LIST OF GENES ANALYZED BY BABYNEXT® CARING FOR LIFE

eurofins | Genoma

Gene	*OMIM	Disease	OMIM	Inheritance	Disease area
ABCD1	*300371	X-Linked Adrenoleukodystrophy	#300100	XLD	Endocrinology and Neurology
ABCD3	*170995	Bile acid synthesis defect congenital 5	#616278	AR	Gastroeneterology
ABCD4	*603214	Methylmalonic Aciduria and Homocystinuria cblJ type	#614857	AR	Metabolic
ACAD8	*604773	Isobutyryl-CoA dehydrogenase deficiency (IBDD)	#611283	AR	Metabolic
ACADM	*607008	Acyl-CoA dehydrogenase medium chain deficiency of	#201450	AR	Metabolic
ACADS	*606885	Acyl-CoA dehydrogenase short-chain deficiency of	#201470	AR	Metabolic
ACADSB	*600301	2-methylbutyrylglycinuria (Short/branched chain acyl-CoA dehydrogenase deficiency)	#610006	AR	Metabolic
ACADVL	*609575	Very long-chain acyl-CoA dehydrogenase deficiency	#201475	AR	Metabolic
ACAT1	*607809	Beta-ketothiolase deficiency	#203750	AR	Metabolic
ACOX2	*601641	Bile acid synthesis defect congenital 6	#617308	AR	Gastroeneterology
ACTG1	*102560	Deafness autosomal dominant 20/26	#604717	AD	Otorhinolaryngology
ADA	*608958	Severe combined immunodeficiency due to ADA deficiency	#102700	AR	Immunology
ADK	*102750	Hypermethioninemia due to adenosine kinase deficiency	#614300	AR	Metabolic
AGL	*610860	Glycogen storage disease type III	#232400	AR	Metabolic
AGXT	*604285	Primary hyperoxaluria type 1	#259900	AR	Metabolic
AHCY	*180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	#613752	AR	Metabolic
AK2	*103020	Reticular dysgenesis	#267500	AR	Immunology
AKR1D1	*604741	Bile acid synthesis defect congenital 2	#235555	AR	Gastroeneterology
ALDH4A1	*606811	Hyperprolinemia type II	#239510	AR	Metabolic
ALDOB	*612724	Fructose intolerance hereditary	#229600	AR	Metabolic

AMACR	*604489	Bile acid synthesis defect congenital 4	#214950	AR	Gastroeneterology
ANK1	*612641	Spherocytosis type 1	#182900	AD/AR	Hematology
ARG1	*608313	Argininemia	#207800	AR	Metabolic
ARSA	*607574	Metachromatic leukodystrophy	#250100	AR	Metabolic
ARSB	*611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	#253200	AR	Metabolic
ASL	*608310	Argininosuccinic aciduria	#207900	AR	Metabolic
ASS1	*603470	Citrullinemia Type 1	#215700	AR	Metabolic
ATP11A	*605868	Deafness autosomal dominant 84	#619810	AD	Otorhinolaryngology
ATP2B2	*108733	Deafness autosomal dominant 82	#619804	AD	Otorhinolaryngology
ATP6V0A4	*605239	Renal tubular acidosis distal 3 with or without sensorineural hearing loss	#602722	AR	Nephrology
ATP6V1B1	*192132	Renal tubular acidosis distal 2 with progressive sensorineural hearing loss	#267300	AR	Nephrology
AUH	* 600529	3-methylglutaconic aciduria type I	#250950	AR	Metabolic
BCKDHA	*608348	Maple syrup urine disease type Ia	#248600	AR	Metabolic
BCKDHB	*248611	Maple syrup urine disease type Ib	#248600	AR	Metabolic
BTD	*609019	Biotinidase deficiency	#253260	AR	Metabolic
BTK	*300300	Agammaglobulinemia X-linked 1	#300755	XLR	Immunology
CABP2	*607314	Deafness autosomal recessive 93	#614899	AR	Otorhinolaryngology
CBS	*613381	Homocystinuria B6-responsive and nonresponsive types	#236200	AR	Metabolic
CD247	*186780	Immunodeficiency 25	# 610163	AR	Immunology
CD320	*606475	Methylmalonic aciduria transient due to transcobalamin receptor defect	#613646	AR	Metabolic
CD3D	*186790	Immunodeficiency 19	#615617	AR	Immunology
CD3E	*186830	Immunodeficiency 18	#615615	AR	Immunology
CDC14A	*603504	Deafness autosomal recessive 32 with or without immotile sperm	#608653	AR	Otorhinolaryngology
CDH23	*605516	Deafness autosomal recessive 12	#601386	AR	Otorhinolaryngology

