005	286	100	100	100
CISD2	611507	97	76	76	76
CIT	605629	131	100	100	99
CITED2	602937	154	100	100	100
CKAP2L	616174	100	88	88	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CLCF1	607672	225	100	100	100
CLCN1	118425	152	100	100	100
CLCN2	600570	275	100	100	100
CLCN4	302910	141	100	100	98
CLCN5	300008	118	100	96	91
CLCN7	602727	244	100	100	100
CLCNKA	602024	213	100	100	99
CLCNKB	602023	213	100	100	99
CLDN1	603718	125	100	100	100
CLDN14	605608	601	100	100	100
CLDN16	603959	120	100	100	100
CLDN19	610036	369	100	100	100
CLEC16A	611303	141	100	99	96
CLEC7A	606264	76	100	100	97
CLIC2	300138	52	100	95	82
CLIP1	179838	122	96	92	91
CLMP	611693	155	100	100	100
CLN3	607042	211	100	100	100
CLN5	608102	113	100	93	89
CLN6	606725	264	100	100	100
CLN8	607837	195	100	100	100
CLP1	608757	213	100	100	100
CLPB	616254	209	100	100	100
CLPP	601119	139	100	100	100
CLPS	120105	247	100	100	100
CLPX	615611	81	100	99	94
CLRN1	606397	92	100	96	92
CLTC	118955	84	100	98	93
CNBP	116955	117	100	98	95
CNDP1	609064	109	100	100	98
CNGA1	123825	76	96	89	81
CNGA3	600053	183	100	100	100
CNGB1	600724	133	100	100	98
CNGB3	605080	83	97	91	86
CNKS2	300724	49	96	82	65
CNNM2	607803	209	100	100	97
CNNM4	607805	260	100	100	99
CNOT3	604910	203	100	100	99
CNTN1	600016	78	100	98	91
CNTNAP2	604569	119	100	100	99
CNTNAP4	610518	86	99	97	92
COA5	613920	159	100	91	78

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
COASY	609855	239	100	100	100
COCH	603196	137	100	100	99
COG1	606973	164	100	100	99
COG2	606974	81	96	92	89
COG4	606976	150	100	100	100
COG5	606821	73	95	92	85
COG6	606977	66	94	83	74
COG7	606978	172	100	100	100
COG8	606979	164	100	100	100
COL10A1	120110	266	100	100	100
COL11A1	120280	86	99	95	89
COL11A2	120290	186	100	100	99
COL12A1	120320	105	97	95	92
COL17A1	113811	157	100	98	97
COL18A1	120328	247	100	100	100
COL1A1	120150	246	100	100	100
COL1A2	120160	131	98	97	96
COL2A1	120140	203	100	100	99
COL3A1	120180	113	100	98	95
COL4A1	120130	121	100	99	96
COL4A2	120090	196	100	100	99
COL4A3	120070	132	100	99	97
CERT1	604677	95	96	92	89
COL4A4	120131	123	100	99	97
COL4A5	303630	84	99	97	92
COL5A1	120215	212	100	100	100
COL5A2	120190	108	99	99	98
COL6A1	120220	286	100	100	100
COL6A2	120240	353	100	100	100
COL6A3	120250	210	100	100	99
COL7A1	120120	279	100	100	100
COL8A2	120252	153	100	100	100
COL9A1	120210	99	100	98	96
COL9A2	120260	153	100	100	100
COL9A3	120270	185	100	100	100
COLEC10	607620	117	100	100	99
COLEC11	612502	234	100	100	100
COLGALT1	617531	178	100	99	95
COLQ	603033	143	100	98	95
COMP	600310	286	100	100	100
COQ2	609825	79	98	92	82
COQ4	612898	176	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
COQ6	614647	206	100	99	97
COQ8A	606980	250	100	100	100
COQ8B	615567	212	100	100	100
COQ9	612837	123	100	100	100
CORIN	605236	110	100	99	97
CORO1A	605000	287	92	92	92
COX10	602125	177	100	99	94
COX14	614478	154	100	100	100
COX15	603646	137	100	100	100
COX20	614698	62	99	86	64
COX4I2	607976	245	100	100	100
COX6B1	124089	158	100	100	100
COX7B	300885	37	100	55	52
CP	117700	92	99	98	94
CPA6	609562	99	96	94	91
CPLANE1	614571	75	97	93	86
CPN1	603103	169	100	100	100
CPOX	612732	103	100	100	100
CPS1	608307	101	100	100	99
CPT1A	600528	163	100	100	99
CPT2	600650	187	100	100	100
CR2	120650	122	99	99	96
CRADD	603454	228	100	100	96
CRB1	604210	214	100	100	100
CRB2	609720	229	100	100	100
CRBN	609262	78	95	93	86
CREB1	123810	91	100	100	99
CREB3L1	616215	178	100	100	100
CREBBP	600140	275	99	99	98
CRELD1	607170	246	100	99	98
CRLF1	604237	269	93	91	91
CRPPA	614631	76	97	84	77
CRTAP	605497	155	100	100	100
CRTC1	607536	206	100	100	100
CRX	602225	158	100	100	100
CRYAA	123580	511	100	100	100
CRYAB	123590	160	100	100	100
CRYBA1	123610	123	100	100	100
CRYBA4	123631	170	100	100	100
CRYBB1	600929	315	100	100	100
CRYBB2	123620	282	100	100	100
CRYBB3	123630	274	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CRYGB	123670	138	100	100	100
CRYGC	123680	201	100	100	100
CRYGD	123690	258	100	100	100
CRYGS	123730	304	100	100	100
CRYM	123740	142	100	100	100
CSF1R	164770	205	100	100	100
CSF2RA	306250	47	46	45	40
CSF2RB	138981	226	100	100	100
CSF3R	138971	236	100	100	100
CSGALNACT1	616615	159	100	100	98
CSNK1D	600864	194	100	100	100
CSNK2A1	115440	110	100	94	94
CSPP1	611654	68	99	91	80
CSRP3	600824	176	100	100	100
CST3	604312	186	100	100	100
CSTA	184600	45	100	75	66
CSTB	601145	132	100	100	100
CTBP1	602618	244	100	94	92
CTC1	613129	174	100	100	100
CTCF	604167	114	100	96	92
CTDP1	604927	173	100	100	100
CTH	607657	91	100	98	92
CTHRC1	610635	111	100	100	97
CTNNA2	114025	106	100	98	93
CTNNA3	607667	93	100	100	97
CTNNB1	116806	133	100	100	99
CTNND1	601045	149	100	99	95
CTNND2	604275	154	98	96	95
CTNS	606272	147	100	97	90
CTSA	613111	181	100	100	100
CTSC	602365	182	100	100	100
CTSD	116840	278	100	100	100
CTSF	603539	232	100	98	96
CTSK	601105	108	100	100	100
CTTNBP2	609772	124	100	99	95
CUBN	602997	107	100	99	96
CUL3	603136	51	95	83	70
CUL4B	300304	43	90	77	62
CUL7	609577	281	100	100	100
CWF19L1	616120	91	100	94	88
CXCR4	162643	190	100	100	100
CXorf56	301012	66	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
CYB5A	613218	109	100	95	89
CYB5R3	613213	188	100	100	100
CYBA	608508	164	100	98	96
CYBB	300481	65	100	98	91
CYC1	123980	228	100	100	100
CYCS	123970	126	100	100	100
CYLD	605018	81	97	88	74
CYP11A1	118485	205	100	100	100
CYP11B1	610613	301	100	100	100
CYP11B2	124080	299	100	100	100
CYP17A1	609300	201	100	97	94
CYP19A1	107910	101	100	100	98
CYP1B1	601771	364	100	100	100
CYP21A2	613815	193	100	100	100
CYP24A1	126065	138	100	100	100
CYP26B1	605207	414	100	100	100
CYP26C1	608428	210	100	100	100
CYP27A1	606530	295	100	100	100
CYP27B1	609506	256	100	100	100
CYP2A6	122720	238	100	100	100
CYP2B6	123930	178	100	100	98
CYP2C19	124020	123	100	95	86
CYP2C8	601129	88	99	97	93
CYP2C9	601130	115	96	86	83
CYP2R1	608713	135	100	96	92
CYP2U1	610670	143	100	100	99
CYP46A1	604087	148	100	100	100
CYP4F22	611495	225	100	100	100
CYP4V2	608614	105	98	93	88
CYP7A1	118455	117	100	100	99
CYP7B1	603711	111	100	99	94
D2HGDH	609186	269	100	100	100
DAB1	603448	135	100	98	93
DAG1	128239	269	100	100	100
DARS1	603084	56	96	88	76
DARS2	610956	79	100	96	94
DAZL	601486	38	82	65	52
DBH	609312	256	100	100	100
DBT	248610	70	100	93	83
DCAF17	612515	71	100	99	93
DCC	120470	114	100	100	98
DCHS1	603057	292	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
DCLRE1C	605988	151	100	97	91
DCN	125255	113	100	100	100
DCPS	610534	179	100	100	100
DCTN1	601143	162	100	100	99
DCX	300121	102	100	96	89
DCXR	608347	314	100	100	100
DDB2	600811	179	100	100	100
DDC	107930	112	100	95	92
DDHD1	614603	143	100	100	97
DDHD2	615003	71	100	94	87
DDOST	602202	211	100	100	100
DDR2	191311	131	100	100	100
DDX11	601150	196	100	100	100
DDX3X	300160	56	100	98	91
DDX59	615464	94	100	98	93
DEAF1	602635	191	100	100	100
DENND5A	617278	136	100	100	98
DEPDC5	614191	157	100	100	98
DES	125660	178	100	100	100
DGAT1	604900	341	99	96	94
DGKE	601440	149	89	85	81
DGUOK	601465	115	100	100	100
DHCR24	606418	207	100	100	100
DHCR7	602858	243	100	100	100
DHDDS	608172	146	100	100	100
DHFR	126060	57	93	71	43
DHH	605423	335	100	100	100
DHODH	126064	225	100	100	99
DHPS	600944	286	100	100	100
DHTKD1	614984	128	100	100	99
DHX37	617362	201	100	100	99
DIABLO	605219	135	100	100	100
DIAPH1	602121	101	100	99	96
DIAPH2	300108	29	86	64	38
DIAPH3	614567	70	86	79	71
DICER1	606241	95	100	99	96
DIP2B	611379	119	100	100	99
DIS3L2	614184	153	100	97	96
DKC1	300126	73	100	93	88
DLAT	608770	131	100	99	92
DLC1	604258	160	100	99	98
DLD	238331	65	99	90	76

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
DLG3	300189	120	100	100	98
DLG4	602887	214	100	100	100
DLL3	602768	150	100	100	100
DLL4	605185	256	100	100	100
DLST	126063	109	100	99	96
DLX3	600525	271	100	100	100
DLX5	600028	257	100	100	100
DMD	300377	68	98	93	87
DMGDH	605849	87	100	96	92
DMP1	600980	92	100	96	96
DMPK	605377	233	100	100	100
DMRT1	602424	169	100	100	100
DMRT2	604935	137	100	99	98
DNA2	601810	61	99	92	83
DNAAF1	613190	146	100	100	97
DNAAF2	612517	213	100	99	97
DNAAF3	614566	243	100	100	100
DNAAF4	608706	55	82	72	64
DNAAF5	614864	217	100	97	96
DNAH11	603339	89	100	96	89
DNAH5	603335	101	99	97	93
DNAI1	604366	151	100	99	97
DNAI2	605483	202	100	100	100
DNAJB2	604139	150	100	100	100
DNAJB6	611332	107	100	94	92
DNAJC12	606060	97	100	98	92
DNAJC19	608977	70	100	98	85
DNAJC5	611203	258	100	100	100
DNAJC6	608375	125	100	100	96
DNAL1	610062	80	90	82	71
DNASE1L3	602244	108	100	100	99
DNHD1	617277	228	100	100	100
DNM1	602377	172	100	100	99
DNM1L	603850	61	99	92	82
DNM2	602378	208	100	100	100
DNMT1	126375	192	100	100	98
DNMT3A	602769	198	100	100	100
DNMT3B	602900	192	100	98	94
DOCK3	603123	140	100	99	99
DOCK6	614194	255	100	100	100
DOCK7	615730	72	99	94	87
DOCK8	611432	118	100	99	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
DOK7	610285	216	98	97	96
DOLK	610746	430	100	100	100
DONSON	611428	78	95	93	89
DPAGT1	191350	151	100	100	100
DPEP1	179780	275	100	100	100
DPF2	601671	164	100	100	100
DPH1	603527	216	100	100	100
DPM1	603503	79	98	89	73
DPM2	603564	164	100	100	100
DPM3	605951	175	100	100	100
DPP6	126141	139	100	95	91
DPY19L2	613893	39	69	52	39
DPYD	612779	73	94	92	86
DPYS	613326	160	100	100	98
DRC1	615288	111	100	99	93
DRD2	126450	274	100	100	100
DRD4	126452	164	100	100	100
DRD5	126453	216	100	100	100
DSC2	125645	84	99	93	89
DSC3	600271	69	97	88	78
DSG1	125670	92	96	88	80
DSG2	125671	104	96	93	86
DSG4	607892	100	100	96	89
DSP	125647	411	100	100	99
DSPP	125485	32	54	49	45
DST	113810	91	99	96	91
DSTYK	612666	167	100	100	100
DTNA	601239	109	100	99	96
DTNBP1	607145	130	100	91	86
DUOX2	606759	183	100	100	100
DUOXA2	612772	142	100	100	100
DUSP6	602748	271	100	100	100
DVL1	601365	251	100	100	100
DVL3	601368	221	100	100	100
DYM	607461	82	100	99	93
DYNC1H1	600112	161	100	100	99
DYNC2H1	603297	57	94	84	73
DYNC2LI1	617083	71	97	83	77
DYRK1A	600855	111	100	97	92
DYSF	603009	184	100	100	100
EARS2	612799	240	100	100	100
EBP	300205	110	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ECE1	600423	185	99	97	97
ECEL1	605896	240	100	100	100
ECHS1	602292	170	100	100	100
ECM1	602201	176	100	100	100
EDA	300451	111	97	97	95
EDAR	604095	260	100	100	100
EDARADD	606603	122	99	90	83
