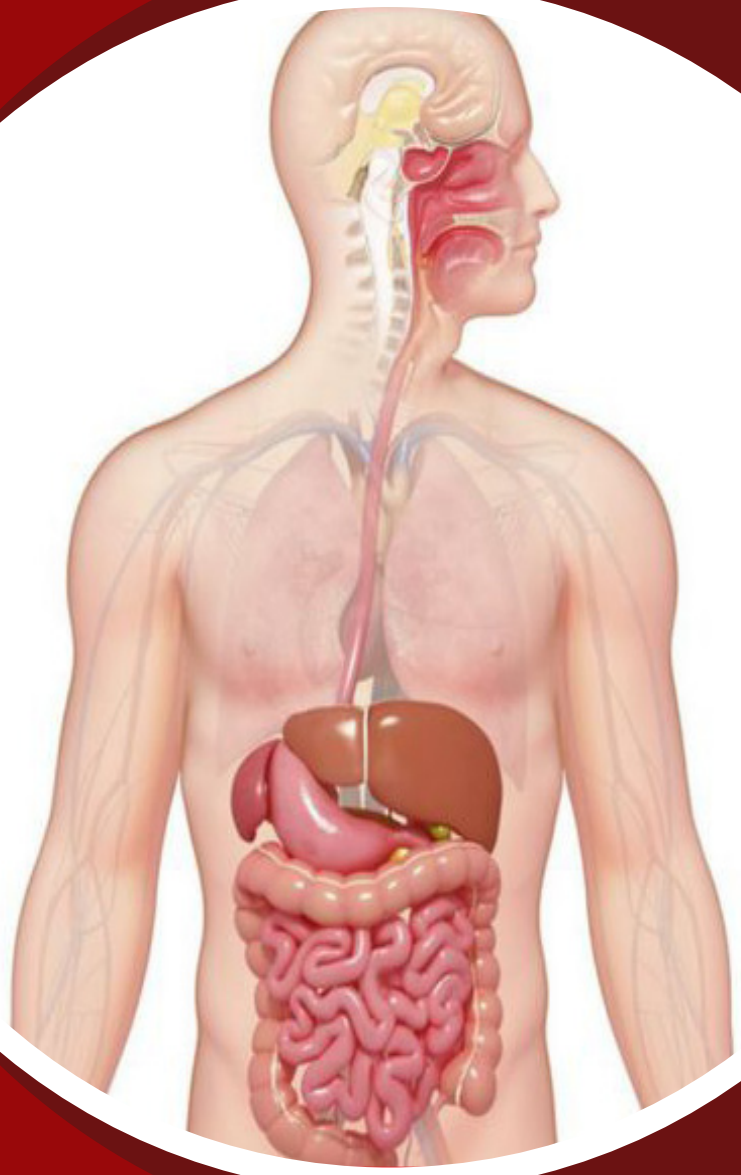

Gastrointestinal Tract

GIT

Gastrointestinal Tract (GIT)

Single Gene

Code	Diseases	Gene
sCe1/00001	17-hydroxylation activity deficiency	CYP17A1
sCe1/00002	2-aminoadipic 2-oxoadipic aciduria	DHTKD1
sCe1/00003	2-methylbutyrylglycinuria	ACADSB
sCe1/00004	3-beta-hydroxysteroid dehydrogenase deficiency type 2	HSD3B2
sCe1/00005	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL
sCe1/00006	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	HMGCS2
sCe1/00007	3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH
sCe1/00008	3-methylglutaconic aciduria type 1	AUH
sCe1/00009	3-methylglutaconic aciduria type 3	OPA3
sCe1/00010	3-methylglutaconic aciduria type 5	DNAJC19
sCe1/00011	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	SERAC1
sCe1/00012	5-oxoprolinase deficiency	OPLAH
sCe1/00013	6q24-related transient neonatal diabetes mellitus type 1	UPD chr. 6
sCe1/00014	Abetalipoproteinemia	MTTP
sCe1/00015	Acetycholinesterase deficiency	ACHE
sCe1/00016	Acetyl-CoA carboxylase deficiency	ACACA
sCe1/00017	Acyl-CoA medium-chain dehydrogenase deficiency	ACADM
sCe1/00018	Acyl-CoA multiple dehydrogenase deficiency	ETFA
sCe1/00019	Acyl-CoA multiple dehydrogenase deficiency	ETFB
sCe1/00020	Acyl-CoA short-chain dehydrogenase deficiency	ACADS
sCe1/00021	Acyl-CoA very long-chain dehydrogenase deficiency	ACADVL
sCe1/00022	Adenine phosphoribosyltransferase deficiency	APRT
sCe1/00023	Adenylosuccinase deficiency	ADSL
sCe1/00024	Adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2
sCe1/00025	Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	POR

sCe1/00026	Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency	CYP11B1
sCe1/00027	Adrenal hypoplasia	NR0B1
sCe1/00028	Alkaptonuria	HGD
sCe1/00029	Alpha-2-macroglobulin deficiency	A2M
sCe1/00030	Alpha-ketoglutarate dehydrogenase deficiency	OGDH
sCe1/00031	Alpha-methylacyl CoA racemase deficiency	AMACR
sCe1/00032	Aminoacylase deficiency	ACY1
sCe1/00033	AMP deaminase deficiency, erythrocytic	AMPD3
sCe1/00034	Amyloidosis, familial visceral	APOA1
sCe1/00035	Andersen disease	GBE1
sCe1/00036	Anemia dyserythropoietic type 1A	CDAN1
sCe1/00037	Anemia dyserythropoietic type 2	SEC23B
sCe1/00038	Antitrypsin-alpha-1 deficiency	SERPINA1
sCe1/00039	Aplastic anemia	PRF1
sCe1/00040	Aplastic anemia	TERC
sCe1/00041	Aplastic anemia, SBDS related	SBDS
sCe1/00042	Apolipoprotein C-II deficiency	APOC2
sCe1/00043	Apparent mineralocorticoid excess	HSD11B2
sCe1/00044	Arginase deficiency	ARG1
sCe1/00045	Arginine-glycine amidinotransferase deficiency	GATM
sCe1/00046	Argininosuccinic aciduria	ASL
sCe1/00047	Aromatic L-amino acid decarboxylase deficiency	DDC
sCe1/00048	Asparaginesynthetase deficiency	ASNS
sCe1/00049	Aspartylglucosaminuria	AGA
sCe1/00050	Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency	ST6GAL2
sCe1/00051	Beta-ureidopropionase deficiency	UPB1
sCe1/00052	Bile acid malabsorption, primary	SLC10A2
sCe1/00053	Bile acid synthesis defect type 2, congenital	AKR1D1
sCe1/00054	Bile acid synthesis defect type 3, congenital	CYP7B1

sCe1/00055	Bile acid synthesis defect type 4, congenital	AMACR
sCe1/00056	Biotinidase deficiency	BTD
sCe1/00057	Bloom syndrome	BLM
sCe1/00058	Branched-chain aminotransferase 1 deficiency	BCAT1
sCe1/00059	Branched-chain aminotransferase 2 deficiency	BCAT2
sCe1/00060	Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK
sCe1/00061	Bronchiectasis with or without elevated sweat chloride type 2	SCNN1A
sCe1/00062	Butyrylcholinesterase deficiency	BCHE
sCe1/00063	Carbamoylphosphate synthetase I deficiency	CPS1
sCe1/00064	Carnitine deficiency	SLC22A5
sCe1/00065	Carnitine palmitoyltransferase 1A deficiency	CPT1A
sCe1/00066	Carnitine palmitoyltransferase 1B deficiency	CPT1B
sCe1/00067	Carnitine palmitoyltransferase 2 deficiency, infantile	CPT2
sCe1/00068	Carnitine palmitoyltransferase 2 deficiency, lethal neonatal	CPT2
sCe1/00069	Carnitine-acylcarnitine translocase deficiency	SLC25A20
sCe1/00070	Catechol-o-methyltransferase deficiency	COMT
sCe1/00071	Ceroid lipofuscinosis neuronal type 1	PPT1
sCe1/00072	Ceroid lipofuscinosis neuronal type 10	CTSD
sCe1/00073	Ceroid lipofuscinosis neuronal type 11	GRN
sCe1/00074	Ceroid lipofuscinosis neuronal type 2	TPP1
sCe1/00075	Ceroid lipofuscinosis neuronal type 3	CLN3
sCe1/00076	Ceroid lipofuscinosis neuronal type 4	DNAJC5
sCe1/00077	Ceroid lipofuscinosis neuronal type 5	CLN5
sCe1/00078	Ceroid lipofuscinosis neuronal type 6	CLN6
sCe1/00079	Ceroid lipofuscinosis neuronal type 7	MFSD8
sCe1/00080	Ceroid lipofuscinosis neuronal type 8	CLN8
sCe1/00081	Chanarin-Dorfman syndrome	ABHD5
sCe1/00082	Chloramphenicol resistance, MT-RNR2 related	MT-RNR2
sCe1/00083	Cholestasis benign recurrent intrahepatic type 2	ABCB11

