Supplementary Table 1: BabyScreen+ gene panel

Gene Name	Model_Of_Inheritance	Phenotypes
AAAS	BIALLELIC	Achalasia-addisonianism-alacrimia syndrome, MIM#231550
ABCC6	BIALLELIC	Arterial calcification, generalized, of infancy, 2, #MIM614473
ABCC8	BOTH monoallelic and biallelic	Hyperinsulinemic hypoglycemia, familial, MIM#256450
ABCD1	X-LINKED: hemizygous mutation in males	Adrenoleukodystrophy, MIM# 300100
ABCD4	BIALLELIC	Methylmalonic aciduria and homocystinuria, cblJ type MIM#614857
ABCG5	BIALLELIC	Sitosterolaemia 2, MIM# 618666
ACAD9	BIALLELIC	Mitochondrial complex I deficiency, nuclear type 20, MIM#611126
ACADM	BIALLELIC	Medium chain acyl CoA dehydrogenase deficiency, MIM#201450
ACADVL	BIALLELIC	VLCAD deficiency, MIM#201475
ACAT1	BIALLELIC	Alpha-methylacetoacetic aciduria, MIM#203750
ACTA2	BOTH monoallelic and biallelic	Aortic aneurysm, familial thoracic 6, MIM# 611788
ACVRL1	MONOALLELIC	Telangiectasia, hereditary hemorrhagic, type 2, MIM#600376
ADA	BIALLELIC	Severe combined immunodeficiency due to ADA deficiency, MIM# 102700, MONDO:0007064
ADA2	BIALLELIC	Vasculitis, autoinflammation, immunodeficiency, and haematologic defects syndrome, MIM# 615688
ADAMTS13	BIALLELIC	Thrombotic thrombocytopenic purpura, familial, MIM#274150
ADGRV1	BIALLELIC	Usher syndrome, type 2C, MIM# 605472
AGL	BIALLELIC	Glycogen storage disease IIIa, MIM#232400
AGRN	BIALLELIC	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, MIM# 615120
AGXT	BIALLELIC	Hyperoxaluria, primary, type 1, MIM# 259900, MONDO:0009823
AHCY	BIALLELIC	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, MIM#613752
AICDA	BIALLELIC	Immunodeficiency with hyper-IgM, type 2, MIM#605258
AIRE	BIALLELIC	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, MIM#240300
AK2	BIALLELIC	Reticular dysgenesis, MIM# 267500;MONDO:0009973
AKR1D1	BIALLELIC	Bile acid synthesis defect, congenital, 2
ALDH4A1	BIALLELIC	Hyperprolinemia, type II MIM#239510
ALDH7A1	BIALLELIC	Epilepsy, pyridoxine-dependent, MIM#266100
ALDOB	BIALLELIC	Fructose intolerance, hereditary, MIM# 229600
ALPL	BIALLELIC	Hypophosphatasia, childhood OMIM#241510;Hypophosphatasia, infantile OMIM#241500
AMACR	BIALLELIC	Bile acid synthesis defect, congenital, 4, MIM# 214950
AMN	BIALLELIC	Megaloblastic anemia-1, Norwegian type, MIM#618882
AP3B1	BIALLELIC	Hermansky-Pudlak syndrome 2, MIM# 608233 MONDO:0011997
AQP2	BOTH monoallelic and biallelic	Diabetes insipidus, nephrogenic, 2, MIM#125800
ARG1	BIALLELIC	Arginase deficiency, MIM#207800
ARPC1B	BIALLELIC	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, MIM#617718
ARSA	BIALLELIC	Metachromatic leukodystrophy, MIM# 250100
ARSB	BIALLELIC	Mucopolysaccharidosis VI (MPS6, MIM# 253200
ASL	BIALLELIC	Argininosuccinic aciduria, MIM#207900
ASS1	BIALLELIC	Citrullinaemia, MIM#215700
ATP6V0A4	BIALLELIC	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, MIM3602722
ATP6V1B1	BIALLELIC	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, MIM# 267300
ATP7A	X-LINKED: hemizygous mutation in males	Menkes disease, MIM# 309400
ATP7B	BIALLELIC	Wilson disease MIM#277900
AVP	MONOALLELIC	Diabetes insipidus, neurohypophyseal MIM#125700