CEACAM16	*614591	Deafness autosomal dominant 4B	#614614	AD	Otorhinolaryngology
CEACAM16	*614591	Deafness autosomal recessive 113	#618410	AR	Otorhinolaryngology
CFTR	*602421	Cystic fibrosis	#219700	AR	Pneumology/Gastroenetrology
CIB2	*605564	Deafness autosomal recessive 48	#609439	AR	Otorhinolaryngology
CLDN14	*605608	Deafness autosomal recessive 29	#614035	AR	Otorhinolaryngology
COCH	*603196	Deafness autosomal dominant 9	#601369	AD	Otorhinolaryngology
COL11A1	*120280	Deafness autosomal dominant 37	#618533	AD	Otorhinolaryngology
COL11A2	*120290	Deafness autosomal dominant 13	#601868	AD	Otorhinolaryngology
COL11A2	*120290	Deafness autosomal recessive 53	#609706	AR	Otorhinolaryngology
COL4A3	*120070	Alport syndrome 3 autosomal dominant	#104200	AD	Nephrology
COL4A3	*120070	Alport syndrome COL4A3-related	#203780	AR	Nephrology
COL4A4	*120131	Alport syndrome autosomal recessive	#203780	AR	Nephrology/Otolaryngology
COL4A5	*303630	Alport syndrome	#301050	XLD	Nephrology/Otolaryngology
CPT1A	* 600528	Carnitine palmitoyltransferase type I deficiency	#255120	AR	Metabolic
CPT2	* 600650	Carnitine palmitoyltransferase type II deficiency	#255110	AD, AR	Metabolic
CRYM	*123740	Deafness autosomal dominant 40	#616357	AD	Otorhinolaryngology
CTNS	*219800	Cystinosis nephropathic	#219800	AR	Metabolic
CYBA	*608508	Chronic granulomatous disease autosomal due to deficiency of CYBA	#233690	AR	Immunology
CYBB	*300481	Chronic granulomatous disease CYBB-related	#306400	XLR	Immunology
CYP7B1	*603711	Bile acid synthesis defect congenital 3	#613812	AR	Gastroeneterology
DBT	*248610	Maple syrup urine disease type II	#248600	AR	Metabolic
DCLRE1C	*605988	Severe combined immunodeficiency Athabascan type	#602450	AR	Immunology
DCLRE1C	*605988	Omenn syndrome / Severe combined immunodeficiency Athabaskan-type	#603554	AR	Immunology
DIABLO	*605219	Deafness autosomal dominant 64	#614152	AD	Otorhinolaryngology

