Supplementary Table 5 Benchmarking subset where we ranked the correct gene as number one out of all candidates in the interval. All entries in this table are correctly predicted genes from the benchmarking set. Note that all files represent diseases/phenotypes even if the title does not reflect this (i.e. MIM 604395 MutL, E. COLI, HOMOLOG). Please inspect the MIM file and/or clinical synopsis for the underlying phenotype. Example in the case of MIM 604395 MutL, E. COLI, HOMOLOG the underlying phenotype is Colorectal cancer which is clearly stated in the subtitle and the text of the file

MIM	Correctly identified disease causing gene		Post. Score	Disease Title
268000	ENSG00000105392		0.1049	266510 REFSUM DISEASE, INFANTILE FORM
144200	ENSG00000171403		0.1087	144200 PALMOPLANTAR KERATODERMA, EPIDERMOLYTIC
171300	ENSG00000117118		0.1109	171300PHEOCHROMOCYTOMA
211980	ENSG00000157764		0.1121	211980 LUNG CANCER ALVEOLAR CELL CARCINOMA, INCLUDED
125270	ENSG00000148218		0.1169	125270 DELTA-AMINOLEVULINATE DEHYDRATASE
235800	ENSG00000084110		0.117	235800 HISTIDINEMIA
278300	ENSG00000158125		0.1176	278300 XANTHINURIA, TYPE I
150800	ENSG00000091483		0.12	150800 LEIOMYOMA, HEREDITARY MULTIPLE, OF SKIN
193510	ENSG00000187098		0.1296	193510 WAARDENBURG SYNDROME, TYPE IIA
102700	ENSG00000196839		0.1322	102700 SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-NEGATIVE, DUE TO ADENOSINE DEAMINASE DEFICIENCY
151670	ENSG00000166035		0.1352	151670 LIPASE, HEPATIC
609227	ENSG00000197535		0.1404	609227 GRISCELLI SYNDROME, TYPE 3
120580	ENSG00000182326		0.1438	120580 COMPLEMENT COMPONENT 1, s SUBCOMPONENT
231200	ENSG00000185245		0.1526	231200 GIANT PLATELET SYNDROME
125851	ENSG00000106633		0.1528	125851 MATURITY-ONSET DIABETES OF THE YOUNG, TYPE II
276710	ENSG00000158104		0.1552	276700 TYROSINEMIA, TYPE I
148210	ENSG00000165474		0.1636	148210 KERATITIS-ICHTHYOSIS-DEAFNESS SYNDROME, AUTOSOMAL DOMINANT
191100	ENSG00000165699		0.1641	191100 TUBEROUS SCLEROSIS
264350	ENSG00000168447		0.1643	264350 PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE
203400	ENSG00000179142		0.1672	203400 ALDOSTERONE DEFICIENCY DUE TO DEFECT IN 18-HYDROXYLASE
607341	ENSG00000165699		0.1716	607341 FOCAL CORTICAL DYSPLASIA OF TAYLOR
176670	ENSG00000160789		0.1725	176670 HUTCHINSON-GILFORD PROGERIA SYNDROME
130070	ENSG00000027847		0.1732	130070 EHLERS-DANLOS SYNDROME, PROGEROID FORM
115310	ENSG00000117118		0.1747	115310 PARAGANGLIOMAS 4
137760	ENSG00000123240		0.1789	137760 GLAUCOMA, PRIMARY OPEN ANGLE, ADULT-ONSET
278400	ENSG00000107165		0.1911	278400 RUFOUS OCULOCUTANEOUS ALBINISM
180800	ENSG00000158887		0.1984	180800 ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA
607736	ENSG00000158887		0.2002	607736 CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J
601240	ENSG00000130005		0.2087	601240 GUANIDINOACETATE METHYLTRANSFERASE
152790	ENSG00000138039		0.2149	152790 LUTEINIZING HORMONE/CHORIOGONADOTROPIN RECEPTOR
202110	ENSG00000148795		0.2197	202110 ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY
605462	ENSG00000185920		0.2211	605462 BASAL CELL CARCINOMA, MULTIPLE
167200	ENSG00000170465		0.2219	167200 PACHYONYCHIA CONGENITA, TYPE 1

142945	ENSG00000164690		0.2256	142945 HOLOPROSENCEPHALY 3