sCe1/00084	Cholestasis intrahepatic, of pregnancy, type 3	ABCB4
sCe1/00085	Cholestasis progressive intrahepatic type 1	ATP8B1
sCe1/00086	Cholestasis progressive intrahepatic type 2	ABCB11
sCe1/00087	Cholestasis progressive intrahepatic type 3	ABCB4
sCe1/00088	Cholestasis, benign recurrent intrahepatic	ATP8B1
sCe1/00089	Cholestasis, intrahepatic, of pregnancy, type 1	ATP8B1
sCe1/00090	Cholesteryl ester storage disease	LIPA
sCe1/00091	Chylomicron retention disease	SAR1B
sCe1/00092	Citrin deficiency	SLC25A13
sCe1/00093	Citrullinemia	ASS1
sCe1/00094	CoA-2 4-dienoyl reductase 1 deficiency	DECR1
sCe1/00095	CoA-3-hydroxyacyl dehydrogenase deficiency	HADH
sCe1/00096	CoA-3-methylcrontonyl carboxylase 1 deficiency	MCCC1
sCe1/00097	CoA-3-methylcrontonyl carboxylase 2 deficiency	MCCC2
sCe1/00098	Colchicine resistance	ABCB1
sCe1/00099	Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1
sCe1/00100	Combined malonic and methylmalonic aciduria	ACSF3
sCe1/00101	Combined oxidative phosphorylation deficiency type 1	GFM1
sCe1/00102	Combined oxidative phosphorylation deficiency type 10	MTO1
sCe1/00103	Combined oxidative phosphorylation deficiency type 11	RMND1
sCe1/00104	Combined oxidative phosphorylation deficiency type 12	EARS2
sCe1/00105	Combined oxidative phosphorylation deficiency type 13	PNPT1
sCe1/00106	Combined oxidative phosphorylation deficiency type 14	FARS2
sCe1/00107	Combined oxidative phosphorylation deficiency type 15	MTFMT
sCe1/00108	Combined oxidative phosphorylation deficiency type 16	MRPL44
sCe1/00109	Combined oxidative phosphorylation deficiency type 17	ELAC2
sCe1/00110	Combined oxidative phosphorylation deficiency type 18	SFXN4
sCe1/00111	Combined oxidative phosphorylation deficiency type 19	LYRM4
sCe1/00112	Combined oxidative phosphorylation deficiency type 2	MRPS16

sCe1/00113	Combined oxidative phosphorylation deficiency type 20	VARS2
sCe1/00114	Combined oxidative phosphorylation deficiency type 21	TARS2
sCe1/00115	Combined oxidative phosphorylation deficiency type 22	ATP5A1
sCe1/00116	Combined oxidative phosphorylation deficiency type 23	GTPBP3
sCe1/00117	Combined oxidative phosphorylation deficiency type 24	NARS2
sCe1/00118	Combined oxidative phosphorylation deficiency type 25	MARS2
sCe1/00119	Combined oxidative phosphorylation deficiency type 26	TRMT5
sCe1/00120	Combined oxidative phosphorylation deficiency type 3	TSFM
sCe1/00121	Combined oxidative phosphorylation deficiency type 4	TUFM
sCe1/00122	Combined oxidative phosphorylation deficiency type 5	MRPS22
sCe1/00123	Combined oxidative phosphorylation deficiency type 6	AIFM1
sCe1/00124	Combined oxidative phosphorylation deficiency type 7	C12ORF65
sCe1/00125	Combined oxidative phosphorylation deficiency type 8	AARS2
sCe1/00126	Combined oxidative phosphorylation deficiency type 9	MRPL3
sCe1/00127	Congenital disorder of glycosylation, type Ip	ALG11
sCe1/00128	Congenital disorder of glycosylation, type Iq	SRD5A3
sCe1/00129	Congenital disorder of glycosylation, type Iw	STT3A
sCe1/00130	Coproporphyria	CPOX
sCe1/00131	Coumarin resistance	VKORC1
sCe1/00132	Coumarin/Warfarin resistance due to CYP2C9 variants	CYP2C9
sCe1/00133	Creatine deficiency syndrome X-linked	SLC6A8
sCe1/00134	CYP2C19 related poor drug metabolism	CYP2C19
sCe1/00135	Cystathioninuria	CTH
sCe1/00136	Cystic fibrosis	CFTR
sCe1/00137	Cystic fibrosis, SLC6A14 related	SLC6A14
sCe1/00138	Cystinosis, nephropathic	CTNS
sCe1/00139	Cytochrome P450 deficiency	CYP1A2
sCe1/00140	D-2-hydroxyglutaric aciduria type 1	D2HGDH
sCe1/00141	D-2-hydroxyglutaric aciduria type 2	IDH2

sCe1/00142	D-bifunctional protein deficiency	HSD17B4
sCe1/00143	D-glyceric aciduria	GLYCTK
sCe1/00144	Diabetes insipidus, nephrogenic, X-linked	AVPR2
sCe1/00145	Diabetes insipidus, neurohypophyseal	AVP
sCe1/00146	Diabetes mellitus type 1	INS
sCe1/00147	Diabetes mellitus, insulin-dependent type 20	HNF1A
sCe1/00148	Diabetes mellitus, insulin-resistant with acanthosis nigricans	INSR
sCe1/00149	Diabetes mellitus, neonatal	GLIS3
sCe1/00150	Diabetes mellitus, noninsulin-dependent	AKT2
sCe1/00151	Diabetes mellitus, noninsulin-dependent	KCNJ11
sCe1/00152	Diabetes mellitus, noninsulin-dependent	ABCC8
sCe1/00153	Diabetes mellitus, permanent neonatal	ABCC8
sCe1/00154	Diabetes mellitus, transient neonatal type 2	ABCC8
sCe1/00155	Diabetes, IGF2 related	IGF2
sCe1/00156	Diarrhea type 1, secretory chloride, congenital	SLC26A3
sCe1/00157	Diarrhea type 4, malabsorptive, congenital	NEUROG3
sCe1/00158	Dihydropyrimidine dehydrogenase deficiency	DPYD
sCe1/00159	Dihydropyrimidinuria	DPYS
sCe1/00160	Dimethylglycine dehydrogenase deficiency	DMGDH
sCe1/00161	Dyggve-Melchior-Clausen disease	DYM
sCe1/00162	Efavirenz, poor metabolism of	CYP2B6
sCe1/00163	Enterokinase deficiency	TMPRSS15
sCe1/00164	Erythrocyte lactate transporter defect	SLC16A1
sCe1/00165	Fabry disease	GLA
sCe1/00166	Factor II deficiency	F2
sCe1/00167	Factor V deficiency	F5
sCe1/00168	Factor XIIIB deficiency	F13B
sCe1/00169	Fanconi anemia type A	FANCA
sCe1/00170	Fanconi anemia type B	FANCB

sCe1/00171	Fanconi anemia type C	FANCC
sCe1/00172	Fanconi anemia type D1	BRCA2
sCe1/00173	Fanconi anemia type D2	FANCD2
sCe1/00174	Fanconi anemia type E	FANCE
sCe1/00175	Fanconi anemia type F	FANCF
sCe1/00176	Fanconi anemia type G	FANCG
sCe1/00177	Fanconi anemia type I	FANCI
sCe1/00178	Fanconi anemia type J	BRIP1
sCe1/00179	Fanconi anemia type L	FANCL
sCe1/00180	Fanconi anemia type M	FANCM
sCe1/00181	Fanconi anemia type N	PALB2
sCe1/00182	Fanconi anemia type P	SLX4
sCe1/00183	Fanconi anemia, XRCCR2 related	XRCC2
sCe1/00184	Fanconi-Bickel syndrome	SLC2A2
sCe1/00185	Farber disease	ASAH1
sCe1/00186	Favism, susceptibility to	G6PD
sCe1/00187	Fish eye disease	LCAT
sCe1/00188	Folate malabsorption, hereditary	SLC46A1
sCe1/00189	Fructose intolerance	ALDOB
sCe1/00190	Fructose uptake deficiency, SLC2A5 related	SLC2A5
sCe1/00191	Fructose-1,6-bisphosphatase deficiency	FBP1
sCe1/00192	Fructosuria essential	KHK
sCe1/00193	Fucosidosis	FUCA1
sCe1/00194	Fumarase deficiency	FH
sCe1/00195	GABA-transaminase deficiency	ABAT
sCe1/00196	Galactokinase deficiency	GALK1
sCe1/00197	Galactose epimerase deficiency	GALE
sCe1/00198	Galactosemia	GALT
sCe1/00199	Galactosialidosis	CTSA