DIAPH1	*602121	Deafness autosomal dominant 1 with or without thrombocytopenia	#124900	AD	Otorhinolaryngology
DNAJC19	*608977	3-methylglutaconic aciduria type V	#610198	AR	Metabolic
DUOX2	*606759	Thyroid dyshormonogenesis 6	#607200	AR	Endocrinology
DUOXA2	*612772	Thyroid dyshormonogenesis 5	#274900	AR	Endocrinology
EPB42	*177070	Spherocytosis type 5	#612690	AR	Hematology
EPS8L2	*614988	Deafness autosomal recessive 106	#617637	AR	Otorhinolaryngology
ESPN	*606351	Deafness autosomal recessive 36	#609006	AR	Otorhinolaryngology
ESPN	*606351	Deafness neurosensory without vestibular involvement autosomal dominant	#609006	AR	Otorhinolaryngology
ESRRB	*602167	Deafness autosomal recessive 35	#608565	AR	Otorhinolaryngology
ETFA	*608053	Glutaric acidemia IIA	#231680	AR	Metabolic
ETFB	*130410	Glutaric acidemia IIB	#231680	AR	Metabolic
ETFDH	*231675	Glutaric acidemia IIC	#231680	AR	Metabolic
EYA4	*603550	Deafness autosomal dominant 10	#601316	AD	Otorhinolaryngology
F8	*300841	Hemophilia A	#306700	XLR	Hematology
F9	* 300746	Hemophilia B	#306900	XLR	Hematology
FAH	*613871	Tyrosinemia type I	#276700	AR	Metabolic
FBP1	*611570	Fructose-16-bisphosphatase deficiency	#229700	AR	Metabolic
FOXI1	*601093	Enlarged vestibular aqueduct	#600791	AR	Otorhinolaryngology
G6PC	*613742	Glycogen storage disease type Ia	#232200	AR	Metabolic
G6PD	*305900	Hemolytic anemia G6PD deficient (Favism)	#300908	XLD/XLR	Hematology
GAA	*606800	Glycogen storage disease II - Pompe disease	#232300	AR	Metabolic
GALC	*606890	Krabbe disease	#245200	AR	Metabolic
GALE	*606953	Galactose epimerase deficiency	#230350	AR	Metabolic
GALK1	*604313	Galactokinase deficiency with cataracts	#230200	AR	Metabolic
GALT	*606999	Galactosemia	#230400	AR	Metabolic
GCDH	*608801	Glutaricaciduria type I	#231670	AR	Metabolic
GCH1	*600225	Hyperphenylalaninemia BH4-deficient B	#233910	AR	Metabolic

GIPC3	*608792	Deafness autosomal recessive 15	#601869	AR	Otorhinolaryngology
GJB2	*121011	Deafness autosomal dominant 3A	#601544	AD	Otorhinolaryngology
GJB2	*121011	Deafness autosomal recessive 1A	#220290	AR, DD	Otorhinolaryngology
GJB3	*603324	Deafness autosomal dominant 2B	#612644	AD	Otorhinolaryngology
GJB3	*603324	Deafness digenic GJB2/GJB3	#220290	AR, DD	Otorhinolaryngology
GJB6	*604418	Deafness autosomal dominant 3B	#612643	AD	Otorhinolaryngology
GJB6	*604418	Deafness autosomal recessive 1B	# 612645	AR	Otorhinolaryngology
GJB6	*604418	Deafness digenic GJB2/GJB6	# 220290	AR, DD	Otorhinolaryngology
GLA	*300644	Fabry disease	#301500	XLR	enzyme replacement oral chaperone terapy
GLIS3	*610192	Diabetes mellitus neonatal with congenital hypothyroidism	# 610199	AR	Endocrinology
GNMT	*606628	Glycine N-methyltransferase deficiency	#606664	AR	Metabolic
GRAP	*604330	Deafness autosomal recessive 114	#618456	AR	Otorhinolaryngology
GREB1L	*617782	Deafness autosomal dominant 80	#619274	AD	Otorhinolaryngology
GRHL2	*608576	Deafness autosomal dominant 28	#608641	AD	Otorhinolaryngology
GRHPR	*604296	Hyperoxaluria primary type II	#260000	AR	Nephrology
GRXCR1	*613283	Deafness autosomal recessive 25	#613285	AR	Otorhinolaryngology
GSDME	*608798	Deafness autosomal dominant 5	#600994	AD	Otorhinolaryngology
GYS2	*138571	Glycogen storage disease 0 liver	#240600	AR	Metabolic
HADH	*601609	Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency	#231530	AR	Metabolic
HADHA	*600890	Long-chain hydroxyacyl-CoA dehydrogenase deficiency	#609016	AR	Metabolic
HADHB	*143450	Trifunctional protein deficiency	# 609015	AR	Metabolic
HAX1	*605998	Neutropenia severe congenital 3 autosomal recessive	# 610738	AR	Immunology
HBB	*141900	Sickle cell anemia	#603903	AR	Hematology