605839	ENSG00000091483		0.229	605839 LEIOMYOMATOSIS AND RENAL CELL CANCER, HEREDITARY
600678	ENSG00000116062		0.245	600678 MutS, E. COLI, HOMOLOG OF, 6
608320	ENSG00000068305		0.2648	608320 CORONARY ARTERY DISEASE, AUTOSOMAL DOMINANT, 1
155255	ENSG00000134982		0.2653	155255 MEDULLOBLASTOMA
256550	ENSG00000184494		0.2668	256550 NEURAMINIDASE DEFICIENCY
103470	ENSG00000187098		0.2677	103470 ALBINISM, OCULAR, WITH SENSORINEURAL DEAFNESS
103500	ENSG00000187098		0.2885	103500 TIETZ SYNDROME
251880	ENSG00000114956		0.2906	51880 MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM
136520	ENSG00000007372		0.3027	36520 FOVEAL HYPOPLASIA AND PRESENILE CATARACT SYNDROME FOVEAL HYPOPLASIA, ISOLATED, INCLUDED
604772	ENSG00000198626		0.3203	04772 VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC
222765	ENSG00000116906		0.3382	22765 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2
276300	ENSG00000122512		0.3416	75355 SQUAMOUS CELL CARCINOMA, HEAD AND NECK
202400	ENSG00000171557		0.3508	202400 AFIBRINOGENEMIA, CONGENITAL HYPOFIBRINOGENEMIA, CONGENITAL, INCLUDED
180849	ENSG00000005339		0.352	180849 RUBINSTEIN-TAYBI SYNDROME
274270	ENSG00000188641		0.3593	274270 DIHYDROPYRIMIDINE DEHYDROGENASE
608328	ENSG00000166147		0.3594	608328 WEILL-MARCHESANI SYNDROME, AUTOSOMAL DOMINANT
600996	ENSG00000198626		0.3677	600996 ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 2
146110	ENSG00000109163		0.3858	146110 HYPOGONADOTROPIC HYPOGONADISM
606762	ENSG00000148672		0.3874	606762 HYPERINSULINISM-HYPERAMMONEMIA SYNDROME
209880	ENSG00000176697		0.4037	209880 AUTONOMIC CONTROL, CONGENITAL FAILURE OF
601399	ENSG00000159216		0.4067	601399 PLATELET DISORDER, FAMILIAL, WITH ASSOCIATED MYELOID MALIGNANCY
176880	ENSG00000184500		0.4108	176880 PROTEIN S, ALPHA
137750	ENSG00000123240		0.4114	137750 GLAUCOMA, PRIMARY OPEN ANGLE, JUVENILE-ONSET, 1
266150	ENSG00000173599		0.4201	266150 PYRUVATE CARBOXYLASE DEFICIENCY
276700	ENSG00000103876		0.4232	276600 TYROSINE TRANSAMINASE DEFICIENCY
271980	ENSG00000112294		0.431	274180 THROMBOXANE A SYNTHASE 1
601110	ENSG00000175142		0.4414	601110 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Id
227810	ENSG00000163581		0.4486	227810 FANCONI-BICKEL SYNDROME
275200	ENSG00000165409		0.449	275200 THYROTROPIN RESISTANCE
603554	ENSG00000166349		0.4588	603554 OMENN SYNDROME
601317	ENSG00000137474		0.4654	601317 DEAFNESS, AUTOSOMAL DOMINANT NONSYNDROMIC SENSORINEURAL 11
124080	ENSG00000179142		0.4727	124080 CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2
203300	ENSG00000107521		0.4783	203300 HERMANSKY-PUDLAK SYNDROME
253010	ENSG00000170266		0.5183	253010 MUCOPOLYSACCHARIDOSIS TYPE IVB
266510	ENSG00000127980		0.5211	266500 REFSUM DISEASE
135290	ENSG00000134982		0.5227	135290 DESMOID DISEASE, HEREDITARY
161400	ENSG00000161610		0.5256	161400 NARCOLEPSY 1
177200	ENSG00000166828		0.5417	177200 LIDDLE SYNDROME
134570	ENSG00000124491		0.5458	134570 FACTOR XIII, A1 SUBUNIT
258870	ENSG00000065154		0.5465	258870 ORNITHINE AMINOTRANSFERASE DEFICIENCY
607855	ENSG00000196569		0.5523	607855 MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A
154700	ENSG00000166147		0.5705	154700 MARFAN SYNDROME
229600	ENSG00000136872		0.5777	229600 FRUCTOSE INTOLERANCE, HEREDITARY