EDC3	609842	226	100	100	100
EDN1	131240	85	100	97	85
EDN3	131242	187	100	100	100
EDNRA	131243	102	100	99	95
EDNRB	131244	135	100	99	94
EDRF1	No ID	66	99	93	82
EEF1A2	602959	218	100	100	100
EFCAB1	No ID	57	89	85	78
EFEMP1	601548	125	100	100	98
EFEMP2	604633	217	100	100	100
EFNA4	601380	253	100	100	100
EFNB1	300035	143	100	100	100
EFTUD2	603892	124	100	100	100
EGF	131530	110	99	95	91
EGFR	131550	156	100	100	99
EGLN1	606425	181	100	100	98
EGR2	129010	402	100	100	100
EHMT1	607001	176	99	99	98
EIF2AK3	604032	95	97	93	90
EIF2AK4	609280	110	98	95	91
EIF2B1	606686	126	100	100	99
EIF2B2	606454	152	100	100	99
EIF2B3	606273	121	100	100	99
EIF2B4	606687	177	100	98	93
EIF2B5	603945	146	100	100	100
EIF2S3	300161	54	99	90	78
EIF4A3	608546	94	100	98	94
EIF4G1	600495	178	100	100	99
ELAC2	605367	162	100	100	100
ELANE	130130	211	100	100	100
ELF4	300775	128	100	100	99
ELN	130160	171	100	100	100
ELOVL4	605512	98	98	89	81
ELP1	603722	No coverage data			
ELP2	616054	86	98	91	83

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
EMC1	616846	147	100	100	99
EMD	300384	184	100	98	95
EMG1	611531	153	100	100	100
EML1	602033	103	100	100	98
EMX2	600035	176	100	99	95
ENAM	606585	120	99	98	96
ENG	131195	281	100	100	100
ENO3	131370	236	100	100	100
ENPP1	173335	67	93	85	78
ENTPD1	601752	102	100	100	99
EOGT	614789	69	100	95	90
EOMES	604615	171	100	100	100
EP300	602700	158	99	98	94
EPAS1	603349	200	100	100	100
EPB41	130500	71	100	96	92
EPB41L1	602879	174	100	100	100
EPB42	177070	172	100	100	100
EPCAM	185535	69	100	94	83
EPG5	615068	106	100	99	98
EPHA2	176946	284	100	100	100
EPHB2	600997	279	98	98	98
EPHX1	132810	136	100	100	98
EPHX2	132811	130	100	100	99
EPM2A	607566	164	90	86	84
EPX	131399	209	100	100	100
ERBB2	164870	209	100	98	98
ERBB3	190151	150	100	100	100
ERBB4	600543	112	100	100	97
ERCC1	126380	143	100	100	100
ERCC2	126340	262	100	100	100
ERCC3	133510	178	100	100	100
ERCC4	133520	101	100	98	92
ERCC5	133530	87	99	96	89
ERCC6	609413	128	100	100	98
ERCC6L2	615667	66	99	93	86
ERCC8	609412	81	100	94	84
ERF	611888	220	100	100	100
ERLIN2	611605	110	100	100	100
ERMARD	615532	102	96	82	73
ESCO2	609353	88	97	89	81
ESPN	606351	187	99	99	98
ESR1	133430	181	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ESRRB	602167	283	100	100	100
ETFA	608053	77	100	92	83
ETFB	130410	179	100	100	100
ETFDH	231675	73	100	92	81
ETHE1	608451	197	100	100	100
ETV6	600618	186	100	100	100
EVC	604831	189	94	92	91
EVC2	607261	181	100	100	99
EWSR1	133450	153	98	97	95
EXOSC2	602238	107	100	100	100
EXOSC3	606489	208	100	100	100
EXPH5	612878	130	99	97	96
EXT1	608177	298	100	100	100
EXT2	608210	142	100	100	100
EXTL3	605744	223	100	100	100
EYA1	601653	111	95	95	89
EYA4	603550	93	100	95	90
EYS	612424	89	100	96	91
EZH2	601573	77	98	94	91
F10	613872	284	97	97	97
F11	264900	151	98	95	89
F12	610619	312	100	100	100
F13A1	134570	148	100	100	100
F13B	134580	64	93	68	48
F2	176930	208	100	100	100
F5	612309	139	98	98	96
F7	613878	328	100	100	100
F8	300841	148	100	99	96
F9	300746	60	97	89	78
FA2H	611026	182	100	100	100
FADD	602457	277	100	100	100
FAH	613871	186	100	100	100
FAM111A	615292	108	100	99	97
FAM111B	615584	98	100	98	94
FAM126A	610531	96	100	100	94
FAM161A	613596	73	98	90	84
FAM20A	611062	139	100	100	100
FAM20C	611061	200	100	100	100
FAM83H	611927	275	100	100	100
FAN1	613534	134	100	100	99
FANCA	607139	163	100	98	96
FANCB	300515	38	94	88	69

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
FANCC	613899	115	100	97	94
FANCD2	613984	92	100	96	92
FANCE	613976	166	100	100	100
FANCF	613897	432	100	100	100
FANCG	602956	171	100	100	100
FANCI	611360	83	100	98	93
FANCL	608111	59	100	92	74
FANCM	609644	66	95	88	78
FAR1	616107	69	100	92	82
FARS2	611592	221	100	100	100
FAS	134637	114	91	83	74
FASLG	134638	106	100	100	99
FASTKD2	612322	72	100	98	91
FAT1	600976	164	100	100	100
FAT2	604269	229	100	100	100
FAT4	612411	170	100	99	98
FBLN1	135820	243	100	100	99
FBLN5	604580	147	100	100	100
FBN1	134797	141	100	100	99
FBN2	612570	139	100	100	99
FBP1	611570	229	100	100	100
FBXL4	605654	94	100	99	95
FBXO11	607871	63	100	96	87
FBXO31	609102	210	100	100	100
FBXO38	608533	111	100	94	84
FBXO7	605648	126	97	94	90
FCGR3A	146740	215	100	100	100
FCGR3B	610665	160	100	100	100
FCN3	604973	191	100	100	100
FECH	612386	139	100	100	99
FERMT1	607900	114	95	88	83
FERMT3	607901	243	100	100	100
FEZF1	613301	262	100	100	100
FGA	134820	155	98	93	93
FGB	134830	76	100	100	96
FGD1	300546	126	100	100	100
FGD4	611104	78	98	96	88
FGF10	602115	130	100	100	100
FGF12	601513	145	100	100	100
FGF14	601515	93	100	97	89
FGF16	300827	74	100	100	95
FGF17	603725	222	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
FGF23	605380	272	100	100	100
FGF3	164950	219	100	100	100
FGF8	600483	142	100	97	94
FGF9	600921	122	100	100	95
FGFR1	136350	185	100	100	100
FGFR2	176943	145	100	100	99
FGFR3	134934	241	100	100	100
FGG	134850	90	100	92	85
FGL1	605776	64	90	89	83
FH	136850	83	100	100	98
FHL1	300163	133	100	100	99
FIBP	608296	172	100	100	100
FIG4	609390	88	100	96	91
FIGLA	608697	143	100	96	95
FIGN	605295	185	100	100	100
FKBP10	607063	195	100	100	100
FKBP14	614505	70	100	100	97
FKRP	606596	339	100	100	100
FKTN	607440	104	90	81	68
FLAD1	610595	269	100	100	100
FLCN	607273	218	100	100	100
FLG	135940	376	100	98	97
FLNA	300017	206	100	100	100
FLNB	603381	193	100	100	99
FLNC	102565	290	100	100	100
FLRT3	604808	140	100	100	100
FLT3	136351	88	100	95	86
FLT4	136352	284	100	100	100
FLVCR1	609144	132	100	95	85
FLVCR2	610865	192	100	100	100
FMN2	606373	113	99	94	86
FMO3	136132	157	100	100	99
FMR1	309550	39	96	84	63
FN1	135600	150	100	100	100
FOLR1	136430	146	100	100	100
FOXC1	601090	137	100	96	93
FOXC2	602402	242	100	100	100
FOXE1	602617	146	100	100	99
FOXE3	601094	100	93	83	82
FOXF1	601089	280	100	100	100
FOXG1	164874	424	100	97	94
FOXH1	603621	355	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
FOXI1	601093	172	100	100	98
FOXL2	605597	274	100	100	100
FOXN1	600838	201	100	100	100
FOXO1	136533	193	100	100	99
FOXP1	605515	121	100	100	100
FOXP2	605317	83	100	98	94
FOXP3	300292	157	100	100	100
FOXRED1	613622	260	100	100	100
FRAS1	607830	141	100	99	99
FREM1	608944	123	100	99	96
FREM2	608945	189	100	100	99
FRMD4A	616305	174	100	100	100
FRMD7	300628	80	100	99	93
FRMPD4	300838	105	100	100	96
FRRS1L	604574	75	100	92	80
FSCN2	607643	333	100	100	100
FSHB	136530	111	100	100	98
FSHR	136435	148	100	100	100
FTCD	606806	270	100	99	98
FTL	134790	188	100	100	100
FTO	610966	149	100	100	100
FTSJ1	300499	155	100	100	100
FUCA1	612280	172	100	100	100
FUS	137070	135	100	100	100
FUT2	182100	243	100	100	100
FUT6	136836	339	100	100	100
FUZ	610622	183	100	100	100
FXN	606829	96	100	100	99
FXYD2	601814	241	100	100	100
FYCO1	607182	166	100	100	100
FZD2	600667	394	100	100	99
FZD4	604579	175	100	100	100
FZD6	603409	110	100	99	96
G6PC	613742	178	100	100	100
G6PC3	611045	161	100	99	95
G6PD	305900	211	100	100	100
GAA	606800	259	100	100	100
GABRA1	137160	96	100	99	97
GABRB1	137190	110	100	100	96
GABRB3	137192	160	100	100	100
GABRG2	137164	98	100	99	93
GAD1	605363	139	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
GALC	606890	85	98	94	88
GALE	606953	200	100	100	100
GALK1	604313	319	100	100	100
GALNS	612222	220	100	100	100
GALNT3	601756	74	99	89	82
GALT	606999	231	100	100	100
GAMT	601240	284	100	100	100
GAN	605379	123	100	99	96
GANAB	104160	156	100	100	100
GARS1	600287	67	100	96	89
GART	142810	104	100	100	99
GAS6	600441	202	99	97	96
GATA1	305371	112	100	100	99
GATA2	137295	235	100	100	100
GATA3	131320	199	100	100	100
GATA4	600576	164	100	100	100
GATA5	611496	213	100	100	100
GATA6	601656	183	100	96	92
GATAD1	614518	162	100	100	99
GATAD2B	614998	153	100	100	100
GATM	602360	105	100	100	99
GBA	606463	207	100	100	100
GBA2	609471	212	100	100	100
GBE1	607839	87	100	100	98
GCDH	608801	239	100	100	100
GCH1	600225	207	94	94	94
GCK	138079	256	100	100	100
GCLC	606857	103	100	96	91
GCM2	603716	215	100	100	100
GCNT2	600429	149	100	100	100
GCSH	238330	43	100	85	69
GDAP1	606598	112	100	95	87
GDF1	602880	150	100	100	99
GDF2	605120	204	100	100	100
GDF3	606522	173	100	100	100
GDF5	601146	246	100	100	100
GDF6	601147	147	100	100	100
GDI1	300104	130	100	100	99
GDNF	600837	114	100	100	100
GFAP	137780	176	100	100	100
GFER	600924	168	100	100	100
GFI1	600871	221	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
GFI1B	604383	283	100	100	100
GFM1	606639	82	100	99	94
GFM2	606544	73	100	94	88
GFPT1	138292	90	100	100	97
GGCX	137167	113	100	100	97
GGT1	612346	97	100	91	80
GH1	139250	200	100	100	100
GHR	600946	105	100	95	88
GHRHR	139191	163	100	100	100
GHSR	601898	463	100	100	99
GIGYF2	612003	80	100	98	91
GIPC3	608792	232	100	100	100
GJA1	121014	143	100	100	100
GJA3	121015	267	100	100	100
GJA5	121013	286	100	100	100
GJA8	600897	207	100	100	100
GJB1	304040	137	100	100	99
GJB2	121011	191	100	100	100
GJB3	603324	289	100	100	100
GJB4	605425	454	100	100	100
GJB6	604418	154	100	100	100
GJC2	608803	283	100	100	98
GK	300474	27	83	59	35
GLA	300644	87	100	100	98
GLB1	611458	149	100	100	100
GLDC	238300	118	100	100	98
GLE1	603371	140	100	100	97
GLI2	165230	260	100	100	100
GLI3	165240	243	100	100	100
GLIS2	608539	245	100	100	100
GLIS3	610192	199	100	100	100
GLMN	601749	39	90	73	58
GLRA1	138491	154	100	100	100
GLRB	138492	74	95	91	79
GLRX5	609588	125	100	100	100
GLUD1	138130	123	100	98	93
GLUL	138290	140	100	100	100
GLYCK	610516	298	100	100	100
GM2A	613109	183	100	100	100
GMNN	602842	45	91	69	59
GMPPA	615495	180	100	100	100
GMPPB	615320	281	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
GMPS	600358	83	100	97	89
GNA11	139313	233	100	100	100
GNAI2	139360	176	100	100	100
GNAI3	139370	70	98	87	75
GNAL	139312	114	100	100	99
GNAO1	139311	173	100	100	99
GNAQ	600998	137	100	100	100
GNAS	139320	269	100	100	100
GNAT1	139330	336	100	100	100
GNAT2	139340	125	100	100	100
GNB1	139380	172	100	100	100
GNB4	610863	67	93	91	84
GNB5	604447	134	100	100	100
GNE	603824	123	100	100	97
GNMT	606628	215	100	100	100
GNPAT	602744	80	97	94	86
GNPTAB	607840	87	100	99	95
GNPTG	607838	302	100	100	100
GNRH1	152760	107	100	100	100
GNRHR	138850	96	100	100	96
GNS	607664	105	100	100	99
GOLGA5	606918	63	100	93	80
GORAB	607983	77	100	93	83
GOSR2	604027	139	100	100	100
GOT1	138180	159	100	100	100
GP1BA	606672	202	100	100	100