sCe1/00200	Gallbladder disease type 1	ABCB4
sCe1/00201	Gaucher disease type 1	GBA
sCe1/00202	Gaucher disease type 2	GBA
sCe1/00203	Gaucher disease type 3	GBA
sCe1/00204	Gaucher disease type 3C	GBA
sCe1/00205	Gaucher disease, atypical	PSAP
sCe1/00206	Gaucher disease, perinatal lethal	GBA
sCe1/00207	Glucocorticoid deficiency type 1	MC2R
sCe1/00208	Glucocorticoid deficiency type 2	MRAP
sCe1/00209	Glucose/Galactose malabsorption	SLC5A1
sCe1/00210	Glutamate formiminotransferase deficiency	FTCD
sCe1/00211	Glutamine deficiency, congenital	GLUL
sCe1/00212	Glutaric acidemia type 1	GCDH
sCe1/00213	Glutaric acidemia type 2C	ETFDH
sCe1/00214	Glutaric aciduria type 3	SUGCT
sCe1/00215	Glutathione S-transferase theta-1 defficiency	GSTT1
sCe1/00216	Glutathione synthetase deficiency	GSS
sCe1/00217	Glycerol kinase deficiency	GK
sCe1/00218	Glycogen storage disease of heart (lethal)	PRKAG2
sCe1/00219	Glycogen storage disease type 0	GYS2
sCe1/00220	Glycogen storage disease type 0 muscle	GYS1
sCe1/00221	Glycogen storage disease type 10	PGAM2
sCe1/00222	Glycogen storage disease type 11	LDHA
sCe1/00223	Glycogen storage disease type 12	ALDOA
sCe1/00224	Glycogen storage disease type 13	ENO3
sCe1/00225	Glycogen storage disease type 14	PGM1
sCe1/00226	Glycogen storage disease type 15	GYG1
sCe1/00227	Glycogen storage disease type 1A	G6PC
sCe1/00228	Glycogen storage disease type 1B	SLC37A4

sCe1/00229	Glycogen storage disease type 1C	SLC37A4
sCe1/00230	Glycogen storage disease type 2	GAA
sCe1/00231	Glycogen storage disease type 3	AGL
sCe1/00232	Glycogen storage disease type 4	GBE1
sCe1/00233	Glycogen storage disease type 5	PYGM
sCe1/00234	Glycogen storage disease type 6B	PYGL
sCe1/00235	Glycogen storage disease type 7	PFKM
sCe1/00236	Glycogen storage disease type 9A	PHKA2
sCe1/00237	Glycogen storage disease type 9B	PHKB
sCe1/00238	Glycogen storage disease type 9C	PHKG2
sCe1/00239	Glycosylation disorder type 1A	PMM2
sCe1/00240	Glycosylation disorder type 1B	MPI
sCe1/00241	Glycosylation disorder type 1C	ALG6
sCe1/00242	Glycosylation disorder type 1D	ALG3
sCe1/00243	Glycosylation disorder type 1E	DPM1
sCe1/00244	Glycosylation disorder type 1F	MPDU1
sCe1/00245	Glycosylation disorder type 1G	ALG12
sCe1/00246	Glycosylation disorder type 1H	ALG8
sCe1/00247	Glycosylation disorder type 1I	ALG2
sCe1/00248	Glycosylation disorder type 1J	DPAGT1
sCe1/00249	Glycosylation disorder type 1K	ALG1
sCe1/00250	Glycosylation disorder type 1L	ALG9
sCe1/00251	Glycosylation disorder type 1M	DOLK
sCe1/00252	Glycosylation disorder type 1N	RFT1
sCe1/00253	Glycosylation disorder type 1O	DPM3
sCe1/00254	Glycosylation disorder type 1S	ALG13
sCe1/00255	Glycosylation disorder type 1U	DPM2
sCe1/00256	Glycosylation disorder type 2A	MGAT2
sCe1/00257	Glycosylation disorder type 2B	MOGS

sCe1/00258	Glycosylation disorder type 2C	SLC35C1
sCe1/00259	Glycosylation disorder type 2D	B4GALT1
sCe1/00260	Glycosylation disorder type 2E	COG7
sCe1/00261	Glycosylation disorder type 2F	SLC35A1
sCe1/00262	Glycosylation disorder type 2G	COG1
sCe1/00263	Glycosylation disorder type 2H	COG8
sCe1/00264	Glycosylation disorder type 2I	COG5
sCe1/00265	Glycosylation disorder type 2J	COG4
sCe1/00266	Glycosylation disorder type 2K	TMEM165
sCe1/00267	Glycosylation disorder type 2M	SLC35A2
sCe1/00268	Glycosylation disorder type 3	COG6
sCe1/00269	Glycosylation disorder type IR	DDOST
sCe1/00270	Glycosylation disorder x-linked	SSR4
sCe1/00271	GM1-gangliosidosis type 1	GLB1
sCe1/00272	GM1-gangliosidosis type 2	GLB1
sCe1/00273	GM2-gangliosidosis type 2	HEXB
sCe1/00274	Guanidinoacetate methyltransferase deficiency	GAMT
sCe1/00275	Hartnup disorder	SLC6A19
sCe1/00276	Hawkinsinuria	HPD
sCe1/00277	HDL deficiency, type 2	ABCA1
sCe1/00278	Hemochromatosis classical	HFE
sCe1/00279	Hemochromatosis type 2A	HFE2
sCe1/00280	Hemochromatosis type 2B	HAMP
sCe1/00281	Hemochromatosis type 3	TFR2
sCe1/00282	Hemochromatosis type 4	SLC40A1
sCe1/00283	Hemolytic anemia due to G6PD deficiency	G6PD
sCe1/00284	Hemophilia A	F8
sCe1/00285	Holocarboxylase synthetase deficiency	HLCS
sCe1/00286	Hurler syndrome	IDUA

sCe1/00287	Hurler-Scheie syndrome	IDUA
sCe1/00288	Hypercalcemia infantile type	CYP24A1
sCe1/00289	Hypercholanemia	BAAT
sCe1/00290	Hypercholanemia	TJP2
sCe1/00291	Hypercholesterolemia autosomal dominant type 3	PCSK9
sCe1/00292	Hypercholesterolemia autosomal recessive	LDLRAP1
sCe1/00293	Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant	LDLR
sCe1/00294	Hypercholesterolemia type B autosomanl dominant	APOB
sCe1/00295	Hyperchylomicronemia type 5	APOA5
sCe1/00296	Hyperinsulinaemia, association with, G6PC2 related	G6PC2
sCe1/00297	Hyperinsulinemic hypoglycemia type 1	ABCC8
sCe1/00298	Hyperinsulinemic hypoglycemia type 2	KCNJ11
sCe1/00299	Hyperinsulinemic hypoglycemia type 3	GCK
sCe1/00300	Hyperinsulinemic hypoglycemia type 6	GLUD1
sCe1/00301	Hyperinsulinemic hypoglycemia type 7	SLC16A1
sCe1/00302	Hyperinsulinism, UCP2 related	UCP2
sCe1/00303	Hyperlipidemia, familial combined, susceptibility to	USF1
sCe1/00304	Hyperlipoproteinemia type 1	LPL
sCe1/00305	Hyperlysinemia type 1	AASS
sCe1/00306	Hypermanganesemia with dystonia, polycythemia and cirrhosis	SLC30A10
sCe1/00307	Hypermethioninemia due to adenosine kinase deficiency	ADK
sCe1/00308	Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome	SLC25A15
sCe1/00309	Hyperoxaluria type 1	AGXT
sCe1/00310	Hyperoxaluria type 2	GRHPR
sCe1/00311	Hyperoxaluria type 3	HOGA1
sCe1/00312	Hyperoxaluria, SLC26A6 related	SLC26A6
sCe1/00313	Hyperphenylalaninemia, BH4 deficient, type C	QDPR
sCe1/00314	Hyperphenylalaninemia, BH4 deficient, type D	PCBD1
sCe1/00315	Hyperphenylalaninemia, BH4-deficient, type A	PTS