AVPR2	X-LINKED: hemizygous mutation in males	Diabetes insipidus, nephrogenic, MIM#304800
BCHE	BIALLELIC	Butyrylcholinesterase deficiency, MIM#617936
BCKDHA	BIALLELIC	Maple syrup urine disease, type Ia, MIM# 248600
BCKDHB	BIALLELIC	Maple syrup urine disease, type Ib, MIM# 248600
BCKDK	BIALLELIC	Branched-chain keto acid dehydrogenase kinase deficiency, MIM#614923
BLNK	BIALLELIC	Agammaglobulinaemia 4, MIM#613502

BMP1	BIALLELIC	Osteogenesis imperfecta, type XIII , MIM#614856
BRCA1	BIALLELIC	Fanconi anemia, complementation group S, MIM# 617883
BRCA2	BIALLELIC	Fanconi anaemia, complementation group D1, MIM# 605724
BRIP1	BIALLELIC	Fanconi anaemia, complementation group J, MIM#609054
BSCL2	BIALLELIC	Lipodystrophy, congenital generalized, type 2, MIM# 269700;Berardinelli-Seip lipodystrophy
BSND	BIALLELIC	Bartter syndrome, type 4a, MIM# 602522
BTD	BIALLELIC	Biotinidase deficiency, MIM#253260
BTK	X-LINKED: hemizygous mutation in males	Agammaglobulinemia, X-linked 1, MIM#300755
C17orf62	BIALLELIC	Chronic granulomatous disease 5, autosomal recessive, MIM# 618935
C2	BIALLELIC	C2 deficiency, MIM#217000
C3	BIALLELIC	C3 deficiency, MIM#613779
C5	BIALLELIC	C5 deficiency, MIM#609536
C6	BIALLELIC	C6 deficiency, MIM#612446
C7	BIALLELIC	C7 deficiency, MIM#610102
C8B	BIALLELIC	C8 deficiency, type II, MIM#613789
C9	BIALLELIC	C9 deficiency, MIM#613825
CA12	BIALLELIC	Hyperchlorhidrosis, isolated MIM#143860
CA2	BIALLELIC	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, MIM#259730
CA5A	BIALLELIC	Hyperammonaemia due to carbonic anhydrase VA deficiency, MIM#615751
CABP2	BIALLELIC	Deafness, autosomal recessive 93, MIM# 614899
CACNA1S	MONOALLELIC	Malignant hyperthermia susceptibility 5, MIM# 601887
CAD	BIALLELIC	Developmental and epileptic encephalopathy 50, MIM#616457
CALM3	MONOALLELIC	Long QT syndrome 16, MIM#618782
CARD11	BOTH monoallelic and biallelic	Immunodeficiency 11A, autosomal recessive, MIM# 615206;Immunodeficiency 11B with atopic dermatitis, autosomal dominant, MIM# 617638
CASR	BOTH monoallelic and biallelic	Hypocalcemia, autosomal dominant MIM#601198;Hyperparathyroidism, neonatal MIM#239200
CAV1	BIALLELIC	Lipodystrophy, congenital generalized, type 3, MIM#612526
CAVIN1	BIALLELIC	Lipodystrophy, congenital generalized, type 4, MIM# 613327
CD19	BIALLELIC	Immunodeficiency, common variable, 3, MIM#613493
CD247	BIALLELIC	Immunodeficiency 25, MIM# 610163
CD27	BIALLELIC	CD27-deficiency MIM# 615122
CD3D	BIALLELIC	Immunodeficiency 19, MIM#615617
CD3E	BIALLELIC	Immunodeficiency 18, MIM#615615
CD3G	BIALLELIC	Immunodeficiency 17;CD3 gamma deficient MIM# 615607
CD40	BIALLELIC	Immunodeficiency with hyper-IgM, type 3, MIM#606843
CD40LG	X-LINKED: hemizygous mutation in males	Immunodeficiency, X-linked, with hyper-IgM MIM# 308230
CD55	BIALLELIC	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, MIM# 226300
CD70	BIALLELIC	Lymphoproliferative syndrome 3, MIM# 618261
CD79A	BIALLELIC	Agammaglobulinaemia 3, MIM#613501
CD79B	BIALLELIC	Agammaglobulinaemia 6, MIM#612692
CDC14A	BIALLELIC	Deafness, autosomal recessive 32, with or without immotile sperm, MIM# 608653