HBB	*141900	Beta-Thalassemia	#613985	AR	Hematology
HGF	*142409	Deafness autosomal recessive 39	#608265	AR	Otorhinolaryngology
HLCS	*609018	Holocarboxylase synthetase deficiency	#253270	AR	Metabolic
HMGCL	*613898	HMG-CoA lyase deficiency o 3-Hydroxy-3-methyglutaric aciduria	#246450	AR	Metabolic
HMGCS2	*600234	HMG-CoA synthase-2 deficiency	#605911	AR	Metabolic
HOGA1	*613597	Hyperoxaluria primary type III	#613616	AR	Metabolic
HPD	*609695	Tyrosinemia type III (HPD causa anche Hawkinsinuria MIM 140350 con trasmissione AD)	#276710	AR	Metabolic
HSD3B2	*613890	Adrenal hyperplasia congenital due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	#201810	AR	Endocrinology
HSD3B7	*607764	Bile acid synthesis defect congenital 1	#607765	AR	Gastroeneterology
IDS	*607764	Mucopolysaccharidosis II	#607765	XLR	Metabolic
IDUA	*252800	Mucopolysaccharidosis type Ih (I-h I-s I-h/s)	#607014	AR	Metabolic
IDUA	*252800	Mucopolysaccharidosis type Ih/s (I-h I-s I-h/s)	#607015	AR	Metabolic
IDUA	*252800	Mucopolysaccharidosis type Is (I-h I-s I-h/s)	#607016	AR	Metabolic
IL2RG	*308380	Severe combined immunodeficiency X-linked (IL2RG causa anche Combined immunodeficiency X-linked moderate OMIM 312863)	#300400	XLR	Immunology
IL7R	*146661	Severe combined immunodeficiency T-cell negative B-cell/natural killer cell- positive type	#608971	AR	Immunology
ILDR1	*609739	Deafness autosomal recessive 42	#609646	AR	Otorhinolaryngology
IVD	*607036	Isovaleric acidemia	#243500	AR	Metabolic
IYD	*612025	Thyroid dyshormonogenesis 4	#274800	AR	Endocrinology
JAK3	*600173	Severe combined immunodeficiency	#600802	AR	Immunology
KARS1	*601421	Deafness autosomal recessive 89	#613916	AR	Otorhinolaryngology

KCNJ10	*602208	Enlarged vestibular aqueduct digenic	#600791	AR	Otorhinolaryngology
KCNQ4	*603537	Deafness autosomal dominant 2A	#600101	AD	Otorhinolaryngology
KITLG	*184745	Deafness autosomal dominant 69 unilateral or asymmetric	#616697	AD	Otorhinolaryngology
LDLR	*606945	Familial hypercholesterolemia	#143890	AD	Metabolic
LHFPL5	*609427	Sordità autosomica recessiva 67	#610265	AR	Otorhinolaryngology
LMBRD1	*612625	Methylmalonic aciduria and homocystinuria cblF type	#277380	AR	Metabolic
LMX1A	*600298	Deafness autosomal dominant 7	#601412	AD	Otorhinolaryngology
LOXHD1	*613072	Deafness autosomal recessive 77	#613079	AR	Otorhinolaryngology
LPL	*609708	Lipoprotein lipase deficiency	#238600	AR	Metabolic
LRTOMT	*612414	Deafness autosomal recessive 63	#611451	AR	Otorhinolaryngology
MARVELD2	*610572	Deafness autosomal recessive 49	#610153	AR	Otorhinolaryngology
MAT1A	*610550	Hypermethioninemia due to methionine adenosyltransferase I/III deficiency	#250850	AR	Metabolic
MCCC1	*609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency	#210200	AR	Metabolic
MCCC2	*609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	#210210	AR	Metabolic
MCEE	*608419	Methylmalonyl-CoA epimerase deficiency	#251120	AR	Metabolic
MIR96	*611606	Deafness autosomal dominant 50	#613074	AD	Otorhinolaryngology
MLYCD	*606761	Malonyl-CoA decarboxylase deficiency	#248360	AR	Metabolic
MMAA	*607481	Methylmalonic aciduria vitamin B12-responsive	#251100	AR	Metabolic
MMAB	*607568	Methylmalonic aciduria vitamin B12-responsive due to defect in synthesis of adenosylcobalamin cblB complementation type	#251110	AR	Metabolic
MMACHC	*609831	Methylmalonic aciduria and homocystinuria cblC type	#277400	AR	Metabolic
MMADHC	*611935	Methylmalonic aciduria and homocystinuria cblD type	#277410	AR	Metabolic