sCe1/00316	Hyperprolinemia type 1	PRODH
sCe1/00317	Hypertriglyceridemia, susceptibility to	LIPI
sCe1/00318	Hypoaldosteronism congenital due to CMO I deficiency	CYP11B2
sCe1/00319	Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2
sCe1/00320	Hypoalphalipoproteinemia	APOA1
sCe1/00321	Hypobetalipoproteinemia type 1	APOB
sCe1/00322	Hypocalcemia, autosomal dominant 2	GNA11
sCe1/00323	Hypocalciuric hypercalcemia, familial type 3	AP2S1
sCe1/00324	Hypoglycemia of infancy, leucine-sensitive	ABCC8
sCe1/00325	Hypoinsulinemic hypoglycemia with hemihypertrophy	AKT2
sCe1/00326	Hypomagnesemia type 1	TRPM6
sCe1/00327	Hypomagnesemia type 2	FXYD2
sCe1/00328	Hypomagnesemia type 3	CLDN16
sCe1/00329	Hypomagnesemia type 4	EGF
sCe1/00330	Hypomagnesemia type 5	CLDN19
sCe1/00331	Hypomagnesemia type 6	CNNM2
sCe1/00332	Hypophosphatasia, adult	ALPL
sCe1/00333	Hypophosphatasia, childhood	ALPL
sCe1/00334	Hypophosphatasia, infantile	ALPL
sCe1/00335	Hypophosphatemic rickets with hypercalciuria	SLC34A3
sCe1/00336	Hypophosphatemic rickets, autosomal dominant	FGF23
sCe1/00337	Hypouricemia, renal type 1	SLC22A12
sCe1/00338	Hypouricemia, renal type 2	SLC2A9
sCe1/00339	Insulin-like growth factor resistance	IGF1R
sCe1/00340	Isobutyryl-CoA dehydrogenase deficiency	ACAD8
sCe1/00341	Isovaleric academia	IVD
sCe1/00342	Krabbe disease	GALC
sCe1/00343	Krabbe disease, atypical	PSAP
sCe1/00344	L-2-hydroxyglutaric aciduria	L2HGDH

sCe1/00345	Lactase deficiency, congenital	LCT
sCe1/00346	Lactate dehydrogenase-B deficiency	LDHB
sCe1/00347	Lacticacidemia due to PDX1 deficiency	PDHX
sCe1/00348	Lactose intolerance, adult type	MCM6
sCe1/00349	LCAD deficiency	ACADL
sCe1/00350	LCAT DEFICIENCY	LCAT
sCe1/00351	Leukocyte adhesion deficiency	ITGB1
sCe1/00352	Leukocyte adhesion deficiency	ITGB2
sCe1/00353	Lipodystrophy generalized type 1	AGPAT2
sCe1/00354	Lipodystrophy generalized type 2	BSCL2
sCe1/00355	Lipodystrophy generalized type 4	PTRF
sCe1/00356	Lipodystrophy type 2, familial partial	LMNA
sCe1/00357	Lipodystrophy, familial partial, type 3	PPARG
sCe1/00358	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA
sCe1/00359	Lung alpha-beta hydrolase deficiency type 1	ABHD1
sCe1/00360	Lysosomal acid phosphatase deficiency	ACP2
sCe1/00361	Malonyl-CoA decarboxylase deficiency	MLYCD
sCe1/00362	Mannose-binding protein deficiency	MBL2
sCe1/00363	Mannosidosis, beta A, lysosomal-like	MANBAL
sCe1/00364	Mannosidosis-alpha	MAN2B1
sCe1/00365	Mannosidosis-beta	MANBA
sCe1/00366	Maple syrup urine disease type 1a	BCKDHA
sCe1/00367	Maple syrup urine disease type 1b	BCKDHB
sCe1/00368	Maple syrup urine disease type 2	DBT
sCe1/00369	Maple syrup urine disease type 3	DLD
sCe1/00370	Maple syrup urine disease, mild variant	PPM1K
sCe1/00371	Maturity-onset diabetes of the young type 1	HNF4A
sCe1/00372	Maturity-onset diabetes of the young type 10	INS
sCe1/00373	Maturity-onset diabetes of the young type 11	BLK

sCe1/00374	Maturity-onset diabetes of the young type 2	GCK
sCe1/00375	Maturity-onset diabetes of the young type 3	HNF1A
sCe1/00376	Maturity-onset diabetes of the young type 4	PDX1
sCe1/00377	Maturity-onset diabetes of the young type 5	HNF1B
sCe1/00378	Maturity-onset diabetes of the young type 6	NEUROD1
sCe1/00379	Maturity-onset diabetes of the young type 7	KLF11
sCe1/00380	Maturity-onset diabetes of the young type 8	CEL
sCe1/00381	Maturity-onset diabetes of the young type 9	PAX4
sCe1/00382	Maturity-onset diabetes of the young, NKX2-2 related	NKX2-2
sCe1/00383	Maturity-onset diabetes of the young, RFX6 related	RFX6
sCe1/00384	Maturity-onset diabetes of the young, ZFP57 related	ZFP57
sCe1/00385	Mediterranean fever	MEFV
sCe1/00386	MELAS syndrome, MT-TL1 related	MT-TL1
sCe1/00387	Metachromatic Leukodystrophy	ARSA
sCe1/00388	Methylacetoacetic aciduria	ACAT1
sCe1/00389	Methylcobalamin deficiency CblG type	MTR
sCe1/00390	Methylmalonate semialdehyde dehydrogenase deficiency	ALDH6A1
sCe1/00391	Methylmalonic aciduria CblA type	MMAA
sCe1/00392	Methylmalonic aciduria CblB type	MMAB
sCe1/00393	Methylmalonic aciduria CblC type	MMACHC
sCe1/00394	Methylmalonic aciduria CblD type	MMADHC
sCe1/00395	Methylmalonic aciduria CblF type	LMBRD1
sCe1/00396	Methylmalonic aciduria CblJ type	ABCD4
sCe1/00397	Methylmalonic aciduria CblR type	CD320
sCe1/00398	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	MUT
sCe1/00399	Methylmalonyl-CoA epimerase deficiency	MCEE
sCe1/00400	Mevalonic aciduria	MVK
sCe1/00401	Microvascular complications of diabetes type 1	VEGFA
sCe1/00402	Microvascular complications of diabetes type 6, susceptibility to	SOD2

sCe1/00403	Mitchell-Riley syndrome	RFX6
sCe1/00404	Mitochondrial complex III deficiency, nuclear type 7	UQCC2
sCe1/00405	Mitochondrial pyruvate carrier deficiency	MPC1
sCe1/00406	Molybdenum cofactor deficiency type A	MOCS1
sCe1/00407	Molybdenum cofactor deficiency type B	MOCS2
sCe1/00408	Molybdenum cofactor deficiency type C	GPHN
sCe1/00409	Monocarboxylate transporter 1 deficiency	SLC16A1
sCe1/00410	Mucolipidosis type 2 alpha/beta	GNPTAB
sCe1/00411	Mucolipidosis type 3	GNPTAB
sCe1/00412	Mucolipidosis type 3 gamma	GNPTG
sCe1/00413	Mucolipidosis type 4	MCOLN1
sCe1/00414	Mucopolysaccharidosis type 2	IDS
sCe1/00415	Mucopolysaccharidosis type 3A	SGSH
sCe1/00416	Mucopolysaccharidosis type 3B	NAGLU
sCe1/00417	Mucopolysaccharidosis type 3C	HGSNAT
sCe1/00418	Mucopolysaccharidosis type 3D	GNS
sCe1/00419	Mucopolysaccharidosis type 4A	GALNS
sCe1/00420	Mucopolysaccharidosis type 4B	GLB1
sCe1/00421	Mucopolysaccharidosis type 6	ARSB
sCe1/00422	Mucopolysaccharidosis type 7	GUSB
sCe1/00423	Mucopolysaccharidosis type 9	HYAL1
sCe1/00424	Mucopolysaccharidosis type IH	IDUA
sCe1/00425	Muscle glycogenosis	PHKA1
sCe1/00426	Myopathy due to myoadenylate deaminase deficiency	AMPD1
sCe1/00427	N-acetylglutamate synthase deficiency	NAGS
sCe1/00428	Neuraminidase deficiency	NEU1
sCe1/00429	Niemann-Pick disease type A/B	SMPD1
sCe1/00430	Niemann-Pick disease type C1	NPC1
sCe1/00431	Niemann-Pick disease type C2	NPC2