276903	ENSG00000137474		0.5959	276710 TYROSINEMIA, TYPE III

209300	ENSG00000091513		0.6229	209300 ATRANSFERRINEMIA
162500	ENSG00000109099		0.625	162500 NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES
121300	ENSG00000080819		0.6317	121300 COPROPORPHYRIA
214100	ENSG00000124587		0.6622	214100 ZELLWEGER SYNDROME
304800	ENSG00000126895		0.6808	304800 DIABETES INSIPIDUS, NEPHROGENIC, X-LINKED
201470	ENSG00000122971		0.6823	201470 ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF
607624	ENSG00000069974		0.6846	607624 GRISCELLI SYNDROME, TYPE 2; GS2
214150	ENSG00000032514		0.6888	214150 CEREBROOCULOFACIOSKELETAL SYNDROME
309900	ENSG00000010404		0.6964	309900 MUCOPOLYSACCHARIDOSIS TYPE II
607941	ENSG00000136574		0.7151	607941 ATRIAL SEPTAL DEFECT 2
133780	ENSG00000174804		0.7265	133780 EXUDATIVE VITREORETINOPATHY, FAMILIAL, AUTOSOMAL DOMINANT
248600	ENSG00000142046		0.7301	248600 MAPLE SYRUP URINE DISEASE
301500	ENSG00000102393		0.742	301500 FABRY DISEASE
125853	ENSG00000108753		0.7466	125853 DIABETES MELLITUS, NONINSULIN-DEPENDENT
191100	ENSG00000103197		0.75	191100 TUBEROUS SCLEROSIS
600121	ENSG00000018510		0.753	600121 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3
607014	ENSG00000127415		0.7581	607014 HURLER SYNDROME
173900	ENSG00000008710		0.7597	173900 POLYCYSTIC KIDNEYS
212066	ENSG00000168282		0.7597	212066 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIa
129600	ENSG00000166147		0.7622	129600 ECTOPIA LENTIS, ISOLATED
240600	ENSG00000111713		0.7744	240600 GLYCOGEN STORAGE DISEASE 0
202370	ENSG00000127980		0.7788	202370 ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM
103850	ENSG00000149925		0.7878	103850 ALDOLASE A, FRUCTOSE-BISPHOSPHATE
230450	ENSG00000001084		0.7918	230450 GAMMA-GLUTAMYLCYSTEINE SYNTHETASE DEFICIENCY, HEMOLYTIC ANEMIA DUE TO
303100	ENSG00000188419		0.7938	303100 CHOROIDEREMIA
607133	ENSG00000100058		0.7948	607133 CATARACT, SUTURAL, WITH PUNCTATE AND CERULEAN OPACITIES
145900	ENSG00000158887		0.7959	145900 HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS
261550	ENSG00000104899		0.7995	261550 PERSISTENT MULLERIAN DUCT SYNDROME, TYPES I AND II
235200	ENSG00000010704		0.8008	235200 HEMOCHROMATOSIS
168600	ENSG00000185345		0.8071	168600 PARKINSON DISEASE
114480	ENSG00000136492		0.8133	114480 BREAST CANCER
606719	ENSG00000147889		0.8136	606719 MELANOMA-PANCREATIC CANCER SYNDROME
203300	ENSG00000100099		0.8214	203300 HERMANSKY-PUDLAK SYNDROME
143890	ENSG00000130164		0.8222	143890 HYPERCHOLESTEROLEMIA, AUTOSOMAL DOMINANT
609192	ENSG00000106799		0.8224	609192 LOEYS-DIETZ SYNDROME
142623	ENSG00000124205		0.8301	142623 HIRSCHSPRUNG DISEASE
150100	ENSG00000111716		0.8316	150100 LACTATE DEHYDROGENASE-B
209880	ENSG00000124205		0.8548	209880 AUTONOMIC CONTROL, CONGENITAL FAILURE OF
603896	ENSG00000145191		0.8626	603896 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
210900	ENSG00000197299		0.874	210900 BLOOM SYNDROME
176200	ENSG00000143224		0.8862	176200 PORPHYRIA VARIEGATA
207900	ENSG00000169910		0.8895	207900 ARGININOSUCCINIC ACIDURIA
215700	ENSG00000130707		0.8896	215700 CITRULLINEMIA, CLASSIC
253270	ENSG00000159267		0.8896	253270 HOLOCARBOXYLASE SYNTHETASE DEFICIENCY