MPL	*159530	Congenital amegakaryocytic thrombocytopenia	#254450	AR	Hematology
MPZL2	*604873	Deafness autosomal recessive 111	#618145	AR	Otorhinolaryngology
MSRB3	*613719	Deafness autosomal recessive 74	#613718	AR	Otorhinolaryngology
MTHFR	*607093	Homocystinuria due to MTHFR deficiency	#236250	AR	Metabolic
MTR	*156570	Homocystinuria-megaloblastic anemia cobalamin G type	#250940	AR	Metabolic
MTRR	*602568	Homocystinuria cobalamin E type	#236270	AR	Metabolic
MTTP	*157147	Abetalipoproteinemia	#200100	AR	Metabolic
MUT	*603058	Methylmalonic aciduria mut(0) type	#251000	AR	Metabolic
MYH14	*608568	Deafness autosomal dominant 4A	#600652	AD	Otorhinolaryngology
MYH9	*160775	Deafness autosomal dominant 17	#603622	AD	Otorhinolaryngology
MYO15A	*602666	Deafness autosomal recessive 3	#600316	AR	Otorhinolaryngology
MYO3A	*606808	Deafness autosomal recessive 30	#607101	AR	Otorhinolaryngology
MYO6	*600970	Deafness autosomal dominant 22	#606346	AD	Otorhinolaryngology
MYO6	*600970	Deafness autosomal dominant 22 with hypertrophic cardiomyopathy	#606346	AD	Otorhinolaryngology
MYO6	*600970	Deafness autosomal recessive 37	#607821	AR	Otorhinolaryngology
MYO7A	*276903	Deafness autosomal dominant 11	#601317	AD	Otorhinolaryngology
MYO7A	*276903	Deafness autosomal recessive 2	#600060	AR	Otorhinolaryngology
NADK2	*615787	24-dienoyl-CoA reductase deficiency	#616034	AR	Metabolic
NAGS	*608300	N-acetylglutamate synthase deficiency	#237310	AR	Metabolic
NF1	*613113	Neurofibromatosis type I	#162200	AD	Syndromic
NHEJ1	*611290	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation	#611291	AR	Immunology
NKX2-5	*600584	Hypothyroidism congenital nongoitrous 5	#225250	AD	Endocrinology

NLRP3	*606416	Deafness autosomal dominant 34 with or without inflammation	#617772	AD	Otorhinolaryngology
OPA3	*606580	3-methylglutaconic aciduria type III	#258501	AR	Metabolic
OSBPL2	*606731	Deafness autosomal dominant 67	#616340	AD	Otorhinolaryngology
OTC	*300461	Ornithine transcarbamylase deficiency	#311250	XL	Metabolic
OTOA	*607038	Deafness autosomal recessive 22	#607039	AR	Otorhinolaryngology
OTOF	*603681	Auditory neuropathy autosomal recessive 1	#601071	AR	Otorhinolaryngology
OTOF	*603681	Deafness autosomal recessive 9	#601071	AR	Otorhinolaryngology
OTOG	*604487	Deafness autosomal recessive 18B	#614945	AR	Otorhinolaryngology
OTOGL	*614925	Deafness autosomal recessive 84B	#614944	AR	Otorhinolaryngology
P2RX2	*600844	Deafness autosomal dominant 41	#608224	AD	Otorhinolaryngology
PAH	*612349	Phenylketonuria	#261600	AR	Metabolic
PAX8	*167415	Hypothyroidism congenital due to thyroid dysgenesis or hypoplasia	#218700	AD	Endocrinology
PCBD1	*126090	Hyperphenylalaninemia BH4-deficient D	#264070	AR	Metabolic
PCCA	*232000	Propionic acidemia	#606054	AR	Metabolic
PCCB	*232050	Propionic acidemia	#606054	AR	Metabolic
PCDH15	*605514	Deafness autosomal recessive 23	#609533	AR	Otorhinolaryngology
PDZD7	*612971	Deafness autosomal recessive 57	#618003	AR	Otorhinolaryngology
PJVK	*610219	Deafness autosomal recessive 59	#610220	AR	Otorhinolaryngology
PLS1	*602734	Deafness autosomal dominant 76	#618787	AD	Otorhinolaryngology
PNP	*164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency	#613179	AR	Immunology
PNPT1	*610316	Deafness autosomal recessive 70	#614934	AR	Otorhinolaryngology
POU1F1	*173110	Combined pituitary hormone deficiency 1	#613038	AR	Endocrinology
POU4F3	*602460	Deafness autosomal dominant 15	#602459	AD	Otorhinolaryngology
PPIP5K2	*611648	Deafness autosomal recessive 100	#618422	AR	Otorhinolaryngology