sCe1/00432	Odontohypophosphatasia	ALPL
sCe1/00433	Ornithine transcarbamoylase deficiency	OTC
sCe1/00434	Orotic aciduria	UMPS
sCe1/00435	Pancreatic agenesis type 2	PTF1A
sCe1/00436	Pancreatic and cerebellar agenesis	PTF1A
sCe1/00437	Pentosuria	DCXR
sCe1/00438	Periodic fever autosomal dominant	TNFRSF1A
sCe1/00439	Phenylketonuria	PAH
sCe1/00440	Phenylketonuria modifier, SLC7A5 related	SLC7A5
sCe1/00441	Phosphoenolpyruvate carboxykinase deficiency, cytosolic	PCK1
sCe1/00442	Phosphoenolpyruvate carboxykinase deficiency, mitochondrial	PCK2
sCe1/00443	Phosphoglycerate dehydrogenase deficiency	PHGDH
sCe1/00444	Phosphoribosylpyrophosphate synthetase superactivity	PRPS1
sCe1/00445	Phosphoserine aminotransferase deficiency	PSAT1
sCe1/00446	Phosphoserine phosphatase deficiency	PSPH
sCe1/00447	Pituitary stalk interruption syndrome, GPR161 related	GPR161
sCe1/00448	Pompe disease	GAA
sCe1/00449	Porphyria acute intermittent	HMBS
sCe1/00450	Porphyria congenital erythropoietic	UROS
sCe1/00451	Porphyria variegate	PPOX
sCe1/00452	Prolidase deficiency	PEPD
sCe1/00453	Propionic academia	PCCB
sCe1/00454	Propionic academia	PCCA
sCe1/00455	Prosaposin deficiency	PSAP
sCe1/00456	Protoporphyria, erythropoietic, X-linked	ALAS2
sCe1/00457	Pseudohermaphroditism with gynecomastia	HSD17B3
sCe1/00458	Pyridoxamine 5'-phosphate oxidase deficiency	PNPO
sCe1/00459	Pyruvate carboxylase deficiency	PC
sCe1/00460	Pyruvate dehydrogenase E1-alpha deficiency	PDHA1

sCe1/00461	Pyruvate dehydrogenase E1-beta deficiency	PDHB
sCe1/00462	Pyruvate dehydrogenase E2 deficiency	DLAT
sCe1/00463	Pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS
sCe1/00464	Pyruvate dehydrogenase phosphatase deficiency	PDP1
sCe1/00465	Pyruvate kinase deficiency with hemolytic anemia	PKLR
sCe1/00466	Refsum disease	PEX7
sCe1/00467	Refsum disease	PHYH
sCe1/00468	Riboflavin deficiency	SLC52A1
sCe1/00469	Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1
sCe1/00470	Rickets, vitamin D dependent, type 1	CYP27B1
sCe1/00471	Saccharopinuria	AASS
sCe1/00472	Sandhoff disease	HEXB
sCe1/00473	Sarcosinemia	SARDH
sCe1/00474	Scheie syndrome	IDUA
sCe1/00475	Schindler disease	NAGA
sCe1/00476	Serine hydrolase deficiency, SERHL2 related	SERHL2
sCe1/00477	Succinic semialdehyde dehydrogenase deficiency	ALDH5A1
sCe1/00478	Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1
sCe1/00479	Sucrase-isomaltase deficiency	SI
sCe1/00480	Sulfatase deficiency	SUMF1
sCe1/00481	Sulfite oxidase deficiency	SUOX
sCe1/00482	Surfactant metabolism dysfunction	SFTPD
sCe1/00483	Surfactant metabolism dysfunction type 1	SFTPB
sCe1/00484	Surfactant metabolism dysfunction type 2	SFTPC
sCe1/00485	Surfactant metabolism dysfunction type 3	ABCA3
sCe1/00486	Surfactant metabolism dysfunction type 4	CSF2RA
sCe1/00487	Surfactant metabolism dysfunction type 5	CSF2RB
sCe1/00488	Tangier disease	ABCA1
sCe1/00489	Tay-Sachs disease	HEXA

sCe1/00490	Tay-Sachs disease AB variant	GM2A
sCe1/00491	Thiamine metabolism dysfunction syndrome type 5	TPK1
sCe1/00492	TJP1 deficiency	TJP1
sCe1/00493	TPMT deficiency	TPMT
sCe1/00494	Transaldolase deficiency	TALDO1
sCe1/00495	Transcobalamin II deficiency	TCN2
sCe1/00496	Trifunctional protein deficiency	HADHA
sCe1/00497	Trimethylaminuria	FMO3
sCe1/00498	Triosephosphate isomerase deficiency	TPI1
sCe1/00499	Tyrosine kinase 2 deficiency	TYK2
sCe1/00500	Tyrosinemia type 1	FAH
sCe1/00501	Tyrosinemia type 1B	GSTZ1
sCe1/00502	Tyrosinemia type 2	TAT
sCe1/00503	Tyrosinemia type 3	HPD
sCe1/00504	Urbach-Wiethe disease	ECM1
sCe1/00505	Von-Gierke disease	G6PC
sCe1/00506	Wilson disease	ATP7B
sCe1/00507	Wolman disease	LIPA
sCe1/00508	Xanthinuria type 1	XDH
sCe5/0001	3MC syndrome type 1	MASP1
sCe5/0002	3MC syndrome type 2	COLEC11
sCe5/0003	Achondrogenesis type 1A	TRIP11
sCe5/0004	Achondrogenesis type 1B	SLC26A2
sCe5/0005	Achondrogenesis type 2	COL2A1
sCe5/0006	Achondroplasia	FGFR3
sCe5/0009	Acrodysostosis 2	PDE4D
sCe5/0010	Acrofacial dysostosis 1, Nager type	SF3B4
sCe5/0011	Adams-Oliver syndrome type 1	ARHGAP31
sCe5/0012	Adams-Oliver syndrome type 2	DOCK6

sCe5/0013	Adams-Oliver syndrome type 3	RBPJ
sCe5/0014	Adams-Oliver syndrome type 4	EOGT
sCe5/0023	Amelogenesis imperfecta type 1A	LAMB3
sCe5/0024	Amelogenesis imperfecta type 1B	ENAM
sCe5/0025	Amelogenesis imperfecta type 1C	ENAM
sCe5/0026	Amelogenesis imperfecta type 1E	AMELX
sCe5/0027	Amelogenesis imperfecta type 1F	AMBN
sCe5/0028	Amelogenesis imperfecta type 1G	FAM20A
sCe5/0029	Amelogenesis imperfecta type 1H	ITGB6
sCe5/0030	Amelogenesis imperfecta type 2A1	KLK4
sCe5/0031	Amelogenesis imperfecta type 2A2	MMP20
sCe5/0032	Amelogenesis imperfecta type 2A3	WDR72
sCe5/0033	Amelogenesis imperfecta type 2A4	C4orf26
sCe5/0034	Amelogenesis imperfecta type 2A5	SLC24A4
sCe5/0035	Amelogenesis imperfecta type 3	FAM83H
sCe5/0036	Amelogenesis imperfecta type 4	DLX3
sCe5/0037	Amelotin deficiency	AMTN
sCe5/0040	Arthrogryposis, distal, type 1A	TPM2
sCe5/0041	Arthrogryposis, distal, type 1B	MYBPC1
sCe5/0042	Arthrogryposis, distal, type 2A	MYH3
sCe5/0043	Arthrogryposis, distal, type 2B	MYH3
sCe5/0044	Arthrogryposis, distal, type 2B	TNNI2
sCe5/0045	Arthrogryposis, distal, type 2B	TNNT3
sCe5/0046	Arthrogryposis, distal, type 3	PIEZO2
sCe5/0047	Arthrogryposis, distal, type 5	PIEZO2
sCe5/0048	Arthrogryposis, distal, type 5D	ECEL1
sCe5/0049	Arthrogryposis, distal, type 7	MYH8
sCe5/0050	Arthrogryposis, mental retardation, and seizures	SLC35A3
sCe5/0051	Arthrogryposis, renal dysfunction, and cholestasis type 1	VPS33B