607791	ENSG00000158887		0.8946	607791 CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D
142900	ENSG00000089225		0.8988	142900 HOLT-ORAM SYNDROME

253000	ENSG00000141012		0.8992	253000 MUCOPOLYSACCHARIDOSIS TYPE IVA
216900	ENSG00000144191		0.9005	216900 ACHROMATOPSIA 2
605373	ENSG00000143252		0.9021	605373 PARAGANGLIOMAS 3
305100	ENSG00000158813		0.9025	305100 ECTODERMAL DYSPLASIA 1, ANHIDROTIC
305900	ENSG00000160211		0.9038	305900 GLUCOSE-6-PHOSPHATE DEHYDROGENASE
253260	ENSG00000169814		0.9062	253260 BIOTINIDASE DEFICIENCY MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET
138040	ENSG00000113580		0.9107	138040 GLUCOCORTICOID RECEPTOR
262190	ENSG00000171105		0.9124	262190 PINEAL HYPERPLASIA, INSULIN-RESISTANT DIABETES MELLITUS, AND SOMATIC ABNORMALITIES
304150	ENSG00000165240		0.9128	304150 CUTIS LAXA, X-LINKED
601386	ENSG00000107736		0.9179	601386 DEAFNESS, AUTOSOMAL RECESSIVE 12
276600	ENSG00000198650		0.9187	276300 TURCOT SYNDROME
125310	ENSG00000074181		0.9224	125310 CEREBRAL ARTERIOPATHY, AUTOSOMAL DOMINANT, WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY
252011	ENSG00000073578		0.9326	252011 MITOCHONDRIAL COMPLEX II DEFICIENCY
276300	ENSG00000076242		0.9355	276300 TURCOT SYNDROME
173900	ENSG00000118762		0.9356	173900 POLYCYSTIC KIDNEYS
233690	ENSG00000051523		0.9357	233690 GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-NEGATIVE
256540	ENSG00000064601		0.9363	256540 NEURAMINIDASE DEFICIENCY WITH BETA-GALACTOSIDASE DEFICIENCY
250940	ENSG00000116984		0.9369	250940 METHYLCOBALAMIN DEFICIENCY, cblG TYPE
603909	ENSG00000003400		0.9369	603909 AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE IIA
230500	ENSG00000170266		0.9399	230500 GANGLIOSIDOSIS, GENERALIZED GM1, TYPE I
203300	ENSG00000110756		0.9414	203300 HERMANSKY-PUDLAK SYNDROME
262300	ENSG00000170289		0.9424	262300 ACHROMATOPSIA 3
130060	ENSG00000164692		0.9441	130060 EHLERS-DANLOS SYNDROME, TYPE VII, AUTOSOMAL DOMINANT
201910	ENSG00000168482		0.9465	201910 ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY
222748	ENSG00000147647		0.9502	222748 DIHYDROPYRIMIDINASE
118220	ENSG00000109099		0.9505	118220 CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A
601544	ENSG00000121742		0.9512	601544 DEAFNESS, AUTOSOMAL DOMINANT NONSYNDROMIC SENSORINEURAL 3
188050	ENSG00000122194		0.956	188050 THROMBOPHILIA VENOUS THROMBOEMBOLISM, INCLUDED
102610	ENSG00000143632		0.9577	102610 ACTIN, ALPHA, SKELETAL MUSCLE 1
603041	ENSG00000025708		0.9592	603041 MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME
264470	ENSG00000161533		0.961	264470 PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY
601547	ENSG00000100058		0.9642	601547 CATARACT, CONGENITAL, CERULEAN TYPE, 2
601067	ENSG00000107736		0.9646	601067 USHER SYNDROME, TYPE ID
600060	ENSG00000137474		0.9671	600060 DEAFNESS, NEUROSENSORY, AUTOSOMAL RECESSIVE 2
605253	ENSG00000158887		0.9676	605253 NEUROPATHY, CONGENITAL HYPOMYELINATING
130060	ENSG00000108821		0.9717	130060 EHLERS-DANLOS SYNDROME, TYPE VII, AUTOSOMAL DOMINANT
109400	ENSG00000185920		0.9747	109400 BASAL CELL NEVUS SYNDROME
601265	ENSG00000156574		0.9758	601265 NODAL, MOUSE, HOMOLOG OF
231900	ENSG00000100983		0.9771	231900 GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES, HEMOLYTIC ANEMIA