CDCA7	BIALLELIC	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, MIM# 616910
CDCA8	BOTH monoallelic and biallelic	Congenital hypothyroidism, MONDO:0018612, CDCA8-related
CDH23	BIALLELIC	Usher syndrome, type 1D (MIM# 601067);Deafness, autosomal recessive 12 (MIM # 601386);Usher syndrome, type 1D/F digenic (MIM #601067)
CDKN1C	MONOALLELIC, imprinted	IMAGe syndrome, MIM# 614732
CEBPE	BIALLELIC	Specific granule deficiency, MIM# 245480
CFD	BIALLELIC	Complement factor D deficiency, MIM#613912
CFH	BIALLELIC	Complement factor H deficiency, MIM# 609814
CFI	BIALLELIC	Complement factor I deficiency MIM#610984
CFP	X-LINKED: hemizygous mutation in males	Properdin deficiency, X-linked, MIM#312060
CFTR	BIALLELIC	Cystic fibrosis, MIM#219700
CHAT	BIALLELIC	Congenital myasthenic syndrome, MIM#254210

CHRNA1	BIALLELIC	Myasthenic syndrome, congenital, 1A, slow-channel, MIM# 601462;Myasthenic syndrome, congenital, 1B, fast-channel , MIM#608930
CHRNB1	BIALLELIC	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, MIM# 616314;Congenital myasthenic syndrome
CHRND	BIALLELIC	Myasthenic syndrome, congenital, 3B, fast-channel, MIM#616322;Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, MIM#616323;Myasthenic syndrome, congenital, 3A, slow-channel, MIM#616321;Multiple pterygium syndrome, lethal type, MIM# 253290;MONDO:0009668
CHRNE	BIALLELIC	Myasthenic syndrome, congenital, 4B, fast-channel, 616324;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, slow-channel congenital, 601462;Myasthenic syndrome, congenital, 4A, slow-channel, 605809
CIB2	BIALLELIC	Deafness, autosomal recessive 48, MIM# 609439
CIITA	BIALLELIC	Bare Lymphocyte Syndrome, type II, complementation group A MIM# 209920
CLCN7	BIALLELIC	Osteopetrosis, autosomal recessive 4, MIM# 611490
CLDN14	BIALLELIC	Deafness, autosomal recessive 29, MIM# 614035
CLPP	BIALLELIC	Perrault syndrome 3, MIM# 614129
COCH	BIALLELIC	Deafness, autosomal recessive 110, MIM# 618094
COL11A1	MONOALLELIC	Stickler syndrome, type II, MIM# 604841
COL11A2	BIALLELIC	Deafness, autosomal recessive 53, MIM# 609706
COL13A1	BIALLELIC	Myasthenic syndrome, congenital, 19, MIM#616720
COL1A1	MONOALLELIC	Osteogenesis imperfecta, type I, MIM#166200
COL1A2	MONOALLELIC	Osteogenesis imperfecta, type II , MIM#166210
COL2A1	MONOALLELIC	Stickler syndrome, type I, MIM# 108300
COL4A3	BIALLELIC	Alport syndrome 2, autosomal recessive, MIM# 203780
COL4A4	BIALLELIC	Alport syndrome 2, autosomal recessive MIM#203780
COL4A5	X-LINKED: hemizygous mutation in males	Alport syndrome 1, X-linked, MIM# 301050
COL9A1	BIALLELIC	Stickler syndrome, type IV, MIM#614134
COL9A2	BIALLELIC	Stickler syndrome, type V, MIM# 614284
COL9A3	BIALLELIC	Stickler syndrome, type VI, MIM# 620022
COLQ	BIALLELIC	Congenital myasthenic syndrome, MIM#603034
COQ2	BIALLELIC	Coenzyme Q10 deficiency, primary, 1, MIM#607426
COQ4	BIALLELIC	Coenzyme Q10 deficiency, primary, 7, MIM#616276
COQ6	BIALLELIC	Coenzyme Q10 deficiency, primary, 6, MIM#614650
COQ8A	BIALLELIC	Coenzyme Q10 deficiency, primary, 4, MIM#612016
CORO1A	BIALLELIC	Immunodeficiency 8 MIM# 615401
CPS1	BIALLELIC	Carbamoylphosphate synthetase I deficiency, MIM#237300