PRF1	*170280	Hemophagocytic lymphohistiocytosis familial 2	#603553	AR	Immunology
PRODH	*606810	Hyperprolinemia type I	#239500	AR	Metabolic
PROP1	*601538	Combined pituitary hormone deficiency 2	#262600	AR	Endocrinology
PTPRC	*151460	Severe combined immunodeficiency PTPRC-related	#608971	AR	Immunology
PTPRQ	*603317	Deafness autosomal dominant 73	#617663	AD	Otorhinolaryngology
PTPRQ	*603317	Deafness autosomal recessive 84A	#613391	AR	Otorhinolaryngology
PTS	*612719	Hyperphenylalaninemia BH4-deficient A	#261640	AR	Metabolic
QDPR	*612676	Hyperphenylalaninemia BH4-deficient C	#261630	AR	Metabolic
RAG1	*179615	Combined cellular and humoral immune defects with granulomas	#233650	AR	Immunology
RAG1	*179615	Severe combined immunodeficiency B cell-negative	#601457	AR	Immunology
RAG1	*179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion severe cytomegalovirus infection and autoimmunity	#609889	AR	Immunology
RAG1	*179615	Omenn syndrome and other RAG1-related disorders	#603554	AR	Immunology
RAG2	*179616	Combined cellular and humoral immune defects with granulomas	#233650	AR	Immunology
RAG2	*179616	Severe combined immunodeficiency B cell-negative	#601457	AR	Immunology
RAG2	*179616	Omenn syndrome RAG2-related	#603554	AR	Immunology
RB1	*614041	Retinoblastoma	#180200	AD	Oncology
RDX	*179410	Deafness autosomal recessive 24	#611022	AR	Otorhinolaryngology
REST	*600571	Deafness autosomal dominant 27	#612431	AD	Otorhinolaryngology
RIPOR2	*611410	Deafness autosomal dominant 21	#607017	AD	Otorhinolaryngology
S1PR2	*605111	Deafness autosomal recessive 68	#610419	AR	Otorhinolaryngology

SIX1	*601205	Deafness autosomal dominant 23	#605192	AD	Otorhinolaryngology
SLC12A2	*600840	Deafness autosomal dominant 78	#619081	AD	Otorhinolaryngology
SLC17A8	*607557	Deafness autosomal dominant 25	#605583	AD	Otorhinolaryngology
SLC22A5	*603377	Carnitine deficiency systemic primary	#212140	AR	Metabolic
SLC25A13	*603859	Citrullinemia type II adult-onset	#603471	AR	Metabolic
SLC25A13	*603859	Citrullinemia type II neonatal-onset	#605814	AR	Metabolic
SLC25A20	*613698	Carnitine-acylcarnitine translocase deficiency	#212138	AR	Metabolic
SLC26A4	*605646	Pendred syndrome/Nonsyndromic Enlarged Vestibular Aqueduct	#274600	AR	Otorhinolaryngology
SLC26A4	*605646	Deafness autosomal recessive 4 with enlarged vestibular aqueduct	#600791	AR	Otorhinolaryngology
SLC37A4	*602671	Glycogen storage disease Ib	#232220	AR	Metabolic
SLC3A1	*104614	Cystinuria	#220100	AR	Nephrology
SLC4A1	*109270	Spherocytosis type 4	#612653	AD	Hematology
SLC4A1	*109270	Renal tubular acidosis distal 1 (DRTA1)	#179800	AD	Nephrology
SLC5A5	*601843	Thyroid dyshormonogenesis 1	#274400	AR	Endocrinology
SLC7A7	*603593	Lysinuric protein intolerance	#222700	AR	Metabolic
SLC7A9	*604144	Cystinuria	#220100	AR	Nephrology
SMARCA4	*603254	Rhabdoid tumor predisposition syndrome 2 (RTPS2)	#613325	AD	Oncology
SMARCB1	*601607	Rhabdoid tumor predisposition syndrome 1	#609322	AD	Oncology
SMN1	*600354	Spinal muscular atrophy-1	#253300	AR	Neurology
SMN2	*600354	Spinal muscular atrophy-2	#253550	AR	Neurology
SMN3	*600354	Spinal muscular atrophy-3	#253400	AR	Neurology
SMN4	*600354	Spinal muscular atrophy-4	#271150	AR	Neurology
SPATA5L1	*619578	Deafness autosomal recessive 119	#619615	AR	Otorhinolaryngology
SPTA1	*182860	Spherocytosis type 3	#270970	AR	Hematology
SPTB	*182870	Spherocytosis type 2	#616649	AD	Hematology