277900	ENSG00000123191		0.9782	277900 WILSON DISEASE
230600	ENSG00000170266		0.9784	230600 GANGLIOSIDOSIS, GENERALIZED GM1, TYPE II, OR JUVENILE TYPE
125853	ENSG00000154415		0.9804	125853 DIABETES MELLITUS, NONINSULIN-DEPENDENT

600258	ENSG00000064933		0.9809	600258 POSTMEIOTIC SEGREGATION INCREASED, S. CEREVISIAE, 1
609015	ENSG00000084754		0.9811	09015 TRIFUNCTIONAL PROTEIN DEFICIENCY
142680	ENSG00000067182		0.9866	142680 PERIODIC FEVER, FAMILIAL, AUTOSOMAL DOMINANT
309400	ENSG00000165240		0.9867	309400 MENKES DISEASE
230650	ENSG00000170266		0.9874	230650 GANGLIOSIDOSIS, GENERALIZED GM1, TYPE III, OR ADULT TYPE
107741	ENSG00000130203		0.9901	107741 APOLIPOPROTEIN E
236250	ENSG00000177000		0.9904	236250 HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY
166220	ENSG00000164692		0.991	166220 OSTEOGENESIS IMPERFECTA, TYPE IV
608810	ENSG00000109846		0.9913	608810 ALPHA-B CRYSTALLINOPATHY
188055	ENSG00000198734		0.9923	188055 THROMBOPHILIA DUE TO DEFICIENCY OF ACTIVATED PROTEIN C COFACTOR
602730	ENSG00000114739		0.9924	602730 ACTIVIN A RECEPTOR, TYPE IIB
188040	ENSG00000178726		0.9926	188040 THROMBOMODULIN
605407	ENSG00000180176		0.9933	605407 SEGAWA SYNDROME, AUTOSOMAL RECESSIVE
603147	ENSG00000088035		0.9938	603147 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ic
300100	ENSG00000101986		0.9938	300100 ADRENOLEUKODYSTROPHY
600501	ENSG00000136160		0.9945	600501 ABCD SYNDROME
155601	ENSG00000147889		0.9946	155601 MELANOMA, CUTANEOUS MALIGNANT, 2
118200	ENSG00000158887		0.9946	118200 CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B
180071	ENSG00000132915		0.9946	180071 PHOSPHODIESTERASE 6A, cGMP-SPECIFIC, ROD, ALPHA
176860	ENSG00000115718		0.9951	176860 PROTEIN C DEFICIENCY, CONGENITAL THROMBOTIC DISEASE DUE TO
264350	ENSG00000111319		0.9954	264350 PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE
170995	ENSG00000117528		0.9965	170995 ATP-BINDING CASSETTE, SUBFAMILY D, MEMBER 3
603073	ENSG00000043355		0.9969	603073 ZINC FINGER PROTEIN OF CEREBELLUM, 2
261630	ENSG00000151552		0.997	261630 PHENYLKETONURIA II
277580	ENSG00000136160		0.9972	277580 WAARDENBURG-SHAH SYNDROME
607785	ENSG00000168638		0.998	607785 JUVENILE MYELOMONOCYTIC LEUKEMIA
250950	ENSG00000148090		0.9981	250950 3-@METHYLGLUTACONIC ACIDURIA, TYPE I
166200	ENSG00000164692		0.9982	166200 OSTEOGENESIS IMPERFECTA, TYPE I
607785	ENSG00000196712		0.9983	607785 JUVENILE MYELOMONOCYTIC LEUKEMIA
259420	ENSG00000164692		0.9983	259420 OSTEOGENESIS IMPERFECTA, PROGRESSIVELY DEFORMING, WITH NORMAL SCLERAE
306000	ENSG00000044446		0.9985	306000 GLYCOGEN STORAGE DISEASE VIII
277580	ENSG00000124205		0.9985	277580 WAARDENBURG-SHAH SYNDROME
166210	ENSG00000108821		0.9988	166210 OSTEOGENESIS IMPERFECTA CONGENITA
166220	ENSG00000108821		0.9988	166220 OSTEOGENESIS IMPERFECTA, TYPE IV
300300	ENSG00000010671		0.9989	300300 BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE
608099	ENSG00000108823		0.999	608099 MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2D
207800	ENSG00000118520		0.9992	207800 ARGININEMIA
121050	ENSG00000138829		0.9992	121050 CONTRACTURAL ARACHNODACTYLY, CONGENITAL
276300	ENSG00000134982		0.9992	276300 TURCOT SYNDROME
606054	ENSG00000175198		0.9992	606054 PROPIONIC ACIDEMIA