GP1BB	138720	113	100	98	94
GP6	605546	291	100	100	100
GP9	173515	330	100	100	100
GPC3	300037	86	100	98	92
GPC4	300168	93	100	99	97
GPC6	604404	135	100	100	100
GPD1	138420	219	100	100	100
GPD1L	611778	123	100	100	100
GPHN	603930	87	100	96	94
GPI	172400	217	100	100	100
GPR143	300808	111	100	100	99
GPR179	614515	401	100	100	100
GPSM2	609245	54	100	96	84
GPT2	138210	169	100	100	96
GPX1	138320	310	100	100	100
GPX4	138322	285	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
GRHL2	608576	120	100	100	98
GRHL3	608317	212	100	100	100
GRHPR	604296	185	100	100	100
GRIA3	305915	62	98	91	79
GRID2	602368	117	100	100	98
GRIK2	138244	66	99	93	86
GRIK5	600283	239	100	100	100
GRIN1	138249	259	100	100	100
GRIN2A	138253	408	100	100	99
GRIN2B	138252	237	100	100	99
GRIN3B	606651	217	97	95	93
GRIP1	604597	117	100	100	98
GRK1	180381	270	100	100	100
GRM1	604473	210	100	100	99
GRM6	604096	285	100	98	96
GRN	138945	336	100	100	100
GRXCR1	613283	100	100	100	100
GSC	138890	158	100	99	95
GSDME	608798	125	100	99	97
GSE1	616886	210	100	100	100
GSN	137350	201	100	100	100
GSS	601002	188	100	100	100
GSTZ1	603758	176	100	100	100
GTF2H5	608780	81	100	100	100
GTPBP3	608536	237	100	100	100
GUCA1A	600364	216	100	100	100
GUCA1B	602275	122	100	100	99
GUCY1A1	139396	116	100	98	94
GUCY2C	601330	91	100	97	92
GUCY2D	600179	218	100	100	100
GUCY2F	300041	69	100	99	93
GUSB	611499	222	100	100	100
GYG1	603942	107	100	100	98
GYS1	138570	231	100	100	100
GYS2	138571	96	99	94	89
H19	103280	No coverage data			
H6PD	138090	279	100	100	100
HACE1	610876	59	97	89	77
HADH	601609	145	100	100	98
HADHA	600890	125	100	99	95
HADHB	143450	74	90	84	76
HAL	609457	182	100	99	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
HAMP	606464	154	100	100	100
HAND1	602406	246	100	100	100
HAND2	602407	217	100	100	100
HARS1	142810	163	100	100	100
HARS2	600783	138	100	100	100
HAX1	605998	120	100	100	100
HBA1	141800	155	100	100	100
HBA2	141850	128	84	76	76
HBB	141900	205	100	100	100
HBD	142000	219	100	100	100
HBG1	142200	40	76	45	30
HBG2	142250	232	100	100	95
HCCS	300056	64	100	98	90
HCFC1	300019	207	100	100	100
HCN1	602780	193	100	99	97
HCN4	605206	306	100	100	100
HCRT	602358	117	100	100	100
HDAC4	605314	227	100	99	97
HDAC6	300272	145	100	100	100
HDAC8	300269	73	100	100	95
HECTD1	No ID	83	96	93	88
HECW2	617245	142	100	100	99
HEPACAM	611642	195	100	100	100
HERC1	605109	116	100	99	97
HERC2	605837	144	99	94	90
HES7	608059	144	100	100	98
HESX1	601802	47	79	62	57
HEXA	606869	162	100	100	100
HEXB	606873	73	97	91	84
HEY2	604674	246	100	98	94
HFE	613609	175	100	100	100
HFM1	615684	34	79	58	43
HGD	607474	123	100	100	98
HGSNAT	610453	93	99	94	92
HIBCH	610690	68	95	91	80
HINT1	601314	123	100	99	92
HIVEP2	143054	196	100	100	99
HJV	608374	248	100	100	100
HK1	142600	158	100	100	99
HLCS	609018	163	100	100	99
HMBS	609806	202	100	100	100
HMGCL	613898	168	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
HMGCS2	600234	176	100	100	99
HMOX1	141250	187	100	100	100
HMX1	142992	94	100	100	100
HNF1A	142410	285	100	100	100
HNF1B	189907	231	100	100	100
HNF4A	600281	221	100	100	100
HNMT	605238	84	100	100	91
HNRNPA1	164017	114	100	99	94
HNRNPH2	300610	110	100	100	100
HNRNPK	600712	69	97	89	77
HNRNPU	602869	88	95	94	92
HOGA1	613597	229	100	100	100
HOXA1	142955	294	100	100	100
HOXA11	142958	217	100	100	100
HOXA13	142959	221	91	85	82
HOXB1	142968	255	100	100	100
HOXC13	142976	165	100	100	100
HOXD10	142984	170	100	100	100
HOXD13	142989	278	100	100	98
HPD	609695	196	100	100	100
HPGD	601688	79	96	77	60
HPRT1	308000	40	95	80	55
HPS1	604982	250	100	100	100
HPS3	606118	92	97	91	85
HPS4	606682	365	100	99	98
HPS5	607521	85	100	97	91
HPS6	607522	246	100	100	100
HPSE2	613469	122	100	100	100
HR	602302	254	100	100	100
HRAS	190020	426	100	100	100
HRG	142640	178	100	99	97
HS6ST1	604846	330	100	100	100
HSD11B1	600713	111	100	100	97
HSD11B2	614232	232	95	92	86
HSD17B10	300256	123	100	100	100
HSD17B3	605573	117	100	98	89
HSD17B4	601860	78	93	91	84
HSD3B2	613890	170	100	100	100
HSD3B7	607764	275	100	100	100
HSF4	602438	224	100	100	100
HSPA9	600548	114	100	99	97
HSPB1	602195	256	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
HSPB3	604624	150	100	100	100
HSPB8	608014	133	100	100	100
HSPD1	118190	67	99	90	77
HSPG2	142461	283	99	99	99
HTR1A	109760	337	100	100	100
HTRA1	602194	144	99	91	89
HTRA2	606441	256	100	100	100
HUWE1	300697	90	100	98	93
HYAL1	607071	247	100	100	100
HYDIN	610812	86	88	81	76
HYLS1	610693	114	100	100	100
IARS1	600709	94	100	97	94
IBA57	615316	227	100	100	100
CILK1	612325	119	99	93	90
ICOS	604558	70	100	95	87
IDH2	147650	236	100	100	100
IDH3B	604526	244	100	100	100
IDS	300823	91	100	98	94
IDUA	252800	224	100	100	98
IER3IP1	609382	147	100	95	85
IFIH1	606951	70	99	91	80
IFITM5	614757	322	100	100	100
IFNGR1	107470	103	92	91	91
IFT122	606045	141	100	100	98
IFT140	614620	199	100	100	100
IFT172	607386	143	100	100	98
IFT43	614068	133	100	100	100
IFT80	611177	49	92	82	69
IFT81	605489	37	96	78	54
IGBP1	300139	86	100	98	89
IGF1	147440	117	100	100	100
IGF1R	147370	176	100	100	100
IGF2R	147280	130	99	96	93
IGFALS	601489	372	100	100	100
IGFBP7	602867	126	100	100	100
IGHMBP2	600502	265	100	100	99
IGLL1	146770	157	100	100	100
IGSF1	300137	91	100	100	98
IHH	600726	276	100	100	100
IKBKB	603258	159	100	100	100
IKBKG	300248	35	36	26	26
IKZF1	603023	257	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
IKZF5	606238	163	100	100	99
IL10RA	146933	181	100	100	100
IL10RB	123889	86	100	98	96
IL11	147681	155	100	99	95
IL11RA	600939	193	100	100	100
IL17F	606496	142	100	100	100
IL17RA	605461	346	100	100	100
IL17RD	606807	160	100	100	99
IL1RAPL1	300206	56	100	92	78
IL1RN	147679	138	100	100	100
IL21R	605383	271	100	100	100
IL2RA	147730	139	100	100	100
IL2RG	308380	88	100	100	97
IL31RA	609510	103	100	99	96
IL36RN	605507	192	100	100	100
IL7R	146661	105	100	100	98
ILDRL1	609739	193	100	100	98
IMPA1	602064	63	94	85	79
BPNT2	614010	192	100	100	100
IMPDH1	146690	255	100	100	100
IMPG2	607056	106	99	97	94
INF2	610982	277	98	97	96
ING1	601566	330	100	100	100
INPP5B	147264	132	100	100	99
INPP5E	613037	259	100	100	100
INPP5K	607875	174	100	100	100
INPPL1	600829	195	100	100	99
INS	176730	321	100	100	100
INSL3	146738	195	100	100	100
INSR	147670	243	100	100	99
INTS1	611345	223	100	99	99
INTS8	611351	53	92	77	67
INVS	243305	123	100	99	97
IQCB1	609237	75	100	99	88
IQCE	617631	203	100	98	96
IQSEC2	300522	128	100	99	98
IRAK4	606883	51	96	85	74
IRF1	147575	204	100	100	100
IRF4	601900	204	100	100	100
IRF6	607199	184	100	100	100
IRF8	601565	157	100	100	97
IRGM	608212	158	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
IRX4	606199	206	100	100	100
IRX5	606195	211	100	100	100
ISCU	611911	91	100	100	100
ISG15	147571	418	100	100	100
ITCH	606409	61	96	90	80
ITGA2	192974	76	99	94	89
ITGA2B	607759	255	100	100	100
ITGA3	605025	224	100	100	100
ITGA6	147556	109	100	98	93
ITGA7	600536	219	100	100	100
ITGA8	604063	102	100	99	97
ITGB2	600065	241	100	100	100
ITGB3	173470	178	100	100	100
ITGB4	147557	269	100	99	99
ITK	186973	104	100	100	96
ITM2B	603904	87	100	96	88
ITPA	147520	208	100	98	93
ITPR1	147265	120	100	99	97
ITPR2	600144	83	95	93	88
ITSN1	602442	109	98	95	91
IVD	607036	170	100	100	100
IYD	612025	92	100	100	96
JAG1	601920	213	100	100	100
JAK2	147796	55	98	91	79
JAK3	600173	266	100	100	100
JAM3	606871	129	100	100	100
JMJD1C	604503	93	95	90	84
JPH2	605267	287	100	100	100
JPH3	605268	362	100	100	100
JUP	173325	266	100	100	100
KALRN	604605	153	100	99	99
KANK1	607704	177	100	100	100
KANSL1	612452	188	100	100	100
KARS1	601421	123	100	100	99
KAT6A	601408	140	100	100	99
KAT6B	605880	131	99	95	92
KATNB1	602703	334	100	100	100
KBTBD13	613727	438	100	100	100
KCNA1	176260	224	100	100	100
KCNA2	176262	209	100	100	100
KCNA4	176266	165	100	100	100
KCNA5	176267	336	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
KCNB1	600397	279	100	100	100
KCNC3	176264	321	94	88	87
KCND3	605411	350	100	100	99
KCNE1	176261	657	100	100	100
KCNE2	603796	301	100	100	100
KCNE3	604433	190	100	100	100
KCNH1	603305	163	100	100	99
KCNH2	152427	275	100	100	100
KCNJ1	600359	156	100	100	100
KCNJ10	602208	271	100	100	100
KCNJ11	600937	382	100	100	100
KCNJ13	603208	143	100	100	100
KCNJ2	600681	189	100	100	100
KCNJ5	600734	275	100	100	100
KCNJ6	600877	177	100	100	100
KCNJ8	600935	559	100	100	100
KCNK3	603220	231	100	100	98
KCNK9	605874	267	100	100	100
KCNMA1	600150	122	100	98	96
KCNQ1	607542	235	100	97	94
KCNQ1OT1	604115	No coverage data			
KCNQ2	602235	274	100	100	100
KCNQ3	602232	186	100	100	98
KCNQ4	603537	245	98	97	96
KCNQ5	607357	107	100	99	93
KCNT1	608167	236	100	100	100
KCNV2	607604	268	100	100	100
KCTD1	613420	249	100	98	95
KCTD7	611725	259	100	100	100
KDM1A	609132	90	98	95	90
KDM4B	609765	285	100	100	100
KDM5B	605393	106	100	99	96
KDM5C	314690	130	100	100	99
KDM6A	300128	57	95	86	73
KDM6B	611577	250	100	100	99
KDR	191306	105	100	98	95
KDSR	136440	104	100	100	99
KERA	603288	118	100	100	100
KHDC3L	611687	178	100	100	100
KHK	614058	255	100	100	100
KIAA0586	610178	62	96	86	75
KIAA0753	617112	101	100	97	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
KIAA1109	611565	71	99	95	88
KIAA1586	No ID	35	95	80	57
KIDINS220	615759	106	100	98	93
KIF11	148760	46	95	80	64
KIF1A	601255	260	100	100	100
KIF1B	605995	104	100	100	98
KIFBP	609367	117	100	100	100
KIF1C	603060	225	100	100	100
KIF20A	605664	143	100	100	100
KIF21A	608283	69	97	91	83
KIF22	603213	243	100	100	100
KIF2A	602591	58	95	79	65
KIF4A	300521	54	100	91	73
KIF5A	602821	126	100	100	100
KIF5C	604593	99	99	93	87
KIF7	611254	224	100	100	98
KIRREL3	607761	297	100	100	100
KISS1	603286	162	100	100	100
KISS1R	604161	180	100	100	100
KIT	164920	103	100	99	97
KITLG	184745	83	100	100	92
KL	604824	172	98	96	95
KLF1	600599	212	100	100	100
KLF11	603301	176	100	100	100
KLF6	602053	176	100	100	100
KLHDC8B	613169	255	100	100	100
KLHL10	608778	151	100	100	99
KLHL15	300980	92	100	100	100
KLHL3	605775	146	100	100	100
KLHL40	615340	236	100	100	100
KLHL41	607701	107	100	99	98
KLHL7	611119	89	100	97	88
KLK4	603767	311	100	100	100
KLKB1	229000	81	96	88	84
KLLN	612105	367	100	100	100
KMT2A	159555	118	100	100	98
KMT2B	606834	247	99	98	97
KMT2C	606833	126	98	96	92
KMT2D	602113	341	100	100	100
KMT2E	608444	101	98	91	86