sCe5/0052	Arthrogryposis, renal dysfunction, and cholestasis type 2	VIPAS39
sCe5/0053	Arthropathy, progressive pseudorheumatoid, of childhood	WISP3
sCe5/0054	Atelosteogenesis type 1	FLNB
sCe5/0055	Atelosteogenesis type 3	FLNB
sCe5/0069	Avascular necrosis of the femoral head, primary	COL2A1
sCe5/0073	Beare-Stevenson cutis gyrata syndrome	FGFR2
sCe5/0074	Bent bone dysplasia syndrome	FGFR2
sCe5/0075	Bone mineral density QTL18, osteoporosis	PLS3
sCe5/0076	Buschke-Ollendorff syndrome	LEMD3
sCe5/0084	Chondrocalcinosis type 2	ANKH
sCe5/0085	Chondrodysplasia punctata, X-linked dominant	EBP
sCe5/0086	Chondrodysplasia punctata, X-linked recessive	ARSE
sCe5/0087	Chondrosarcoma, familial	EXT1
sCe5/0088	Cleidocranial dysplasia	RUNX2
sCe5/0094	Contractural arachnodactyly, congenital	FBN2
sCe5/0095	Cornelia de Lange syndrome type 1	NIPBL
sCe5/0096	Cornelia de Lange syndrome type 2	SMC1A
sCe5/0097	Cornelia de Lange syndrome type 3	SMC3
sCe5/0098	Cornelia de Lange syndrome type 4	RAD21
sCe5/0099	Cornelia de Lange syndrome type 5	HDAC8
sCe5/0100	Craniofacial-skeletal-dermatologic dysplasia	FGFR2
sCe5/0101	Crouzon syndrome with acanthosis nigricans	FGFR3
sCe5/0112	Czech dysplasia	COL2A1
sCe5/0113	Dentin dysplasia, type 2	DSPP
sCe5/0114	Dentinogenesis imperfecta, Shields type 2	DSPP
sCe5/0115	Dentinogenesis imperfecta, Shields type 3	DSPP
sCe5/0118	Diaphyseal medullary stenosis with malignant fibrous histiocytoma	MTAP
sCe5/0130	Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2
sCe5/0178	Epiphyseal dysplasia, multiple, type 1	COMP

sCe5/0179	Epiphyseal dysplasia, multiple, type 3	COL9A3
sCe5/0180	Epiphyseal dysplasia, multiple, type 5	MATN3
sCe5/0185	Exostoses, multiple, type 1	EXT1
sCe5/0186	Exostoses, multiple, type 2	EXT2
sCe5/0188	Feingold syndrome type 2	MIR17HG
sCe5/0189	Fibrochondrogenesis 2	COL11A2
sCe5/0190	Fibrochondrogenesis type 1	COL11A1
sCe5/0191	Fibrodysplasia ossificans progressive	ACVR1
sCe5/0192	Fibrosis of extraocular muscles, congenital type 2	PHOX2A
sCe5/0193	Floating-Harbor syndrome	SRCAP
sCe5/0195	Frank-ter Haar syndrome	SH3PXD2B
sCe5/0196	Geleophysic dysplasia type 1	ADAMTSL2
sCe5/0198	Ghosal hematodiaphyseal syndrome	TBXAS1
sCe5/0199	Gnathodiaphyseal dysplasia	ANO5
sCe5/0200	Gracile bone dysplasia	FAM111A
sCe5/0206	Greenberg skeletal dysplasia	LBR
sCe5/0209	Haim-Munk syndrome	CTSC
sCe5/0210	Heimler syndrome type 1	PEX1
sCe5/0223	Hypertrophic osteoarthropathy type 1	HPGD
sCe5/0224	Hypertrophic osteoarthropathy type 2	SLCO2A1
sCe5/0225	Hypochondroplasia	FGFR3
sCe5/0226	Hypophosphatemic rickets with hypercalciuria	SLC34A3
sCe5/0227	Hypophosphatemic rickets, autosomal dominant	FGF23
sCe5/0228	Hypophosphatemic rickets, autosomal recessive type 1	DMP1
sCe5/0229	Hypophosphatemic rickets, autosomal recessive type 2	ENPP1
sCe5/0230	Hypophosphatemic rickets, X-linked	PHEX
sCe5/0231	Hypotrichosis type 1	APCDD1
sCe5/0317	Legg-Calve-Perthes disease	COL2A1
sCe5/0321	Loeys-Dietz syndrome type 1A	TGFBR1

sCe5/0322	Loeys-Dietz syndrome type 1B	TGFBR2
sCe5/0323	Loeys-Dietz syndrome type 1C	SMAD3
sCe5/0324	Loeys-Dietz syndrome type 2A	TGFBR1
sCe5/0325	Loeys-Dietz syndrome type 2B	TGFBR2
sCe5/0332	Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24
sCe5/0333	Marfan syndrome, TGFBR1 related	TGFBR1
sCe5/0334	Marfan syndrome, TGFBR2 related	TGFBR2
sCe5/0335	McKusick-Kaufman syndrome	MKKS
sCe5/0338	MERRF syndrome, MT-TK related	MT-TK
sCe5/0339	MERRF syndrome, MT-TP related	MT-TP
sCe5/0340	Metaphyseal chondrodysplasia, Schmid type	COL10A1
sCe5/0341	Metaphyseal chondromatosis with increased urinary excretion of D-2-hydroxyglutarate	IDH1
sCe5/0343	Multicentric carpotarsal osteolysis syndrome	MAFB
sCe5/0344	Multicentric osteolysis, nodulosis, and arthropathy	MMP2
sCe5/0345	Multiple pterygium syndrome lethal type	CHRNA1
sCe5/0346	Multiple pterygium syndrome lethal type	CHRND
sCe5/0349	Nail-Patella syndrome	LMX1B
sCe5/0350	Nephrolithiasis/osteoporosis, hypophosphatemic, type 2	SLC9A3R1
sCe5/0361	Odontoonychodermal dysplasia	WNT10A
sCe5/0365	Omodysplasia type 1	GPC6
sCe5/0366	Opsismodysplasia	INPPL1
sCe5/0367	Osseous heteroplasia, progressive	GNAS
sCe5/0368	Osteoarthritis with mild chondrodysplasia	COL2A1
sCe5/0369	Osteogenesis and dental anomalies, CSF1 related	CSF1
sCe5/0370	Osteogenesis disorders, CREB3L1 related	CREB3L1
sCe5/0371	Osteogenesis imperfect	COL1A1
sCe5/0372	Osteogenesis imperfect	COL1A2
sCe5/0373	Osteogenesis imperfecta type 10	SERPINH1
sCe5/0374	Osteogenesis imperfecta type 11	FKBP10

sCe5/0375	Osteogenesis imperfecta type 12	SP7
sCe5/0376	Osteogenesis imperfecta type 13	BMP1
sCe5/0377	Osteogenesis imperfecta type 14	TMEM38B
sCe5/0378	Osteogenesis imperfecta type 15	WNT1
sCe5/0379	Osteogenesis imperfecta type 5	IFITM5
sCe5/0380	Osteogenesis imperfecta type 6	SERPINF1
sCe5/0381	Osteogenesis imperfecta type 7	CRTAP
sCe5/0382	Osteogenesis imperfecta type 8	P3H1
sCe5/0383	Osteogenesis imperfecta type 9	PPIB
sCe5/0384	Osteogenesis imperfecta with congenital joint contractures	PLOD2
sCe5/0385	Osteolysis, familial expansile	TNFRSF11A
sCe5/0386	Osteomyelitis, sterile multifocal, with periostitis and pustulosis	IL1RN
sCe5/0387	Osteopathia striata with cranial sclerosis	AMER1
sCe5/0388	Osteopetrosis of infancy, malignant	SNX10
sCe5/0389	Osteopetrosis, autosomal dominant type 1	CLCN7
sCe5/0390	Osteopetrosis, autosomal recessive type 1	TCIRG1
sCe5/0391	Osteopetrosis, autosomal recessive type 2	TNFSF11
sCe5/0392	Osteopetrosis, autosomal recessive type 3	CA2
sCe5/0393	Osteopetrosis, autosomal recessive type 4	CLCN7
sCe5/0394	Osteopetrosis, autosomal recessive type 5	OSTM1
sCe5/0395	Osteopetrosis, autosomal recessive type 6	PLEKHM1
sCe5/0396	Osteopetrosis, autosomal recessive type 7	TNFRSF11A
sCe5/0397	Osteoporosis pseudoglioma syndrome	LRP5
sCe5/0398	Pachyonychia congenita type 1	KRT16
sCe5/0399	Pachyonychia congenita type 2	KRT17
sCe5/0400	Pachyonychia congenita type 3	KRT6A
sCe5/0401	Pachyonychia congenita type 4	KRT6B
sCe5/0402	Paget disease of bone	SQSTM1
sCe5/0403	Paget disease, juvenile	TNFRSF11B