125700	ENSG00000101200		0.9993	125700 DIABETES INSIPIDUS, NEUROHYPOPHYSEAL TYPE
166200	ENSG00000108821		0.9993	166200 OSTEOGENESIS IMPERFECTA, TYPE I
261600	ENSG00000171759		0.9993	261600 PHENYLKETONURIA
177820	ENSG00000185245		0.9994	177820 PSEUDO-VON WILLEBRAND DISEASE
600155	ENSG00000136160		0.9995	600155 HIRSCHSPRUNG DISEASE 2

236200	ENSG00000160200		0.9995	236200 HOMOCYSTINURIA
237300	ENSG00000021826		0.9997	237300 CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO
604307	ENSG00000100058		0.9997	604307 CATARACT, COPPOCK-LIKE
166210	ENSG00000164692		0.9997	166210 OSTEOGENESIS IMPERFECTA CONGENITA
608799	ENSG00000000419		0.9998	608799 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ie
259420	ENSG00000108821		0.9998	259420 OSTEOGENESIS IMPERFECTA, PROGRESSIVELY DEFORMING, WITH NORMAL SCLERAE
233700	ENSG00000158517		0.9998	233700 GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-POSITIVE, TYPE I
607136	ENSG00000112592		0.9998	607136 SPINOCEREBELLAR ATAXIA 17
120435	ENSG00000095002		0.9999	120435 COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1
604395	ENSG00000119684		0.9999	604395 MutL, E. COLI, HOMOLOG OF, 3
214100	ENSG00000157911		0.9999	214100 ZELLWEGER SYNDROME
306900	ENSG00000101981		0.9999	306900 HEMOPHILIA B
300514	ENSG00000139618		0.9999	300514 FANCONI ANEMIA, COMPLEMENTATION GROUP B
238970	ENSG00000102743		0.9999	238970 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME
602092	ENSG00000006611		0.9999	602092 DEAFNESS, NEUROSENSORY, AUTOSOMAL RECESSIVE 18
227650	ENSG00000158169		0.9999	227650 FANCONI ANEMIA
306700	ENSG00000185010		0.9999	306700 HEMOPHILIA A
158320	ENSG00000095002		0.9999	158320 MUIR-TORRE SYNDROME
607822	ENSG00000080815		0.9999	607822 ALZHEIMER DISEASE, FAMILIAL, TYPE 3
118300	ENSG00000109099		0.9999	118300 CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS
609227	ENSG00000115648		1	609227 GRISCELLI SYNDROME, TYPE 3
608089	ENSG00000095002		1	608089 ENDOMETRIAL CANCER
202200	ENSG00000185231		1	202200 GLUCOCORTICOID DEFICIENCY 1
601665	ENSG00000166603		1	601665 OBESITY LEANNESS, INCLUDED
201450	ENSG00000117054		1	201450 ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF
276904	ENSG00000006611		1	276904 USHER SYNDROME, TYPE IC
312170	ENSG00000131828		1	312170 PYRUVATE DECARBOXYLASE DEFICIENCY
145900	ENSG00000109099		1	145900 HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS
232800	ENSG00000152556		1	232800 GLYCOGEN STORAGE DISEASE VII
302045	ENSG00000198947		1	302045 CARDIOMYOPATHY, DILATED, X-LINKED
278800	ENSG00000032514		1	278800 DE SANCTIS-CACCHIONE SYNDROME
125853	ENSG00000171105		1	125853 DIABETES MELLITUS, NONINSULIN-DEPENDENT
300400	ENSG00000147168		1	300400 SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED
233710	ENSG00000116701		1	233710 GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-POSITIVE, TYPE II
193300	ENSG00000134086		1	193300 VON HIPPEL-LINDAU SYNDROME
600119	ENSG00000108823		1	600119 SARCOGLYCAN, ALPHA
603896	ENSG00000111361		1	603896 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
232700	ENSG00000100504		1	232700 GLYCOGEN STORAGE DISEASE VI
601224	ENSG00000151348		1	601224 POTOCKI-SHAFFER SYNDROME
603360	ENSG00000121680		1	603360 PEROXISOME BIOGENESIS FACTOR 16