KMT5B	610881	86	99	95	89
KNL1	609173	80	99	96	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
KPTN	615620	242	100	100	100
KRAS	190070	59	90	78	77
KRBOX4	300585	149	100	100	97
KRIT1	604214	74	100	96	87
KRT1	139350	155	100	100	98
KRT10	148080	99	100	98	96
KRT12	601687	143	100	100	100
KRT13	148065	170	100	100	100
KRT14	148066	208	100	100	100
KRT16	148067	180	100	100	99
KRT17	148069	168	100	100	99
KRT18	148070	172	100	100	100
KRT2	600194	154	100	100	100
KRT3	148043	154	100	100	99
KRT4	123940	199	100	100	100
KRT5	148040	282	100	100	100
KRT6A	148041	309	100	100	100
KRT6B	148042	319	100	100	100
KRT6C	612315	263	100	100	100
KRT74	608248	147	100	100	100
KRT8	148060	118	100	100	100
KRT81	602153	170	82	80	76
KRT83	602765	196	100	100	100
KRT85	602767	241	100	100	100
KRT86	601928	177	81	79	74
KRT9	607606	192	100	100	98
KYNU	605197	65	99	82	71
L1CAM	308840	164	100	100	100
L2HGDH	609584	73	97	96	93
LAMA1	150320	140	100	99	98
LAMA2	156225	96	100	98	93
LAMA3	600805	121	100	99	96
LAMA4	600133	117	100	98	94
LAMB1	150240	144	100	98	94
LAMB2	150325	282	100	100	100
LAMB3	150310	222	100	100	100
LAMC1	150290	152	100	98	95
LAMC2	150292	140	100	100	98
LAMC3	604349	249	100	100	100
LAMP2	309060	54	100	89	76
LAMTOR2	610389	239	100	100	100
LARGE1	603590	204	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
LARP7	612026	37	95	79	62
LARS2	604544	126	100	100	99
LAS1L	300964	128	100	100	99
LBR	600024	72	96	94	86
LCA5	611408	64	95	94	88
LCAT	606967	392	100	100	100
LCT	603202	230	100	100	100
LDB3	605906	213	100	100	100
LDHA	150000	81	100	98	92
LDHB	150100	107	100	98	89
LDLR	606945	268	100	100	100
LDLRAP1	605747	227	100	100	100
LEF1	153245	127	100	100	100
LEFTY2	601877	342	100	100	100
LEMD3	607844	108	96	91	83
LEP	164160	135	100	100	100
LEPR	601007	80	93	86	79
LFNG	602576	221	88	86	85
LGI1	604619	84	98	93	84
LHB	152780	161	100	100	100
LHCGR	152790	93	99	94	87
LHFPL5	609427	263	100	100	100
LHX3	600577	232	100	100	100
LHX4	602146	206	100	100	100
LIAS	607031	78	95	93	87
LIFR	151443	66	87	80	72
LIG1	126391	172	100	100	100
LIG4	601837	86	100	97	93
LIM2	154045	298	100	100	100
LINS1	610350	80	100	96	89
LIPA	613497	85	100	100	99
LIPC	151670	151	100	92	91
LIPH	607365	121	100	100	98
LIPI	609252	49	95	87	76
LIPN	613924	80	98	88	87
LITAF	603795	173	100	100	100
LMAN1	601567	104	95	90	83
LMAN2L	609552	141	100	100	100
LMBR1	605522	45	99	84	65
LMBRD1	612625	51	89	71	61
LMF1	611761	228	100	100	100
LMNA	150330	238	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
LMNB1	150340	110	100	97	92
LMOD1	602715	134	100	100	100
LMOD3	616112	120	90	89	88
LMX1B	602575	257	100	100	100
LONP1	605490	264	100	100	100
LORICRIN	152445	67	100	100	100
LOX	153455	226	100	100	100
LOXHD1	613072	197	100	100	100
LPAR6	609239	55	100	99	83
LPIN1	605518	118	100	99	95
LPIN2	605519	125	100	100	99
LPL	609708	124	100	100	99
LPP	600700	183	100	100	100
LRAT	604863	260	100	100	100
LRBA	606453	90	99	96	91
LRIG2	608869	107	98	95	91
LRIT3	615004	173	100	100	100
LRMDA	614537	148	100	100	100
LRP2	600073	118	100	99	97
LRP4	604270	228	100	100	99
LRP5	603506	325	98	98	98
LRPAP1	104225	173	100	100	99
LRPPRC	607544	61	98	90	79
LRRC6	614930	73	93	91	86
LRRC8A	608360	319	100	100	100
LRRK1	610986	225	99	98	97
LRRK2	609007	65	98	92	84
LRSAM1	610933	180	100	100	100
LRTOMT	612414	174	100	100	100
LTBP2	602091	241	100	100	100
LTBP3	602090	223	100	99	99
LTBP4	604710	271	100	100	99
LTC4S	246530	118	100	100	98
LYST	606897	87	97	92	89
LYZ	153450	91	100	100	99
LZTFL1	606568	43	84	76	70
LZTR1	600574	242	100	100	100
LZTS1	606551	223	100	100	100
MAB21L2	604357	511	100	100	100
MACF1	608271	104	100	99	97
MACROD2	611567	58	96	90	83
MAD1L1	602686	164	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
MAF	177075	206	89	85	82
MAFB	608968	275	100	100	100
MAGEC3	300469	106	100	100	99
MAGEL2	605283	355	100	100	100
MAGT1	300715	69	93	87	78
MAK	154235	81	94	92	88
MAML2	607537	171	100	100	100
MAMLD1	300120	125	100	100	100
MAN1B1	604346	251	100	100	100
MAN2B1	609458	213	100	100	100
MANBA	609489	93	100	99	94
MAOA	309850	74	100	99	92
MAP1A	600178	213	100	100	99
MAP2K1	176872	148	100	100	99
MAP2K2	601263	276	100	100	100
MAP3K1	600982	97	99	98	95
MAP3K7	602614	83	99	96	90
MAP3K8	191195	108	100	100	97
MAPRE2	605789	98	100	100	98
MAPT	157140	170	100	100	99
MARS2	609728	343	100	100	100
MARVELD2	610572	142	90	85	84
MASP1	600521	195	100	100	100
MASP2	605102	160	100	100	100
MAST1	612256	274	100	100	100
MASTL	608221	85	100	100	97
MAT1A	610550	273	100	100	100
MAT2A	601468	102	100	100	96
MATN3	602109	144	100	95	91
MATR3	164015	86	98	92	85
MBD5	611472	141	100	99	97
MBOAT7	606048	228	100	100	100
MBTPS2	300294	52	99	94	81
MC2R	607397	210	100	100	100
MC4R	155541	124	100	100	100
MCC	159350	147	100	100	100
MCCC1	609010	111	100	96	95
MCCC2	609014	99	100	98	95
MCEE	608419	115	100	100	99
MCF2L	609499	230	100	100	100
MCFD2	607788	107	100	97	92
MCM3AP	603294	161	100	99	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
MCM4	602638	154	97	96	92
MCM6	601806	97	100	99	98
MCM8	608187	54	97	86	72
MCM9	610098	121	100	100	99
MCOLN1	605248	216	100	100	100
MCOLN3	607400	65	100	97	89
MCPH1	607117	114	87	84	81
MDGA1	609626	223	100	100	100
MDH2	154100	167	100	100	100
MECP2	300005	200	100	100	99
MECR	608205	137	100	100	99
MED12	300188	99	100	100	98
MED13	603808	93	100	97	91
MED13L	608771	124	100	100	100
MED17	603810	82	97	87	68
MED23	605042	79	100	97	92
MED25	610197	200	100	100	100
MEF2C	600662	157	100	100	99
MEFV	608107	158	100	100	100
MEGF10	612453	173	100	100	98
MEGF8	604267	242	100	100	100
MEIS2	601740	161	100	100	100
MEN1	613733	267	100	100	100
MEOX1	600147	212	100	100	100
MERTK	604705	123	100	100	96
MESP2	605195	184	100	100	100
MET	164860	111	100	99	97
METTL23	615262	109	100	100	100
MFAP5	601103	96	100	96	85
MFN2	608507	182	100	100	100
MFRP	606227	212	100	100	100
MFSD2A	614397	161	100	100	100
MFSD8	611124	82	99	93	83
MGAT2	602616	328	100	100	100
MGME1	615076	103	100	100	97
MGP	154870	68	89	89	89
MIB1	608677	96	100	98	94
MICU1	605084	88	98	92	84
MID1	300552	112	100	100	97
MID2	300204	81	99	94	87
MINPP1	605391	210	97	83	76
MIP	154050	203	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
MIR17HG	609415	No coverage data			
MIR184	613146	No coverage data			
MIR96	611606	No coverage data			
MITF	156845	119	100	100	93
MKKS	604896	251	100	100	100
MKRN3	603856	236	100	100	100
MKS1	609883	157	100	100	100
MLC1	605908	181	100	100	95
MLH1	120436	140	100	100	99
MLH3	604395	100	100	99	97
MLLT10	602409	85	97	92	90
MLLT11	604684	75	100	100	95
MLPH	606526	150	100	100	98
MLYCD	606761	134	100	100	99
MMAA	607481	82	100	99	95
MMAB	607568	156	100	100	100
MMACHC	609831	205	100	100	100
MMADHC	611935	56	85	85	80
MMP1	120353	104	100	100	99
MMP13	600108	94	100	100	98
MMP14	600754	213	100	100	100
MMP2	120360	178	100	100	100
MMP20	604629	131	100	96	92
MMP21	608416	151	100	100	99
MMP9	120361	228	100	100	100
MMUT	609058	101	100	95	92
MN1	156100	253	100	100	100
MNX1	142994	107	84	80	77
MOCOS	613274	150	100	100	97
MOCS1	603707	198	100	100	100
MOCS2	603708	81	100	100	97
MOCS3	No ID	432	100	100	100
MOG	159465	142	100	100	97
MOGS	601336	229	100	100	100
MPC1	614738	118	100	100	94
MPDU1	604041	164	100	100	100
MPDZ	603785	104	100	99	95
MPI	154550	228	100	100	100
MPL	159530	229	100	100	100
MPLKIP	609188	126	100	100	100
MPO	606989	286	100	100	100
MPV17	137960	133	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
MPZ	159440	187	100	100	100
MRAP	609196	201	100	100	100
MRAS	608435	160	100	100	100
MRE11	600814	61	97	88	76
MRPL3	607118	104	100	96	87
MRPS16	609204	119	100	100	100
MRPS22	605810	72	100	98	85
MRTFA	606078	212	100	100	100
MS4A1	112210	59	95	82	67
MSH2	609309	88	96	91	84
MSH3	600887	75	99	92	80
MSH6	600678	137	100	100	98
MSL2	614802	128	100	100	100
MSMO1	607545	50	100	93	81
MSR1	153622	78	97	93	86
MSRB3	613719	64	92	79	78
MSTN	601788	91	100	97	91
MSX1	142983	173	100	100	100
MSX2	123101	170	100	100	100
MTAP	156540	109	100	100	97
MTFMT	611766	93	100	100	97
MTHFD1	172460	118	100	99	96
MTHFR	607093	191	100	100	100
MTHFS	604197	138	100	100	100
MTM1	300415	38	95	76	55
MTMR2	603557	92	100	100	99
MTMR9	606260	94	100	99	94
MTO1	614667	115	99	93	89
MTOR	601231	147	100	100	100
MTPAP	613669	69	99	89	77
MTR	156570	119	100	100	98
MTRR	602568	95	100	100	96
MTPP	157147	89	100	100	98
MUC1	158340	181	100	100	100
MUSK	601296	116	100	99	96
MUTYH	604933	248	100	100	100
MVK	251170	207	100	100	100
MXI1	600020	84	100	93	83
MYBPC1	160794	90	100	99	95
MYBPC3	600958	272	100	100	100
MYCN	164840	275	100	100	100
MYD88	602170	228	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
MYF6	159991	154	100	100	100
MYH11	160745	171	100	100	99
MYH14	608568	204	100	100	100
MYH2	160740	134	100	99	97
MYH3	160720	137	100	100	99
MYH6	160710	225	100	100	100
MYH7	160760	221	100	100	100
MYH7B	609928	235	100	100	100
MYH8	160741	117	100	98	97
MYH9	160775	156	100	100	99
MYL2	160781	172	100	100	100
MYL3	160790	219	100	100	100
MYL9	609905	264	100	100	99
MYLK	600922	195	100	100	100
MYLK2	606566	193	100	100	100
MYMK	615345	241	100	100	100
MYO15A	602666	285	100	100	100
MYO18B	607295	248	100	100	98
MYO1A	601478	200	100	100	99
MYO1E	601479	148	100	97	96
MYO3A	606808	86	97	87	80
MYO5A	160777	106	96	93	90
MYO5B	606540	165	100	100	99
MYO6	600970	57	93	82	66
MYO7A	276903	214	100	100	100
MYOC	601652	195	100	100	100
MYOCD	606127	150	100	100	100
MYOT	604103	68	96	93	86
MYOZ2	605602	71	100	96	85
MYPN	608517	119	100	99	97
MYRF	608329	219	100	100	99
MYT1L	613084	174	100	97	91
NAA10	300013	98	100	100	100
NAA15	608000	59	99	91	78
NACC1	610672	272	100	100	100
NAGA	104170	226	100	100	100
NAGLU	609701	182	100	100	100
NAGS	608300	311	100	100	100
NALCN	611549	87	100	97	90
NANOS1	608226	102	99	97	89
NANS	605202	140	100	100	99
NARS2	612803	79	97	93	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
NAT10	609221	146	99	96	96
NAT8L	610647	265	98	91	86
NBAS	608025	103	100	98	94
NBEA	604889	68	98	93	82
NBEAL2	614169	294	100	100	100
NBN	602667	57	98	83	71
NCAPD2	615638	136	100	100	100
NCAPD3	609276	113	100	100	97
NCAPH	602332	103	95	95	94
NCAPH2	611230	256	100	100	100
NCF1	608512	62	63	58	56
NCF2	608515	213	100	100	100
NCF4	601488	170	100	100	100
NCOA4	601984	106	100	100	98
NCOR2	600848	183	100	100	99