STRC	*606440	Deafness autosomal recessive 16	#603720	AR	Otorhinolaryngology
STX11	*605014	Hemophagocytic lymphohistiocytosis familial 4	#603552	AR	Immunology
SYNE4	*615535	Deafness autosomal recessive 76	#615540	AR	Otorhinolaryngology
TAT	*613018	Tyrosinemia type II	#276600	AR	Metabolic
TAZ	*300394	3-methylglutaconic aciduria type II - Barth syndrome	#302060	XLR	Metabolic
TBC1D24	*613577	Deafness autosomal dominant 65	#616044	AD	Otorhinolaryngology
TBC1D24	*613577	Deafness autosomal recessive 86	#614617	AR	Otorhinolaryngology
TECTA	*602574	Deafness autosomal dominant 8/12	#601543	AD	Otorhinolaryngology
TECTA	*602574	Deafness autosomal recessive 21	#603629	AR	Otorhinolaryngology
TG	*612025	Thyroid dyshormonogenesis 3	#274800	AR	Endocrinology
THRA	*190120	Congenital nongoitrous hypothryoidism 6	#614450	AD	Endocrinology
TMC1	*606706	Deafness autosomal dominant 36	#606705	AD	Otorhinolaryngology
TMC1	*606706	Deafness autosomal recessive 7	#600974	AR	Otorhinolaryngology
TMEM132E	*616178	Deafness autosomal recessive 99	#618481	AR	Otorhinolaryngology
TMIE	*607237	Deafness autosomal recessive 6	#600971	AR	Otorhinolaryngology
TMPRSS3	*605511	Deafness autosomal recessive 8/10	#601072	AR	Otorhinolaryngology
TNC	*187380	Deafness autosomal dominant 56	#615629	AD	Otorhinolaryngology
TPO	*606765	Thyroid dyshormonogenesis 2	#274500	AR	Endocrinology
TPRN	*613354	Deafness autosomal recessive 79	#613307	AR	Otorhinolaryngology
TRHR	*188545	Generalized thyrotropin-releasing hormone resistance	#618573	AR	Endocrinology
TRIOBP	*609761	Deafness autosomal recessive 28	#609823	AR	Otorhinolaryngology
TRMU	*610230	Acute infantile liver failure	#613070	AR	Metabolic
TSHB	*188540	Congenital nongoitrous hypothyroidism 4	#275100	AR	Endocrinology
TSHR	*603372	Hypothyroidism congenital nongoitrous 1	#275200	AR	Endocrinology

UNC13D	*608897	Hemophagocytic lymphohistiocytosis familial 3	#608898	AR	Immunology
USH1C	*605242	Deafness autosomal recessive 18A	#602092	AR	Otorhinolaryngology
WBP2	*606962	Deafness autosomal recessive 107	#617639	AR	Otorhinolaryngology
WFS1	*606201	Deafness autosomal dominant 6/14/38	#600965	AD	Otorhinolaryngology
WHRN	*607928	Deafness autosomal recessive 31	#607084	AR	Otorhinolaryngology
WT1	*607102	Wilms tumor type 1 and other WT1-related disorders	#194070	AD	Oncology
ZAP70	*176947	Immunodeficiency 48	#269840	AR	Immunology