604286	ENSG00000163069		1	604286 MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2E
600259	ENSG00000122512		1	600259 POSTMEIOTIC SEGREGATION INCREASED, S. CEREVISIAE, 2
300376	ENSG00000198947		1	300376 MUSCULAR DYSTROPHY, BECKER TYPE
215300	ENSG00000182197		1	215300 CHONDROSARCOMA
175100	ENSG00000134982		1	175100 ADENOMATOUS POLYPOSIS OF THE COLON

253700	ENSG00000102683		1	253700 MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2C
245349	ENSG00000110435		1	245349 PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY
246450	ENSG00000117305		1	246450 3-@HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY
306400	ENSG00000165168		1	306400 GRANULOMATOUS DISEASE, CHRONIC, X-LINKED
260500	ENSG00000141510		1	260500 PAPILLOMA OF CHOROID PLEXUS
151623	ENSG00000147889		1	151623 LI-FRAUMENI SYNDROME 1
137150	ENSG00000183044		1	137150 4-@AMINOBUTYRATE AMINOTRANSFERASE
277450	ENSG00000115486		1	277450 VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 1
608089	ENSG00000113318		1	608089 ENDOMETRIAL CANCER
214100	ENSG00000139197		1	214100 ZELLWEGER SYNDROME
248610	ENSG00000137992		1	248610 DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE
132600	ENSG00000168036		1	132600 PILOMATRIXOMA
604391	ENSG00000020922		1	604391 ATAXIA-TELANGIECTASIA-LIKE DISORDER; ATLD
203750	ENSG00000075239		1	203750 ALPHA-METHYLACETOACETIC ACIDURIA
214100	ENSG00000108733		1	214100 ZELLWEGER SYNDROME
311250	ENSG00000036473		1	311250 ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO
278760	ENSG00000175595		1	278760 XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F
214100	ENSG00000034693		1	214100 ZELLWEGER SYNDROME
231200	ENSG00000169704		1	231200 GIANT PLATELET SYNDROME
158320	ENSG00000076242		1	158320 MUIR-TORRE SYNDROME
133700	ENSG00000182197		1	133700 EXOSTOSES, MULTIPLE, TYPE I
216400	ENSG00000134899		1	216400 COCKAYNE SYNDROME, TYPE A
608089	ENSG00000076242		1	608089 ENDOMETRIAL CANCER
133701	ENSG00000151348		1	133701 EXOSTOSES, MULTIPLE, TYPE II
201460	ENSG00000115361		1	201460 ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF
133510	ENSG00000163161		1	133510 EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 3
601366	ENSG00000175387		1	601366 MOTHERS AGAINST DECAPENTAPLEGIC, DROSOPHILA, HOMOLOG OF, 2
227650	ENSG00000183161		1	227650 FANCONI ANEMIA
227650	ENSG00000165281		1	227650 FANCONI ANEMIA
278740	ENSG00000134574		1	278740 XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP E
601789	ENSG00000162928		1	601789 PEROXISOME BIOGENESIS FACTOR 13
310200	ENSG00000198947		1	310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE
176930	ENSG00000180210		1	176930 COAGULATION FACTOR II
172490	ENSG00000102893		1	172490 PHOSPHORYLASE KINASE, BETA SUBUNIT
202370	ENSG00000162928		1	202370 ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM
120436	ENSG00000076242		1	120436 MutL, E. COLI, HOMOLOG OF, 1
151623	ENSG00000141510		1	151623 LI-FRAUMENI SYNDROME 1
208900	ENSG00000149311		1	208900 ATAXIA-TELANGIECTASIA
227650	ENSG00000112039		1	227650 FANCONI ANEMIA
227650	ENSG00000187741		1	227650 FANCONI ANEMIA
278720	ENSG00000154767		1	278720 XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP C
601675	ENSG00000163161		1	601675 TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; TTDP