NCSTN	605254	140	100	100	100
NDE1	609449	133	100	100	100
NDN	602117	268	100	100	100
NDP	300658	155	100	100	100
NDRG1	605262	165	100	100	100
NDST1	600853	243	100	100	100
NDUFA1	300078	106	100	100	100
NDUFA10	603835	99	97	92	87
NDUFA11	612638	243	100	100	100
NDUFA12	614530	119	100	100	100
NDUFA2	602137	189	100	100	100
NDUFA9	603834	137	100	100	97
NDUFAF1	606934	87	100	100	98
NDUFAF2	609653	77	100	99	90
NDUFAF3	612911	230	100	100	100
NDUFAF4	611776	61	90	53	38
NDUFAF5	612360	73	96	94	87
NDUFAF6	612392	66	92	81	79
NDUFB3	603839	72	100	100	100
NDUFS1	157655	90	100	99	96
NDUFS2	602985	143	100	100	100
NDUFS3	603846	180	100	100	100
NDUFS4	602694	88	100	100	98
NDUFS6	603848	99	100	100	95
NDUFS7	601825	250	100	100	100
NDUFS8	602141	334	100	100	100
NDUFV1	161015	257	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
NDUFV2	600532	68	98	90	77
NEB	161650	118	89	87	84
NECAP1	611623	150	100	100	100
NECTIN1	600644	175	100	100	100
NECTIN4	609607	227	100	100	100
NEDD4L	606384	98	98	96	93
NEFL	162280	244	100	99	95
NEK1	604588	53	92	81	70
NEK9	609798	100	100	100	97
NEU1	608272	238	100	100	100
NEUROD1	601724	236	100	100	100
NEUROG3	604882	384	100	100	100
NEXMIF	300524	94	100	100	100
NEXN	613121	30	92	66	41
NF1	613113	88	97	91	85
NF2	607379	165	100	100	100
NFATC1	600489	265	100	100	100
NFE2L2	600492	137	100	100	99
NFIA	600727	112	100	100	100
NFIX	164005	273	100	100	100
NFKB2	164012	245	100	100	100
NFKBIA	164008	168	100	100	100
NFU1	608100	56	88	75	70
NGF	162030	267	100	100	100
NGLY1	610661	70	100	97	89
NHEJ1	611290	108	100	100	96
NHLRC1	608072	312	100	100	100
NHP2	606470	210	100	100	100
NHS	300457	96	100	99	97
NID1	131390	181	100	100	100
NIN	608684	97	98	94	91
NIPA1	608145	158	100	100	99
NIPAL4	609383	127	100	99	98
NIPBL	608667	93	98	90	82
NKX2-1	600635	216	100	100	100
NKX2-5	600584	223	100	100	100
NKX2-6	611770	265	100	100	100
NKX3-2	602183	185	100	100	100
NLGN3	300336	154	100	100	99
NLGN4X	300427	201	100	100	100
NLRP12	609648	246	100	100	100
NLRP3	606416	404	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
NLRP7	609661	215	100	100	98
NME8	607421	45	87	74	62
NMNAT1	608700	118	100	100	96
NNT	607878	110	100	96	93
NOBOX	610934	222	100	100	100
NOD2	605956	268	100	100	100
NODAL	601265	262	100	100	100
NOG	602991	322	100	100	100
NOL3	605235	314	100	100	100
NONO	300084	75	100	90	83
NOP10	606471	174	100	100	100
NOP56	614154	128	100	100	100
NOS1	163731	201	100	100	100
NOTCH1	190198	318	100	99	99
NOTCH2	600275	188	100	98	97
NOTCH3	600276	291	100	100	99
NOVA2	601991	210	99	96	94
NPC1	607623	138	100	100	99
NPC2	601015	119	100	100	99
NPHP1	607100	89	97	96	89
NPHP3	608002	89	94	89	87
NPHP4	607215	258	100	100	100
NPHS1	602716	238	100	100	100
NPHS2	604766	93	100	99	94
NPM1	164040	56	94	80	63
NPPA	108780	192	100	100	100
NPPC	600296	156	100	100	100
NPR2	108961	199	100	100	100
NPRL2	607072	317	100	100	100
NPRL3	600928	184	100	100	100
NR0B1	300473	193	100	100	100
NR0B2	604630	199	100	100	100
NR2E3	604485	217	100	100	100
NR2F1	132890	208	100	99	96
NR2F2	107773	270	100	100	98
NR3C1	138040	127	100	100	100
NR3C2	600983	146	100	100	98
NR4A3	600542	136	100	100	98
NR5A1	184757	293	100	100	100
NR5A2	604453	127	100	100	97
NRAS	164790	107	100	100	99
NRL	162080	186	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
NRXN1	600565	190	100	99	98
NSD1	606681	208	100	99	98
NSD3	607083	90	100	100	98
NSDHL	300275	88	100	100	95
NSMF	608137	215	100	100	100
NSUN2	610916	113	100	99	94
NT5C	191720	149	100	100	100
NT5C2	600417	78	95	90	84
NT5C3A	606224	76	96	79	71
NT5E	129190	164	100	100	99
NTF4	162662	315	100	100	100
NTRK1	191315	272	100	100	100
NTRK2	600456	153	100	100	100
NUBPL	613621	108	100	95	88
NUMA1	164009	166	100	100	100
NUP107	607617	55	97	91	79
NUP214	114350	137	100	100	97
NUP62	605815	345	100	100	100
NUS1	610463	109	93	82	70
NYX	300278	184	100	100	100
OAT	613349	75	89	83	73
OBSL1	610991	283	100	100	100
OCA2	611409	152	100	97	96
OCLN	602876	100	85	81	69
OCRL	300535	75	100	98	91
ODAPH	614829	114	100	100	100
ODC1	165640	94	100	100	98
OFD1	300170	51	97	80	64
OGDH	613022	195	100	100	100
OGG1	601982	201	100	100	100
OPA1	605290	55	90	79	67
OPA3	606580	317	100	100	100
OPHN1	300127	71	100	99	91
OPLAH	614243	347	100	100	100
OPN1LW	300822	105	89	87	81
OPN1MW	300821	24	53	35	31
OPN1SW	613522	133	100	100	100
OPTN	602432	81	99	90	77
ORA11	610277	276	100	99	99
ORC1	601902	148	100	100	100
ORC4	603056	43	96	83	61
ORC6	607213	104	100	98	89

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
OSMR	601743	106	100	99	94
OSTM1	607649	118	93	70	64
OTC	300461	49	99	86	71
OTOA	607038	120	81	78	75
OTOF	603681	267	100	100	99
OTOG	604487	246	100	100	100
OTOGL	614925	70	95	91	83
OTX2	600037	208	100	100	100
OXCT1	601424	93	100	100	95
P2RX1	600845	201	100	100	100
P2RX2	600844	216	100	100	100
P2RX6	608077	254	100	100	100
P2RY12	600515	102	100	100	97
P3H1	610339	199	100	100	100
P3H2	610341	102	100	100	96
P4HB	176790	174	100	100	100
PABPN1	602279	82	98	83	72
PACS1	607492	151	100	99	97
PACS2	610423	210	100	100	99
PAFAH1B1	601545	72	86	86	85
PAH	612349	121	100	100	99
PAK3	300142	41	94	88	73
PALB2	610355	103	100	99	97
PAM16	614336	190	100	100	100
PANK2	606157	117	100	99	97
PANX1	608420	112	100	100	97
PAPPA2	PAPPA2	199	100	100	100
PAPSS2	603005	124	100	100	100
PARK7	602533	76	100	99	85
PAX1	167411	289	100	100	100
PAX2	167409	211	100	100	100
PAX3	606597	178	100	100	100
PAX4	167413	212	100	100	100
PAX6	607108	267	100	100	100
PAX8	167415	221	100	100	100
PAX9	167416	352	100	100	100
PBX1	176310	97	100	100	96
PC	608786	344	100	100	100
PCARE	613425	861	100	100	100
PCBD1	126090	127	100	100	100
PCCA	232000	71	98	93	87
PCCB	232050	159	100	98	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PCDH12	605622	267	100	100	100
PCDH15	605514	152	99	97	95
PCDH19	300460	221	100	100	98
PCGF2	600346	190	100	100	100
PCK1	614168	214	100	100	100
PCK2	614095	245	100	100	100
PCLO	604918	135	100	99	97
PCM1	600299	52	95	85	70
PCNT	605925	234	100	96	94
PCSK1	162150	114	100	100	98
PCSK9	607786	259	100	100	100
PCYT1A	123695	126	100	100	99
PDCD10	609118	62	88	87	85
PDE11A	604961	113	100	97	94
PDE4D	600129	88	100	96	90
PDE6A	180071	174	100	99	95
PDE6B	180072	237	100	100	100
PDE6C	600827	82	96	84	76
PDE6G	180073	229	100	100	100
PDE6H	601190	93	100	100	100
PDE8B	603390	100	100	100	97
PDGFB	190040	184	100	100	100
PDGFRA	173490	115	100	100	99
PDGFRB	173410	235	100	100	100
PDGFRL	604584	130	100	99	96
PDHA1	300502	78	100	94	87
PDHB	179060	104	100	100	99
PDHX	608769	79	100	95	89
PDK1	602524	89	100	99	92
PDK2	602525	224	100	100	100
PDK3	300906	67	100	97	90
PDK4	602527	91	100	97	87
PDP1	605993	194	100	100	100
PDP2	615499	268	100	100	100
PDSS5B	605333	46	90	78	65
PDSS1	607429	69	100	96	82
PDSS2	610564	152	99	93	88
PDX1	600733	153	100	100	100
PDXK	179020	254	100	100	100
PDYN	131340	181	100	100	100
PDZD7	612971	247	100	100	100
PEPD	613230	216	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PER2	603426	160	98	98	97
PET100	614770	178	100	100	100
PEX1	602136	80	95	94	90
PEX10	602859	266	100	100	100
PEX11B	603867	105	100	99	95
PEX12	601758	131	100	100	100
PEX13	601789	113	100	100	100
PEX14	601791	188	100	100	100
PEX16	603360	212	100	100	100
PEX19	600279	205	100	100	100
PEX2	170993	93	100	100	100
PEX26	608666	151	100	100	100
PEX3	603164	60	100	97	88
PEX5	600414	172	100	100	100
PEX6	601498	253	100	100	99
PEX7	601757	94	100	100	94
PFKM	610681	157	100	100	100
PFN1	176610	190	100	100	100
PGAM2	612931	286	100	100	100
PGAP1	611655	42	92	74	56
PGAP2	615187	219	100	100	100
PGAP3	611801	208	100	100	100
PGK1	311800	89	100	100	95
PGM1	171900	121	100	100	99
PGM3	172100	97	100	97	91
PHC1	602978	141	100	100	98
PHEX	300550	69	100	98	87
PHF21A	608325	110	100	97	95
PHF23	612910	172	100	100	100
PHF6	300414	22	86	52	22
PHF8	300560	98	100	100	98
PHGDH	606879	199	100	100	100
PHIP	612870	68	94	90	84
PHKA1	311870	71	100	98	91
PHKA2	300798	94	100	98	92
PHKB	172490	86	98	93	87
PHKG1	172470	268	100	100	100
PHKG2	172471	307	100	100	100
PHOX2A	602753	163	100	100	100
PHOX2B	603851	140	100	95	89
PHRF1	611780	267	100	100	99
PHYH	602026	99	100	97	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PI4KA	600286	153	100	100	99
PICALM	603025	80	90	83	79
PIEZO1	611184	276	100	100	100
PIEZO2	613629	95	99	96	91
PIGA	311770	62	100	99	86
PIGC	601730	139	100	100	100
PIGG	616918	146	100	99	96
PIGL	605947	159	100	100	100
PIGM	610273	151	100	100	100
PIGN	606097	50	92	79	65
PIGO	614730	253	100	100	100
PIGQ	605754	264	100	100	100
PIGT	610272	194	100	100	100
PIGV	610274	186	100	100	100
PIGW	610275	131	100	100	100
PIGY	610662	69	100	100	100
PIK3CA	171834	69	100	95	89
PIK3CD	602839	284	100	100	100
PIK3R1	171833	65	100	93	82
PIK3R2	603157	272	96	94	93
PIK3R5	611317	219	100	100	100
PIKFYVE	609414	93	99	96	92
PINK1	608309	258	100	100	98
PIP5K1C	606102	220	100	99	97
PITPNM3	608921	213	100	99	98
PITX1	602149	285	100	100	100
PITX2	601542	197	100	100	97
PITX3	602669	225	100	100	100
PJKV	610219	107	100	100	99
PKD1	601313	252	97	97	97
PKD1L1	609721	142	100	99	97
PKD2	173910	102	100	95	88
PKHD1	606702	136	100	100	99
PKLR	609712	320	100	100	100
PKP1	601975	181	100	100	100
PKP2	602861	150	100	100	100
PLA2G4A	600522	69	98	93	83
PLA2G5	601192	212	100	100	100
PLA2G6	603604	287	100	100	100
PLA2G7	601690	71	94	87	81
PLAG1	603026	162	100	100	100
PLAU	191840	181	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PLCB1	607120	80	98	94	87
PLCB4	600810	68	100	95	82
PLCD1	602142	245	100	100	100
PLCE1	608414	112	100	99	96
PLCG2	600220	146	100	100	98
PLEC	601282	393	100	100	100
PLEKHG5	611101	229	100	100	100
PLEKHM1	611466	191	99	98	98
PLG	173350	118	100	99	97
PLIN1	170290	238	100	100	100
PLK4	605031	77	98	90	83
PLN	172405	43	100	99	85
PLOD1	153454	233	100	100	100
PLOD2	601865	68	91	89	83
PLOD3	603066	212	100	100	100
PLP1	300401	86	100	98	92
PLPBP	604436	120	100	100	99
PLS3	300131	50	100	92	73
PLXNA3	300022	179	100	100	100
PLXND1	604282	243	100	99	99
PML	102578	255	100	100	100
PMM2	601785	115	100	100	99
PMP22	601097	127	100	100	100
PMPCA	613036	189	100	100	100
PMS2	600259	108	99	94	87
PNKP	605610	246	100	100	100
PNLIP	246600	96	100	100	98
PNP	164050	329	100	100	98
PNPLA1	612121	220	100	100	99
PNPLA2	609059	214	100	100	100
PNPLA6	603197	269	100	100	100