sCe5/0416	Pseudoachondroplasia	COMP
sCe5/0424	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 1	TERT
sCe5/0425	Pulmonary fibrosis and/or bone marrow failure, telomere-related, type 4	PARN
sCe5/0426	Pycnodysostosis	CTSK
sCe5/0429	Radioulnar synostosis, FGFRL1 related	FGFRL1
sCe5/0430	Raine syndrome	FAM20C
sCe5/0436	Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1
sCe5/0437	Rickets, vitamin D dependent, type 1	CYP27B1
sCe5/0438	Rickets, vitamin D-resistant, type 2A	VDR
sCe5/0450	Smith-McCort dysplasia	DYM
sCe5/0451	Spondylocarpotarsal synostosis syndrome	FLNB
sCe5/0452	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like	SLC39A13
sCe5/0453	Spondylocostal dysostosis, autosomal recessive type 1	DLL3
sCe5/0454	Spondylocostal dysostosis, autosomal recessive type 2	MESP2
sCe5/0455	Spondylocostal dysostosis, autosomal recessive type 3	LFNG
sCe5/0456	Spondyloenchondrodysplasia with immune dysregulation	ACP5
sCe5/0457	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6
sCe5/0458	Spondyloepimetaphyseal dysplasia, MATN3 related	MATN3
sCe5/0459	Spondyloepiphyseal dysplasia with congenital joint dislocations	CHST3
sCe5/0460	Spondylometaepiphyseal dysplasia, short limb-hand type	DDR2
sCe5/0470	Terminal osseous dysplasis	FLNA
sCe5/0471	Trichodontoosseous syndrome	DLX3
sCe5/0472	Trichohepatoenteric syndrome type 1	TTC37
sCe5/0473	Trichohepatoenteric syndrome type 2	SKIV2L
sCe5/0474	Trichorhinophalangeal syndrome type 1	TRPS1
sCe5/0475	Trichothiodystrophy	ERCC3
sCe5/0476	Trichothiodystrophy	ERCC2
sCe5/0477	Trichothiodystrophy	GTF2H5
sCe5/0478	Trichothiodystrophy, nonphotosensitive type 1	MPLKIP

sCe5/0479	Tuftelin deficiency	TUFT1
sCe5/0483	Van Buchem disease	SOST
sCe5/0491	Winchester Syndrome	MMP14
sCe5/0492	Wolcott-Rallison syndrome	EIF2AK3
sCe5/0501	XFE progeroid syndrome	ERCC4
sCe8/0001	17-beta hydroxysteroid dehydrogenase X deficiency	HSD17B10
sCe8/0002	Achalasia addisonianism alacrimia syndrome	AAAS
sCe8/0003	Acromegaly, predisposition to, due to germline GPR101 mutation	GPR101
sCe8/0004	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	CYP11A1
sCe8/0005	Adrenocorticotropic hormone deficiency	TBX19
sCe8/0009	Androgen insensitivity	AR
sCe8/0010	Androgen insensitivity, partial, with or without breast cancer	AR
sCe8/0011	Androgen-binding protein deficiency	SHBG
sCe8/0013	Bardet-Biedl syndrome type 1	BBS1
sCe8/0014	Bardet-Biedl syndrome type 10	BBS10
sCe8/0015	Bardet-Biedl syndrome type 11	TRIM32
sCe8/0016	Bardet-Biedl syndrome type 12	BBS12
sCe8/0017	Bardet-Biedl syndrome type 13	MKS1
sCe8/0018	Bardet-Biedl syndrome type 14	CEP290
sCe8/0019	Bardet-Biedl syndrome type 15	WDPCP
sCe8/0020	Bardet-Biedl syndrome type 2	BBS2
sCe8/0021	Bardet-Biedl syndrome type 3	ARL6
sCe8/0022	Bardet-Biedl syndrome type 4	BBS4
sCe8/0023	Bardet-Biedl syndrome type 5	BBS5
sCe8/0024	Bardet-Biedl syndrome type 6	MKKS
sCe8/0025	Bardet-Biedl syndrome type 7	BBS7
sCe8/0026	Bardet-Biedl syndrome type 8	TTC8
sCe8/0027	Bardet-Biedl syndrome type 9	BBS9
sCe8/0028	Bardet-Biedl syndrome, LZTFL1 related	LZTFL1

sCe8/0029	Bardet-Biedl syndrome, modifier of, CCDC28B related	CCDC28B
sCe8/0030	Bartter syndrome	SLC12A7
sCe8/0031	Bartter syndrome	SLC12A5
sCe8/0032	Bartter syndrome	SLC12A3
sCe8/0033	Bartter syndrome	SLC12A2
sCe8/0034	Bartter syndrome type 1	SLC12A1
sCe8/0035	Bartter syndrome type 2	KCNJ1
sCe8/0036	Bartter syndrome type 3	CLCNKB
sCe8/0037	Bartter syndrome type 4a	BSND
sCe8/0038	Bartter syndrome type 4b	CLCNKA
sCe8/0039	Cholestasis, infantile, NR1H4 related	NR1H4
sCe8/0040	Crigler-Najjar syndrome, type 1	UGT1A1
sCe8/0041	Crigler-Najjar syndrome, type 2	UGT1A1
sCe8/0044	Cystinuria	PREPL
sCe8/0045	Cystinuria	SLC7A9
sCe8/0046	Cystinuria	SLC3A1
sCe8/0049	Dubin-Johnson syndrome	ABCC2
sCe8/0051	Estrogen resistance	ESR1
sCe8/0056	Fanconi-Bickel syndrome	SLC2A2
sCe8/0069	Gilbert syndrome	UGT1A1
sCe8/0072	Glucocorticoid resistance, generalized	NR3C1
sCe8/0073	Glycine encephalopathy	AMT
sCe8/0074	Glycine encephalopathy	GLDC
sCe8/0075	Glycine N-methyltransferase deficiency	GNMT
sCe8/0076	Goitre, multinodular	KEAP1
sCe8/0077	Growth hormone deficiency	GH1
sCe8/0078	Growth hormone deficiency	GHRHR
sCe8/0079	Growth hormone insensitivity with immunodeficiency	STAT5B
sCe8/0090	Hepatic failure, early onset, and neurologic disorder	SCO1

sCe8/0091	Hepatic venoocclusive disease with immunodeficiency	SP110
sCe8/0092	Hyperaldosteronism type 3	KCNJ5
sCe8/0093	Hyperbilirubinemia, familial transient neonatal	UGT1A1
sCe8/0094	Hyperbilirubinemia, Rotor type	SLCO1B3
sCe8/0095	Hyperbilirubinemia, Rotor type	SLCO1B1
sCe8/0096	Hyperparathyroidism type 1, familial	CDC73
sCe8/0097	Hyperparathyroidism type 2, familial	CDC73
sCe8/0098	Hyperparathyroidism, neonatal severe	CASR
sCe8/0099	Hypertriglyceridemia, transient infantile	GPD1
sCe8/0101	Hypocalcemia, autosomal dominant, with Bartter syndrome	CASR
sCe8/0102	Hypocalciuric hypercalcemia, type 1	CASR
sCe8/0103	Hypogonadotropic hypogonadism	LHB
sCe8/0104	Hypogonadotropic hypogonadism	KISS1R
sCe8/0105	Hypogonadotropic hypogonadism	NSMF
sCe8/0106	Hypogonadotropic hypogonadism type 10 with or without anosmia	TAC3
sCe8/0107	Hypogonadotropic hypogonadism type 11 with or without anosmia	TACR3
sCe8/0108	Hypogonadotropic hypogonadism type 12 with or without anosmia	GNRH1
sCe8/0109	Hypogonadotropic hypogonadism type 15 with or without anosmia	HS6ST1
sCe8/0110	Hypogonadotropic hypogonadism type 3 with or without anosmia	PROKR2
sCe8/0111	Hypogonadotropic hypogonadism type 7 with or without anosmia	GNRHR
sCe8/0112	Hypoparathyroidism	PTH
sCe8/0113	Hypoparathyroidism, familial isolated	GCM2
sCe8/0114	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3
sCe8/0115	Hypoparathyroidism-retardation-dysmorphism syndrome	TBCE
sCe8/0116	Hypophosphatemic rickets	CLCN5
sCe8/0117	Hypothyroidism congenital nongoitrous type 1	TSHR
sCe8/0118	Hypothyroidism congenital nongoitrous type 2, familial	PAX8
sCe8/0119	Hypothyroidism congenital nongoitrous type 4	TSHB
sCe8/0120	Hypothyroidism congenital nongoitrous type 6	THRA