PNPO	603287	181	100	100	100
PNPT1	610316	57	84	76	65
POC1A	614783	216	100	100	100
POF1B	300603	51	99	85	67
POFUT1	607491	199	100	100	100
POGLUT1	615618	87	100	99	91
POGZ	614787	154	100	100	99
POLD1	174761	291	100	100	100
POLE	174762	195	100	100	100
POLG	174763	188	100	100	100
POLG2	604983	110	97	89	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
POLH	603968	117	100	100	100
POLR1A	616404	161	100	100	100
POLR1C	610060	136	100	100	100
POLR1D	613715	123	100	100	100
POLR3A	614258	143	100	100	99
POLR3B	614366	93	100	97	92
POLR3H	POLR3H	183	100	100	100
POMC	176830	351	100	100	100
POMGNT1	606822	228	100	100	100
POMGNT2	614828	358	100	100	100
POMK	615247	206	100	100	100
POMP	613386	76	85	85	85
POMT1	607423	155	100	100	100
POMT2	607439	154	100	100	97
POP1	602486	129	100	100	99
POR	124015	296	100	100	100
PORCN	300651	129	100	100	99
POU1F1	173110	65	92	91	85
POU3F3	602480	134	81	72	68
POU3F4	300039	397	100	100	100
POU4F3	602460	364	100	100	100
PPA2	609988	58	89	72	58
PPARG	601487	108	100	100	99
PPFIA4	603145	219	100	100	100
PIIB	123841	189	100	100	100
PPM1D	605100	133	100	100	98
PPM1K	611065	152	100	99	96
PPOX	600923	195	100	100	100
PPP1CB	600590	90	100	100	97
PPP1R13L	607463	219	100	100	100
PPP1R15B	613257	183	100	100	100
PPP1R3A	600917	81	98	96	93
PPP2CA	176915	86	99	94	81
PPP2R1A	605983	241	100	100	100
PPP2R1B	603113	95	96	95	92
PPP2R2B	604325	108	100	100	99
PPP2R3C	615902	58	97	83	64
PPP2R5B	601644	155	100	100	100
PPP2R5C	601645	84	95	89	79
PPP2R5D	601646	172	100	100	100
PPP3CA	114105	97	100	99	93
PPT1	600722	145	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PQBP1	300463	124	100	100	100
PRCC	179755	168	100	100	96
PRCD	610598	137	100	100	100
PRDM16	605557	245	100	100	100
PRDM5	614161	96	95	90	85
PRDM6	616982	152	100	100	98
PRELID2	No ID	96	95	89	88
PRF1	170280	383	100	100	100
PRG4	604283	113	100	96	93
PRICKLE1	608500	107	100	100	99
PRICKLE2	608501	239	100	100	100
PRIM1	176635	68	91	87	79
PRIMPOL	615421	41	92	76	60
PRKAG2	602743	154	93	90	86
PRKAR1A	188830	98	100	100	99
PRKCA	176960	120	100	100	100
PRKCE	176975	205	100	100	100
PRKCG	176980	212	100	100	100
PRKCSH	177060	260	100	100	100
PRKD1	605435	108	99	94	90
PRKG1	176894	84	98	91	89
PRKN	602544	138	100	100	99
PRKRA	603424	101	100	100	99
PRLR	176761	149	100	99	95
PRMT7	610087	208	100	100	99
PRNP	176640	470	100	100	100
PROC	612283	275	100	100	100
PRODH	606810	222	100	99	98
PROK2	607002	84	100	100	100
PROKR2	607123	340	100	100	100
PROM1	604365	67	95	88	82
PROP1	601538	186	100	100	100
PROS1	176880	68	93	91	84
PRPF3	607301	74	100	100	96
PRPF31	606419	249	100	100	100
PRPF6	613979	186	100	100	100
PRPF8	607300	159	100	100	100
PRPH2	179605	418	100	100	100
PRPS1	311850	70	100	98	92
PRR12	616633	202	100	100	100
PRRT2	614386	179	100	100	98
PRRX1	167420	124	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PRSS1	276000	209	100	100	100
PRSS12	606709	157	100	100	100
PRSS56	613858	203	100	100	100
PRUNE1	617413	140	100	100	100
PRX	605725	412	100	100	100
PSAP	176801	134	100	100	99
PSAT1	610936	103	100	98	90
PSENN	607632	150	100	100	100
PSMB8	177046	205	100	100	100
PSMC3IP	608665	107	100	99	95
PSMD12	604450	69	100	98	92
PSPH	172480	97	96	83	78
PSTPIP1	606347	257	100	100	100
PTCH1	601309	201	100	100	98
PTCH2	603673	280	100	100	100
PTCHD1	300828	101	100	100	99
PTDSS1	612792	101	100	100	100
PTEN	601728	100	99	94	87
PTF1A	607194	138	100	97	93
PTGIS	601699	193	100	98	96
PTGR1	601274	87	100	98	96
PTH	168450	89	100	100	95
PTH1R	168468	266	100	100	100
PTHLH	168470	208	100	100	100
PTPN11	176876	80	98	94	89
PTPN12	600079	76	96	87	80
PTPN14	603155	194	100	99	97
PTPRC	151460	62	93	84	76
PTPRJ	600925	117	96	93	91
PTPRO	600579	92	99	97	95
PTPRQ	603317	57	96	88	75
PTRH2	608625	192	100	100	100
PTRHD1	617342	264	100	100	100
PTS	612719	119	100	100	100
PUF60	604819	237	100	100	100
PUM1	607204	145	100	100	100
PURA	600473	481	100	100	100
PUS1	608109	233	100	100	100
PUS3	616283	128	100	100	99
PUS7	616261	88	100	96	93
PYCR1	179035	285	100	100	100
PYCR2	616406	201	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PYGL	613741	128	100	100	99
PYGM	608455	232	100	100	100
QARS1	603727	206	100	100	100
QDPR	612676	134	100	100	100
RAB18	602207	68	91	90	85
RAB23	606144	64	100	88	85
RAB27A	603868	88	100	100	97
RAB28	612994	53	85	72	58
RAB33B	605950	201	100	100	98
RAB39B	300774	156	100	100	99
RAB3GAP1	602536	86	100	99	95
RAB3GAP2	609275	96	97	91	81
RAB40AL	300405	174	100	100	100
RAB7A	602298	115	100	100	100
RAC1	602048	113	100	95	86
RAC2	602049	233	100	100	100
RAD21	606462	63	100	98	93
RAD50	604040	51	94	76	57
RAD51	179617	102	88	88	88
RAD51C	602774	98	100	90	88
RAD54B	604289	63	95	82	72
RAD54L	603615	150	100	100	100
RAF1	164760	124	100	98	97
RAG1	179615	199	100	100	100
RAG2	179616	130	100	100	100
RAI1	607642	405	100	100	100
RALA	179550	45	80	75	65
RAP1GDS1	179502	58	100	96	85
RAPSN	601592	334	100	100	100
RARB	180220	98	100	98	92
RARS2	611524	70	100	93	77
RASA1	139150	62	95	80	66
RASGRP2	605577	310	100	100	100
RAX2	610362	192	100	100	97
RB1	614041	54	90	74	62
RB1CC1	606837	56	86	72	61
RBBP8	604124	53	99	89	79
RBCK1	610924	200	100	100	100
RBFOX1	605104	143	95	95	91
RBM10	300080	130	100	100	100
RBM20	613171	248	100	100	98
RBM28	612074	110	100	99	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
RBM8A	605313	82	100	100	95
RBP4	180250	281	100	100	100
RBPJ	147183	79	99	91	81
RBSN	609511	172	100	100	99
RCBTB1	607867	129	100	100	96
RD3	180040	148	100	100	100
RDH12	608830	147	100	100	100
RDH5	601617	283	100	100	100
RDX	179410	58	86	72	61
RECQL4	603780	312	100	100	100
REEP1	609139	164	100	100	98
RELN	600514	114	100	99	97
REN	179820	168	100	100	100
RERE	605226	243	100	100	99
RET	164761	217	100	100	100
RETREG1	613114	112	96	89	85
REV3L	602776	86	98	96	91
RFT1	611908	134	100	95	89
RFTN2	618215	135	100	100	96
RFX5	601863	180	100	100	100
RFX6	612659	109	98	95	91
RFXANK	603200	218	100	100	100
RFXAP	601861	136	100	100	100
RGR	600342	138	100	100	100
RGS9	604067	162	100	100	98
RGS9BP	607814	268	100	100	100
RHAG	180297	95	100	100	100
RHBDF2	614404	259	100	100	100
RHCE	111700	189	100	99	95
RHEB	601293	28	90	59	39
RHO	180380	270	100	100	100
RHOBTB2	607352	232	100	100	100
RIMS1	606629	110	100	97	92
RIN2	610222	151	100	100	99
RIPK4	605706	383	100	100	100
RIPPLY2	609891	129	90	64	64
RIT1	609591	123	100	100	100
RLBP1	180090	205	100	100	100
RLIM	300379	59	100	100	94
RMND1	614917	87	95	83	77
RMRP	157660	No coverage data			
RNASEH2A	606034	265	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	100	100	100
RNASEL	180435	120	100	100	99
RNASET2	612944	99	93	92	84
RNF113A	300951	95	100	100	100
RNF123	614472	206	100	100	100
RNF125	610432	101	100	97	84
RNF135	611358	145	100	100	98
RNF139	603046	109	100	100	99
RNF145	No ID	71	97	92	83
RNF168	612688	137	99	97	93
RNF170	614649	64	100	91	76
RNF212	612041	95	100	91	86
RNF216	609948	149	100	98	97
RNF6	604242	139	100	100	100
RNU4ATAC	601428	No coverage data			
ROBO1	602430	133	100	100	99
ROBO2	602431	106	100	99	96
ROBO3	608630	213	100	100	100
ROBO4	607528	199	100	100	100
ROGDI	614574	222	100	99	97
ROM1	180721	171	100	100	100
ROR2	602337	278	100	100	98
RORA	600825	125	100	98	93
RP1	603937	109	100	99	95
RP1L1	608581	188	100	99	96
RP2	300757	71	100	100	88
RPE65	180069	92	100	100	96
RPGR	312610	35	70	62	51
RPGRIP1	605446	119	100	99	97
RPGRIP1L	610937	80	95	91	83
RPIA	180430	151	100	100	100
RPL10	312173	136	100	100	100
RPL11	604175	155	100	100	100
RPL35A	180468	109	89	89	89
RPL5	603634	69	89	85	74
RPS10	603632	115	100	100	96
RPS14	130620	180	100	100	100
RPS17	180472	1	5	5	0
RPS19	603474	178	100	100	100
RPS24	602412	138	100	100	100
RPS26	603701	118	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
RPS6KA3	300075	37	96	80	55
RPS7	603658	93	100	97	88
RPSA	150370	110	100	100	95
RRAS2	600098	90	100	90	82
RRM2B	604712	117	100	100	98
RS1	300839	110	100	100	94
RSPH1	609314	91	100	100	98
RSPH4A	612647	131	100	100	97
RSPH9	612648	215	100	100	100
RSPO1	609595	204	100	100	100
RSPO4	610573	234	100	100	100
RSPRY1	616585	93	100	96	94
RTEL1	608833	260	100	100	100
RTN2	603183	218	100	100	100
RTTN	610436	74	98	94	87
RUBCN	613516	163	100	100	100
RUNX1	151385	200	100	96	96
RUNX2	600211	160	100	100	100
RUSC2	611053	241	100	100	100
RXFP2	606655	72	93	86	78
RXYLT1	605862	75	99	91	82
RYR1	180901	256	100	99	99
RYR2	180902	133	99	94	90
SACS	604490	97	100	100	98
SAG	181031	133	100	100	100
SALL1	602218	214	100	100	100
SALL4	607343	229	100	100	100
SAMD9	610456	73	99	94	89
SAMHD1	606754	97	100	99	93
SAR1B	607690	66	99	88	88
SARDH	604455	274	100	100	100
SARS2	612804	272	100	100	100
SART3	611684	126	100	97	93
SAT1	313020	87	100	100	97
SATB1	602075	106	100	100	97
SATB2	608148	139	100	100	99
SBDS	607444	131	100	100	99
SBF2	607697	100	100	98	94
SC5D	602286	79	100	98	90
SCAF4	616023	104	98	95	90
SCARB2	602257	79	100	97	91
SCARF2	613619	234	100	99	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SCN10A	604427	150	100	100	99
SCN11A	604385	100	100	99	94
SCN1A	182389	97	100	97	91
SCN1B	600235	224	100	100	100
SCN2A	182390	93	98	94	86
SCN2B	601327	167	100	100	100
SCN3A	182391	92	100	96	90
SCN3B	608214	139	100	100	100
SCN4A	603967	238	100	100	100
SCN4B	608256	173	100	100	100
SCN5A	600163	208	100	100	99
SCN8A	600702	137	100	100	98
SCN9A	603415	87	98	92	82
SCNN1A	600228	236	100	100	100
SCNN1B	600760	192	100	100	100
SCNN1G	600761	186	100	100	100
SCO1	603644	245	100	100	100
SCO2	604272	377	100	100	100
SCP2	184755	81	98	90	90
SDCCAG8	613524	66	100	91	78
SDHA	600857	155	100	100	100
SDHAF1	612848	141	100	100	100
SDHAF2	613019	76	100	100	98
SDHB	185470	123	100	100	98
SDHC	602413	143	100	100	100
SDHD	602690	123	100	100	100
SEC23A	610511	76	100	95	88
SEC23B	610512	125	97	97	96
SEC24D	607186	96	100	99	96
SEC63	608648	50	98	89	73
SECISBP2	607693	88	100	98	92
SELENON	606210	160	91	90	90
SEMA3A	603961	102	100	99	97
SEMA3E	608166	91	93	91	87
SEMA4A	607292	197	100	100	100
SEPSECS	613009	88	99	96	90
SEPTIN12	611562	149	100	100	100
SEPTIN9	604061	287	100	100	100
SERAC1	614725	98	98	95	89
SERPINA1	107400	176	100	100	100
SERPINA3	107280	159	100	100	100
SERPINA6	122500	172	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SERPINA7	314200	80	100	100	98
SERPINB6	173321	151	100	100	95
SERPINB7	603357	92	100	100	94
SERPINC1	107300	157	100	100	100
SERPIND1	142360	139	100	100	100
SERPINE1	173360	217	100	100	100
SERPINF1	172860	117	100	100	98
SERPINF2	613168	235	100	100	100
SERPING1	606860	113	100	100	99
SERPINH1	600943	369	100	100	100
SERPINI1	602445	58	100	98	89
SETBP1	611060	171	100	100	99
SETD1A	611052	215	100	99	99
SETD2	612778	113	100	99	98
SETD5	615743	121	100	99	96
SETX	608465	100	100	97	95
SF1	601516	130	92	86	82
SF3B1	605590	73	97	94	89
SF3B4	605593	172	100	100	100
SFTPA2	178642	255	100	100	100
SFTPB	178640	241	100	100	100
SFTPC	178620	186	100	100	100
SFXN4	615564	71	99	88	78
SGCA	600119	238	100	100	100
SGCB	600900	86	100	99	93
SGCD	601411	80	100	99	94
SGCE	604149	104	97	86	82
SGCG	608896	102	100	100	97
SGO1	609168	55	96	90	82
SGSH	605270	272	99	94	94
SH2B3	605093	211	100	100	100
SH2D1A	300490	81	100	95	85
SH3BP2	602104	248	94	91	91
SH3KBP1	300374	85	100	97	92
SH3PXD2B	613293	196	100	100	100
SH3TC2	608206	171	100	100	100
SHANK2	603290	323	98	98	98
SHANK3	606230	246	99	98	97
SHH	600725	259	100	100	100
SHMT1	182144	177	100	100	100
SHMT2	138450	252	100	100	100
SHOC2	602775	74	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SHOX	312865	45	50	50	48
SHROOM4	300579	116	100	99	96
SI	609845	65	94	87	79
SIGMAR1	601978	318	100	100	100
SIK1	605705	309	100	100	100
SIL1	608005	202	100	100	98
SIM1	603128	243	100	100	100
SIN3A	607776	114	100	100	99
SIX1	601205	284	100	100	100
SIX3	603714	279	100	100	100
SIX5	600963	167	100	100	100
SIX6	606326	241	100	100	100
SKI	164780	208	100	100	98
SKIV2L	600478	190	100	100	100
SLC10A2	601295	106	100	100	98
SLC10A7	611459	67	94	81	68
SLC11A2	600523	102	100	99	97
SLC12A1	600839	88	100	96	88
SLC12A3	600968	199	100	100	99
SLC12A6	604878	98	100	97	95
SLC13A5	608305	194	100	100	100
SLC16A1	600682	117	100	100	99
SLC16A12	611910	97	100	100	98
SLC16A2	300095	101	100	100	100
SLC17A5	604322	82	100	94	85
SLC17A8	607557	109	100	100	98
SLC19A1	600424	308	100	100	100
SLC19A2	603941	208	100	99	96
SLC19A3	606152	108	100	100	100
SLC1A1	133550	114	100	100	99
SLC1A2	600300	139	100	100	100
SLC1A3	600111	106	100	99	95
SLC1A4	600229	125	100	100	100
SLC20A2	158378	185	100	95	95
SLC22A12	607096	206	100	100	100
SLC22A18	602631	166	100	100	100
SLC22A5	603377	177	100	100	99
SLC23A2	603791	138	100	99	95
SLC24A1	603617	139	100	100	99
SLC24A5	609802	76	96	84	73
SLC25A1	190315	204	100	100	100
SLC25A12	603667	94	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC25A13	603859	88	100	98	89
SLC25A15	603861	152	100	100	100
SLC25A19	606521	168	100	100	100
SLC25A20	613698	155	100	100	100
SLC25A22	609302	317	100	100	100
SLC25A24	608744	95	97	91	89
SLC25A3	600370	131	100	100	100
SLC25A32	610815	106	100	100	98
SLC25A38	610819	181	100	100	100
SLC25A4	103220	200	100	100	100
SLC26A2	606718	138	100	100	100
SLC26A3	126650	78	100	95	88
SLC26A4	605646	121	100	96	91
SLC26A5	604943	86	98	96	94
SLC26A8	608480	102	100	99	96
SLC27A4	604194	318	100	100	99
SLC29A3	612373	233	100	100	100
SLC2A1	138140	354	100	100	100
SLC2A10	606145	216	100	100	100
SLC2A2	138160	76	100	98	94
SLC2A9	606142	158	100	100	98
SLC30A10	611146	209	100	100	100
SLC30A2	609617	210	100	100	100
SLC33A1	603690	147	98	93	85
SLC34A1	182309	200	100	100	100
SLC34A2	604217	134	100	100	100
SLC34A3	609826	248	100	100	100
SLC35A1	605634	69	100	99	95
SLC35A2	314375	196	100	100	99
SLC35A3	605632	49	93	85	76
SLC35C1	605881	329	100	100	100
SLC35D1	610804	89	92	88	76
SLC36A2	608331	135	100	98	93
SLC37A4	602671	193	100	100	100
SLC38A8	615585	213	100	100	100
SLC39A12	608734	85	100	97	88
SLC39A13	608735	246	100	100	100
SLC39A14	608736	159	100	100	100
SLC39A4	607059	284	100	100	100
SLC39A8	608732	91	100	93	85
SLC3A1	104614	133	99	96	89
SLC40A1	604653	105	100	98	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC45A2	606202	127	100	100	97
SLC46A1	611672	271	100	100	100
SLC4A1	109270	268	100	100	100
SLC4A11	610206	304	100	100	100
SLC4A4	603345	110	100	99	96
SLC52A1	607883	285	100	100	100
SLC52A2	607882	210	100	100	100
SLC52A3	613350	271	100	100	100
SLC5A1	182380	136	100	100	98
SLC5A2	182381	258	100	100	100
SLC5A5	601843	198	100	100	100
SLC5A7	608761	130	100	100	98
SLC6A1	137165	177	100	100	98
SLC6A17	610299	180	100	100	100
SLC6A19	608893	229	100	100	100
SLC6A20	605616	205	100	100	100
SLC6A3	126455	226	100	100	100
SLC6A5	604159	154	100	100	100
SLC6A8	300036	134	100	100	97
SLC7A14	615720	166	100	100	100
SLC7A7	603593	146	100	100	100
SLC7A9	604144	154	100	100	99
SLC9A3R1	604990	174	100	100	100
SLC9A6	300231	89	98	88	81
SLC9A7	300368	70	100	96	91
SLCO1B1	604843	49	94	80	66
SLCO1B3	605495	48	94	81	67
SLCO2A1	601460	152	100	100	99
SLCO5A1	613543	148	91	91	89
SLITRK1	609678	210	100	100	100
SLITRK6	609681	134	100	100	100
SLURP1	606119	390	100	100	100
SLX4	613278	192	100	100	100
SMAD2	601366	87	100	100	94
SMAD3	603109	239	100	100	100
SMAD4	600993	78	100	97	95
SMAD6	602931	188	100	100	100
SMAD9	603295	117	100	99	97
SMARCA2	600014	95	100	98	96
SMARCA4	603254	225	100	100	100
SMARCAD1	612761	52	92	78	64
SMARCAL1	606622	146	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SMARCB1	601607	216	100	100	100
SMARCC2	601734	143	100	100	100
SMARCE1	603111	102	100	94	83
SMC1A	300040	96	100	96	95
SMC3	606062	46	93	80	60
SMCHD1	614982	63	97	89	74
SMN1	600354	3	7	7	7
SMO	601500	213	100	100	100
SMOC1	608488	133	100	99	97
SMOC2	607223	171	100	100	100
SMPD1	607608	377	100	100	100
SMPD4	610457	243	100	99	97
SMPX	300226	34	100	80	40
SMS	300105	61	100	93	86
SNAI2	602150	151	100	100	100
SNAP25	600322	96	100	100	96
SNAP29	604202	107	100	100	95
SNCA	163890	106	100	100	100
SNCB	602569	155	100	100	100
SNIP1	608241	141	100	100	98
SNRNP200	601664	164	100	100	100
SNRNP70	180740	159	100	100	100
SNRPB	182282	124	100	100	100
SNRPE	128260	42	94	83	70
SNRPN	182279	141	100	100	100
SNTA1	601017	195	100	100	98
SNX10	614780	71	100	100	98
SNX14	616105	43	83	65	53
SOBP	613667	172	100	98	97
SOD1	147450	154	100	100	100
SON	182465	170	100	98	94
SOS1	182530	74	94	89	79
SOS2	601247	64	95	80	67
SOST	605740	330	100	100	100
SOX10	602229	381	100	100	100
SOX11	600898	201	100	100	100
SOX17	610928	198	100	100	100
SOX18	601618	130	95	92	87
SOX2	184429	257	100	100	100
SOX3	313430	155	100	100	100
SOX5	604975	117	100	100	98
SOX6	607257	118	96	96	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SOX8	605923	169	100	100	100
SOX9	608160	207	100	100	99
SP110	604457	117	96	93	92
SP7	606633	258	100	100	100
SPAG1	603395	59	93	80	68
SPARC	182120	141	100	100	100
SPART	607111	96	100	98	95
SPAST	604277	82	99	84	69
SPATA16	609856	74	93	91	84
SPATA5	613940	95	100	99	94
SPATA7	609868	114	88	81	77
SPECC1L	614140	109	100	97	92
SPG11	610844	105	99	94	88
SPG21	608181	87	100	98	90
SPG7	602783	227	100	100	100
SPINK1	167790	70	100	100	100
SPINK5	605010	65	96	87	77
SPINT2	605124	169	100	100	100
SPOCK1	602264	143	100	100	98
SPR	182125	142	100	100	100
SPRED1	609291	108	100	100	98
SPRY4	607984	277	100	100	100
SPTA1	182860	115	100	99	97
SPTAN1	182810	147	100	99	98
SPTB	182870	186	100	100	99
SPTBN2	604985	211	100	99	98
SPTLC1	605712	98	100	99	95
SPTLC2	605713	111	100	100	99
SQSTM1	601530	268	100	100	100
SRC	190090	215	100	100	100
SRCAP	611421	186	100	100	100
SRD5A2	607306	114	100	100	100
SRD5A3	611715	138	100	100	100
SRP72	602122	79	100	93	87
SRPX2	300642	102	100	100	100
SRY	480000	382	100	100	100
SSR3	606213	97	100	98	83
SSR4	300090	139	100	100	100
SSTR5	182455	184	100	100	100
ST14	606797	210	100	100	100
ST3GAL3	606494	156	100	100	100
ST3GAL5	604402	100	100	93	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
STAC3	615521	150	100	98	95
STAG1	604358	73	95	89	83
STAG3	608489	143	100	100	99
STAMBP	606247	126	100	92	92
STAR	600617	186	100	100	100
STAT1	600555	91	97	95	90
STAT3	102582	135	100	100	99
STAT5B	604260	157	100	100	100
STIL	181590	192	100	99	95
STIM1	605921	202	100	100	100
STK11	602216	310	100	100	100
STK4	604965	97	100	95	81
STOX1	609397	106	93	89	88
STRA6	610745	229	100	100	100
STRADA	608626	154	100	100	96
STRC	606440	67	60	50	46
STS	300747	106	100	100	97
STT3A	601134	119	100	100	100
STT3B	608605	67	99	92	83
STX1B	601485	139	100	100	100
STXBP1	602926	126	100	100	100
STXBP2	601717	220	100	100	100
SUCLA2	603921	74	100	98	88
SUCLG1	611224	90	100	100	96
SUCO	No ID	73	93	90	82
SUFU	607035	206	100	100	100
SUGCT	609187	121	100	93	90
SULF1	610012	99	100	98	92
SUMF1	607939	174	100	100	100
SUMO1	601912	29	76	60	34
SUOX	606887	254	100	100	100
SURF1	185620	158	100	98	95
SUZ12	606245	49	79	75	65
SYCE1	611486	159	100	100	100
SYCP3	604759	44	86	72	68
SYN1	313440	151	100	100	100
SYNCRIP	616686	88	100	98	90
SYNE1	608441	113	100	98	95
SYNE2	608442	74	98	91	84
SYNE4	615535	173	100	100	97
SYNGAP1	603384	201	100	98	98
SYNJ1	604297	83	98	95	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
SYP	313475	110	100	100	99
SYT1	185605	69	97	88	79
SYT14	610949	71	100	94	82
SZT2	615463	206	100	100	100
TAB2	605101	120	100	99	93
TAC3	162330	133	100	100	100
TACO1	612958	109	100	100	100
TACR3	162332	160	100	100	96
TACSTD2	137290	617	100	100	100
TAF1	313650	71	100	99	95
TAF2	604912	66	98	89	80
TAL1	187040	141	100	98	94
TAL2	186855	133	100	100	100
TALDO1	602063	133	100	100	100
TANGO2	616830	253	100	100	100
TAP1	170260	172	100	100	100
TAP2	170261	166	100	100	100
TAPBP	601962	233	100	100	100
TAPT1	612758	58	94	80	66
TARDBP	605078	152	100	100	97
TAT	613018	141	100	100	100
TAZ	300394	145	100	100	100
TBC1D20	611663	149	100	100	96
TBC1D24	613577	390	100	100	100
TBC1D7	612655	94	95	85	85
TBCD	604649	210	99	97	95
TBCE	604934	69	100	96	87
TBCK	616899	56	95	88	76
TBL1XR1	608628	68	100	90	81
TBP	600075	140	100	99	90
TBR1	604616	270	100	100	100
TBX1	602054	131	94	90	87
TBX15	604127	193	100	100	99
TBX19	604614	114	100	100	99
TBX20	606061	158	100	100	100
TBX21	604895	177	100	100	100
TBX22	300307	63	100	94	84
TBX3	601621	175	100	100	100
TBX4	601719	204	100	100	100
TBX5	601620	233	100	100	98
TBX6	602427	280	100	100	100
TBXAS1	274180	174	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
TCAP	604488	300	100	100	100
TCF12	600480	94	100	99	96
TCF20	603107	182	100	100	100
TCF4	602272	145	100	99	97
TCIRG1	604592	242	100	100	100
TCN1	189905	73	97	92	84
TCN2	613441	190	100	100	100
TCOF1	606847	201	100	100	98
DYNLT2B	617353	146	100	100	88
TCTN1	609863	117	98	97	95