sCe8/0121	Hypothyroidism, isolated, TRHR related	TRHR
sCe8/0122	Hypotonia-cystinuria syndrome	PREPL
sCe8/0125	Immunodeficiency with natural killer cell deficiency	MCM4
sCe8/0126	Immunodeficiency, common variable type 7	CR2
sCe8/0128	Infantile liver failure syndrome type 2	NBAS
sCe8/0130	Intrahepatic cholestasis of pregnancy, NR1H4 related	NR1H4
sCe8/0131	Johanson Blizzard syndrome	UBR1
sCe8/0132	Kallmann syndrome type 1	ANOS1
sCe8/0133	Kallmann syndrome type 2	FGFR1
sCe8/0134	Kallmann syndrome type 4	PROK2
sCe8/0135	Kallmann syndrome, SEMA3A related	SEMA3A
sCe8/0136	Laron syndrome	GHR
sCe8/0137	Liddle syndrome	SCNN1B
sCe8/0138	Liddle syndrome	SCNN1G
sCe8/0139	Lipoid congenital adrenal hyperplasia	STAR
sCe8/0140	Lipoprotein glomerulopathy	APOE
sCe8/0141	Liver failure transient infantile	TRMU
sCe8/0142	Lowe oculocerebrorenal syndrome	OCRL
sCe8/0143	LYSINURIC PROTEIN INTOLERANCE	SLC7A7
sCe8/0152	Menkes disease	ATP7A
sCe8/0179	Obesity	MC4R
sCe8/0180	Obesity due to leptin deficiency	LEP
sCe8/0181	Obesity with adrenal insufficiency and red hair	POMC
sCe8/0182	Obesity with impaired prohormone processing	PCSK1
sCe8/0183	Obesity, early-onset, susceptibility to	POMC
sCe8/0184	Obesity, severe	PPARG
sCe8/0185	Obesity, susceptibility to, SLC6A14 related	SLC6A14
sCe8/0186	Ovalocytosis	SLC4A1
sCe8/0187	Pancreatitis	CTRC

sCe8/0188	Pancreatitis	SPINK1
sCe8/0189	Pancreatitis	PRSS1
sCe8/0190	Pancreatitis, chronic, early onset	CPA1
sCe8/0191	Pancreatitis, chronic, protection against	PRSS2
sCe8/0192	Panhypopituitarism, X-linked	SOX3
sCe8/0193	Parathyroid adenoma with cystic changes, familial	CDC73
sCe8/0194	Pendred syndrome	SLC26A4
sCe8/0195	Pigmented nodular adrenocortical disease type 1, primary	PRKAR1A
sCe8/0196	Pituitary hormone deficiency, combined type 3	LHX3
sCe8/0197	Pituitary hormone deficiency, combined type 4	LHX4
sCe8/0198	Polycystic kidney and hepatic disease	PKHD1
sCe8/0202	Polycystic liver disease	PRKCSH
sCe8/0203	Polycystic ovary syndrome type 1	SULT2B1
sCe8/0204	Polycystic ovary syndrome type 1	SULT2A1
sCe8/0205	Precocious puberty, male	LHCGR
sCe8/0206	Premature ovarian failure type 1	FMR1
sCe8/0207	Primary aldosteronism, seizures, and neurologic abnormalities	CACNA1D
sCe8/0209	Pseudohypoaldosteronism, type 1, autosomal dominant	NR3C2
sCe8/0210	Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1B
sCe8/0211	Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1G
sCe8/0212	Pseudohypoaldosteronism, type 1, autosomal recessive	SCNN1A
sCe8/0213	Pseudohypoaldosteronism, type 2B	WNK4
sCe8/0214	Pseudohypoparathyroidism type 1A	GNAS
sCe8/0215	Pseudohypoparathyroidism type 1B	GNAS
sCe8/0216	Pseudohypoparathyroidism type 1C	GNAS
sCe8/0217	Pseudopseudohypoparathyroidism	GNAS
sCe8/0232	SERKAL syndrome	WNT4
sCe8/0233	Thromocytopenia-Absent-Radius-Syndrome	RBM8A
sCe8/0234	Thyroid dyshormonogenesis type 1	SLC5A5
sCe8/0235	Thyroid dyshormonogenesis type 2A	TPO

sCe8/0236	Thyroid dyshormonogenesis type 3	TG
sCe8/0237	Thyroid dyshormonogenesis type 4	IYD
sCe8/0238	Thyroid dyshormonogenesis type 5	DUOXA2
sCe8/0239	Thyroid dyshormonogenesis type 6	DUOX1
sCe8/0240	Thyroid dyshormonogenesis type 6	DUOX2
sCe8/0241	Thyroid hormone metabolism abnormal	SECISBP2
sCe8/0242	Thyroid hormone resistance	THRB
sCe8/0243	Trifunctional protein deficiency	HADHB
sCe8/0248	Wolcott-Rallison syndrome	EIF2AK3

Panel

Code	Product Name	TAT-day
	Clinical Exome Whole exome Solo	
pCe1/00001	Brain iron accumulation syndromes panel	25
pCe1/00002	Ceroid lipofuscinosis panel	25
pCe1/00003	Congenital glycosylation disease panel	25
pCe1/00004	Diabetes neonatal panel	25
pCe1/00005	Diamond-Blackfan anemia panel	25
pCe1/00006	Familial hypercholesterolemia panel	25
pCe1/00007	Fatty acid oxidation disorder panel	25
pCe1/00008	Glycogen storage disease panel (advanced)	25
pCe1/00009	Glycogen storage disease panel (basic)	25
pCe1/00010	Hyperinsulinemic hypoglycemia panel	25
pCe1/00011	Leigh syndrome and mitochondrial encephalopathy panel	25
pCe1/00012	Lipodystrophy panel	25
pCe1/00013	Lysosomal storage disease panel	25
pCe1/00014	Methylmalonic acidemia panel (advanced)	25
pCe1/00015	Methylmalonic acidemia panel (basic)	25
pCe1/00016	MODY panel	25
pCe1/00017	Mucopolysaccharidosis panel	25
pCe1/00018	Non ketotic hyperglycinemia panel	25
pCe1/00019	Obesity panel	25
pCe1/00020	Refsum disease panel	25
pCe1/00021	Renal tubular acidosis panel	25
pCe1/00022	Surfactant metabolism dysfunction panel	25
pCe1/00023	Urea cycle disorder panel	25
pCe1/00024	Zellweger syndrome panel	25
pCe1/00025	CentoSphingo Enzyme Panel	
pCe1/00026	CentoSphingo Enzyme Panel X-TRA	

pCe1/00027	CentoMPS Enzyme Panel	
pCe1/00028	CentoMPS Enzyme Panel ***X-TRA***	
pCe1/00029	CentoNCL Enzyme Panel	
pCe1/00030	CentoNCL Enzyme Panel X-TRA	
pCe1/00031	CentoLSD Enzyme Panel	
pCe1/00032	CentoLSD Enzyme Panel ***X-TRA***	
pCe5/0001	Abnormal mineralization panel	25
pCe5/0008	Cornelia de Lange syndrome panel	25
pCe5/0018	Osteogenesis imperfecta and low bone density disorders panel	25
pCe5/0019	Osteopetrosis and high bone density disorders panel	25
pCe8/0002	Bardet Biedl panel	25
pCe8/0003	Bartter Syndrome panel	25
pCe8/0004	Combined pituitary hormone deficiency panel	25
pCe8/0005	Congenital adrenal hyperplasia panel	25
pCe8/0007	Intrahepatic cholestasis panel	25
pCe8/0008	Kallmann syndrome and Hypogonadotropic hypogonadism panel	25
pCe8/0011	Neonatal mitochondrial hepatopathies panel	25
pCe8/0014	Pancreatitis panel	25
pCe8/0016	Pseudohypoaldosteronism panel	25

DNAFit

Code	Product
DF1	Diet Pro
DF2	Diet Fitness Pro 360