TCTN2	613846	108	100	95	93
TCTN3	613847	126	100	100	98
TDGF1	187395	148	100	100	100
TDO2	191070	55	100	92	74
TDP1	607198	104	100	99	93
TDP2	605764	121	89	88	84
TDRD7	611258	94	97	92	87
TEAD1	189967	116	100	100	100
TECPR2	615000	195	100	99	97
TECR	610057	156	100	100	100
TECTA	602574	226	100	100	100
TEK	600221	110	100	99	96
TELO2	611140	246	100	100	100
TENM3	610083	151	100	100	99
TERC	602322	No coverage data			
TET2	612839	123	100	100	98
TEX28	300092	No coverage data			
TF	190000	119	100	100	99
TFAP2A	107580	236	100	100	100
TFAP2B	601601	162	100	100	100
TFE3	314310	142	100	100	100
TFG	602498	86	99	89	83
TFR2	604720	243	100	100	100
TG	188450	177	100	100	100
TGDS	616146	51	94	78	62
TGFB1	190180	274	100	100	100
TGFB2	190220	126	100	100	97
TGFB3	190230	182	100	100	100
TGFBI	601692	153	100	100	100
TGFBR1	190181	110	97	94	92
TGFBR2	190182	214	100	100	99
TGIF1	602630	133	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
TGM1	190195	276	100	100	100
TGM5	603805	201	100	100	100
TGM6	613900	265	100	100	100
TH	191290	266	100	100	100
THAP1	609520	115	100	100	96
THBD	188040	398	100	100	100
THOC2	300395	34	82	62	47
THOC6	615403	231	100	100	100
THPO	600044	378	100	100	100
THRA	190120	260	100	100	100
THRB	190160	137	100	100	100
TIA1	603518	82	100	98	89
TIMM8A	300356	98	100	100	100
TIMP3	188826	203	100	100	100
TINF2	604319	229	100	100	100
TJP2	607709	125	100	100	98
TK2	188250	119	100	97	88
TLK2	608439	83	98	90	80
TLL1	606742	82	100	97	91
TLR4	603030	130	100	100	100
TM4SF20	615404	75	100	100	99
TMC1	606706	86	99	95	89
TMC6	605828	223	100	100	100
TMC8	605829	156	100	100	100
TMCO1	614123	96	91	89	82
TMEM126A	612988	66	100	93	77
TMEM138	614459	110	100	100	100
TMEM165	614726	92	100	99	93
TMEM181	613209	103	97	96	93
TMEM199	616815	142	100	100	100
TMEM216	613277	142	100	100	100
TMEM231	614949	271	100	100	98
TMEM237	614423	77	98	98	95
TMEM240	616101	287	100	100	100
TMEM38B	611236	89	100	100	93
TMEM67	609884	61	90	74	62
TMEM70	612418	103	100	98	94
TMIE	607237	135	94	76	76
TMLHE	300777	63	89	86	82
TMPRSS15	606635	61	88	82	77
TMPRSS3	605511	207	100	100	100
TMPRSS6	609862	264	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
TMPRSS7	No ID	106	100	100	97
TMTC3	617218	47	96	79	62
TMX2	616715	147	100	100	100
TNC	187380	225	100	100	100
TNFRSF10B	603612	159	100	100	100
TNFRSF11A	603499	178	95	95	95
TNFRSF11B	602643	122	100	100	97
TNFRSF13B	604907	241	100	100	100
TNFRSF13C	606269	135	100	98	90
TNFRSF1A	191190	204	100	100	100
TNFSF11	602642	119	100	100	100
TNIK	610005	107	100	98	92
TNNC1	191040	309	100	100	100
TNNI2	191043	312	100	100	100
TNNI3	191044	218	100	100	100
TNNT1	191041	157	100	100	99
TNNT2	191045	141	100	100	100
TNNT3	600692	203	100	100	100
TNXB	600985	243	95	93	92
TOE1	613931	204	100	100	100
TOP1	126420	61	93	83	76
TOP2A	126430	62	99	91	79
TOPORS	609507	125	100	100	100
TP53	191170	246	100	100	100
TP63	603273	134	100	100	99
TPI1	190450	190	100	100	100
TPK1	606370	69	100	98	85
TPM1	191010	128	100	97	87
TPM2	190990	126	100	100	98
TPM3	191030	90	100	100	97
TPMT	187680	61	100	95	86
TPO	606765	222	100	100	99
TPP1	607998	223	100	100	100
TPRN	613354	185	93	88	83
TRAIP	605958	175	100	100	100
TRAPPC11	614138	77	97	94	90
TRAPPC2	300202	40	88	60	43
TRAPPC6B	610397	41	91	56	49
TRAPPC9	611966	213	100	100	100
TRDN	603283	43	68	52	41
TREH	275360	218	100	100	99
TREM2	605086	225	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
TREX1	606609	313	100	100	100
TRHR	188545	144	100	100	98
TRIM24	603406	75	95	88	75
TRIM32	602290	222	100	100	100
TRIM33	605769	100	100	95	90
TRIM37	605073	69	100	97	91
TRIO	601893	159	99	99	97
TRIOBP	609761	259	100	100	100
TRIP11	604505	44	90	73	58
TRIP12	604506	87	100	99	97
TRMT1	611669	217	100	100	100
TRMT10A	616013	63	100	94	84
TRMU	610230	192	100	99	91
TRPA1	604775	72	89	81	77
TRPC6	603652	113	97	96	92
TRPM1	603576	140	100	99	95
TRPM3	608961	179	100	100	99
TRPM4	606936	174	100	100	100
TRPM6	607009	192	98	97	96
TRPS1	604386	210	100	100	100
TRPV3	607066	177	100	97	97
TRPV4	605427	299	100	100	100
TSC1	605284	153	100	100	100
TSC2	191092	314	100	100	100
TSEN15	608756	92	99	93	93
TSEN2	608753	190	87	87	84
TSEN34	608754	349	100	100	100
TSEN54	608755	201	100	100	98
TSFM	604723	182	100	99	97
TSG101	601387	86	100	99	92
TSHB	188540	84	100	100	97
TSHR	603372	139	100	99	95
TSHZ1	614427	282	100	98	98
TSPAN12	613138	96	100	98	90
TSPAN7	300096	97	100	100	99
TSPEAR	612920	207	100	100	100
TSPYL1	604714	185	100	100	100
TTBK2	611695	141	100	100	100
TTC19	613814	101	100	98	93
TTC21B	612014	63	99	95	83
TTC37	614589	65	99	96	91
TTC7A	609332	192	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
TTC8	608132	104	96	91	86
TTI2	614426	133	100	100	100
TTN	188840	136	99	98	96
TTPA	600415	93	100	100	97
TTR	176300	168	100	100	100
TUBA1A	602529	219	100	100	100
TUBA8	605742	185	100	100	100
TUBB	191130	186	100	100	100
TUBB1	612901	212	100	100	99
TUBB2A	615101	109	100	90	81
TUBB2B	612850	113	100	90	80
TUBB3	602661	330	100	100	97
TUBB4A	602662	233	100	100	100
TUBG1	191135	334	100	100	100
TUBGCP2	617817	193	100	100	100
TUBGCP4	609610	122	100	100	98
TUBGCP6	610053	356	100	100	100
TUFM	602389	218	100	100	100
TULP1	602280	172	100	97	92
TUSC3	601385	101	100	100	96
TWIST1	601622	187	100	100	98
TWIST2	607556	126	100	100	100
TWNK	606075	231	100	100	100
TXNL4A	611595	193	100	91	75
TYK2	176941	276	100	100	100
TYMP	131222	204	100	100	100
TYMS	188350	172	100	100	100
TYR	606933	147	100	99	98
TYROBP	604142	161	100	100	100
TYRP1	115501	137	100	100	100
UBA1	314370	122	100	100	98
UBA5	610552	52	93	78	66
UBE2A	312180	96	100	98	80
UBE3A	601623	79	98	89	83
UBE3B	608047	134	100	100	99
UBIAD1	611632	203	100	100	100
UBQLN2	300264	116	100	100	100
UBR1	605981	71	96	92	85
UGT1A1	191740	273	100	100	100
UMOD	191845	259	100	100	99
UMPS	613891	146	100	100	99
UNC80	612636	125	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
UNG	191525	123	100	100	99
UPB1	606673	213	100	100	100
UPF3B	300298	40	83	64	48
UQCC2	614461	124	100	100	100
UQCRB	191330	98	100	100	97
UQCRC2	191329	97	100	100	96
UQCRQ	612080	169	100	100	91
UROC1	613012	245	100	100	100
UROD	613521	191	100	100	100
UROS	606938	95	100	100	100
USB1	613276	145	100	100	93
USF1	191523	171	100	100	100
USH1C	605242	209	100	100	99
USH1G	607696	427	100	100	100
USH2A	608400	116	100	99	97
USP18	607057	132	95	95	95
USP21	604729	212	100	100	99
USP27X	300975	106	100	100	100
USP7	602519	80	99	95	90
USP9X	300072	45	96	86	71
USP9Y	400005	55	97	87	74
UTP4	607456	122	100	100	99
UTRN	128240	72	99	94	85
UVSSA	614632	237	100	100	100
VANGL1	610132	145	100	100	100
VANGL2	600533	187	100	100	100
VAPB	605704	71	100	96	82
VAX1	604294	125	97	92	89
VCAN	118661	128	100	100	99
VCL	193065	137	100	99	96
VCP	601023	140	100	100	100
VDR	601769	176	100	100	100
VHL	608537	136	100	100	100
VIM	193060	138	100	99	96
VIPAS39	613401	106	100	100	100
VKORC1	608547	146	100	98	88
VLDLR	192977	120	100	100	98
VPS11	608549	184	100	100	100
VPS13A	605978	48	95	81	63
VPS13B	607817	98	99	95	89
VPS13C	608879	63	96	87	77
VPS33B	608552	145	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
VPS35	601501	82	100	97	91
VPS37A	609927	61	100	92	80
VPS45	610035	72	100	93	92
VPS53	615850	133	100	100	100
VRK1	602168	56	100	89	72
VSX1	605020	136	100	99	98
VSX2	142993	227	100	100	100
VWA3B	614884	101	100	99	94
VWF	613160	209	99	98	98
WAC	615049	78	96	88	82
WAS	300392	113	100	100	100
WASHC4	615748	45	93	80	65
WASHC5	610657	82	100	94	87
WBP11	618083	96	98	90	83
WDPCP	613580	87	98	91	82
WDR11	606417	87	99	96	93
WDR13	300512	199	100	100	100
WDR19	608151	75	100	96	92
WDR26	617424	80	100	96	92
DYNC2I2	613363	177	100	100	100
WDR35	613602	75	97	93	87
WDR36	609669	83	98	91	84
WDR4	605924	188	100	100	100
WDR45	300526	161	100	100	100
DYNC2I1	615462	85	99	95	88
WDR62	613583	214	100	100	100
WDR72	613214	79	100	98	93
WDR73	616144	194	100	100	100
WDR81	614218	293	100	100	100
WFS1	606201	268	100	100	100
WHRN	607928	288	100	100	100
WIPF1	602357	186	100	100	99
WNK1	605232	123	100	99	96
WNK4	601844	161	100	100	100
WNT1	164820	280	100	100	100
WNT10A	606268	286	100	100	100
WNT10B	601906	270	100	100	100
WNT3	165330	317	100	100	100
WNT4	603490	289	100	92	92
WNT5A	164975	316	100	100	100
WNT7A	601570	347	100	100	100
WRAP53	612661	213	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
WRN	604611	62	95	87	77
WT1	607102	175	100	100	100
WWOX	605131	128	100	94	94
XDH	607633	164	100	100	100
XIAP	300079	63	94	89	81
XK	314850	107	100	100	100
XPA	611153	49	99	92	75
XPC	613208	138	100	100	99
XPNPEP3	613553	157	100	100	99
XRCC4	194363	39	95	77	54
XYLT1	608124	191	100	98	95
XYLT2	608125	298	100	100	99
YAP1	606608	104	100	100	97
YARS1	604061	130	100	100	100
YARS2	610957	213	100	98	95
YIF1B	No ID	196	100	100	100
YME1L1	607472	68	93	88	80
YWHAE	605066	85	87	85	82
YY1	600013	156	100	100	99
YY1AP1	607860	173	100	100	99
ZAP70	176947	312	100	100	100
ZBTB16	176797	309	100	100	100
ZBTB18	608433	289	100	100	100
ZBTB20	606025	302	100	100	100
ZBTB24	614064	155	100	100	100
ZC3H14	613279	76	98	96	88
ZC4H2	300897	88	100	100	100
ZDHHC15	300576	50	96	68	56
ZDHHC9	300646	77	100	100	98
ZEB1	189909	99	99	94	86
ZEB2	605802	289	100	100	100
ZFP57	612192	179	100	99	98
ZFPM2	603693	127	100	100	99
ZFYVE26	612012	176	100	100	99
ZFYVE27	610243	213	100	100	100
ZIC1	600470	322	100	100	100
ZIC2	603073	171	95	93	91
ZIC3	300265	137	100	100	99
ZIC4	608948	291	100	100	100
ZMPSTE24	606480	73	98	87	77
ZMYND10	607070	232	100	100	100
ZMYND11	608668	86	100	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ZNF101	603983	106	100	100	100
ZNF292	616213	98	100	99	98
ZNF335	610827	252	100	100	100
ZNF407	615894	224	100	100	100
ZNF41	314995	87	100	99	96
ZNF423	604557	316	100	100	100
ZNF469	612078	331	100	100	100
ZNF513	613598	344	100	100	100
ZNF592	613624	263	100	100	100
ZNF644	614159	86	100	99	97
ZNF674	300573	77	100	95	77
ZNF711	314990	58	99	92	81
ZNF750	610226	258	100	100	100
ZNF81	314998	59	100	99	93
ZNRF3	612062	198	97	96	96
ZSWIM6	615951	118	97	94	91

- OMIM release used: 8-9-2019

- The statistics above are based on a set of 100 samples

- Median depth is the median of the mean sequence depth over the protein coding exons (± 10 bp flanking introns) of the longest transcript

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