CPT1A	BIALLELIC	Carnitine palmitoyltransferase I deficiency, MIM#255120
CPT2	BIALLELIC	CPT II deficiency, infantile 600649;CPT II deficiency, lethal neonatal 608836;CPT II deficiency, myopathic, stress-induced 255110
CRTAP	BIALLELIC	Osteogenesis imperfecta, type VII, MIM# MIM#610682
CSF3R	BIALLELIC	Neutropenia, severe congenital, 7, autosomal recessive, MIM#617014
CTNS	BIALLELIC	Cystinosis, nephropathic MIM#219800
CTPS1	BIALLELIC	Immunodeficiency 24, MIM#615897
CUBN	BIALLELIC	Megaloblastic anaemia-1, Finnish type, MIM#261100
CUL3	MONOALLELIC	Pseudohypoaldosteronism, type IIE 614496
CXCR4	MONOALLELIC	WHIM syndrome 1, MIM#193670
CYB561	BIALLELIC	Orthostatic hypotension 2, MIM#618182
CYBA	BIALLELIC	Chronic granulomatous disease, MIM#233690
CYBB	X-LINKED: hemizygous mutation in males	Chronic granulomatous disease, MIM#306400
CYP11A1	BIALLELIC	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, MIM#613743
CYP11B1	BIALLELIC	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, MIM#202010
CYP11B2	BIALLELIC	Hypoaldosteronism, congenital, due to CMO I deficiency, MIM#203400;Hypoaldosteronism, congenital, due to CMO II deficiency, MIM#610600
CYP17A1	BIALLELIC	17,20-lyase deficiency, isolated, MIM#202110
CYP21A2	BIALLELIC	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, MIM#201910

CYP27A1	BIALLELIC	Cerebrotendinous xanthomatosis, MIM# 213700
CYP27B1	BIALLELIC	Vitamin D-dependent rickets, type I MIM#264700
CYP2R1	BIALLELIC	Rickets due to defect in vitamin D 25-hydroxylation deficiency MIM#600081
CYP7B1	BIALLELIC	Bile acid synthesis defect, congenital, 3, MIM# 613812
DBT	BIALLELIC	Maple syrup urine disease, MIM#248600
DCLRE1C	BIALLELIC	Severe combined immunodeficiency, Athabascan type MIM# 602450;Omenn syndrome, MIM# 603554
DDC	BIALLELIC	Aromatic L-amino acid decarboxylase deficiency, MIM#608643
DFNB59	BIALLELIC	Deafness, autosomal recessive 59, MIM# 610220
DGAT1	BIALLELIC	Diarrhea 7, protein-losing enteropathy type , MIM#615863
DHCR7	BIALLELIC	Smith-Lemli-Opitz syndrome, MIM#270400
DHFR	BIALLELIC	Megaloblastic anaemia due to dihydrofolate reductase deficiency, MIM# 613839
DICER1	MONOALLELIC	DICER1 syndrome, MONDO:0017288
DLAT	BIALLELIC	Pyruvate dehydrogenase E2 deficiency, MIM#245348
DMP1	BIALLELIC	Hypophosphatemic rickets MIM#241520
DNAJC12	BIALLELIC	Hyperphenylalaninemia, mild, non-BH4-deficient, MIM#617384
DNAJC21	BIALLELIC	Bone marrow failure syndrome 3, MIM#617052
DNASE2	BIALLELIC	Autoinflammatory-pancytopenia syndrome, MIM# 619858
DNMT3B	BIALLELIC	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, MIM# 242860
DOCK2	BIALLELIC	Immunodeficiency 40 MIM# 616433
DOCK8	BIALLELIC	Hyper-IgE syndrome, MIM#243700
DOK7	BIALLELIC	Congenital myasthenic syndrome, MIM# 254300
DPAGT1	BIALLELIC	Congenital disorder of glycosylation, type Ij, MIM# 608093;DPAGT1-CDG MONDO:0011964;Myasthenic syndrome, congenital, 13, with tubular aggregates, MIM# 614750
DUOX2	BIALLELIC	Thyroid dyshormonogenesis 6, MIM# 607200
DUOXA2	BIALLELIC	Thyroid dyshormonogenesis 5, MIM# 274900
ECHS1	BIALLELIC	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency MIM# 616277
EDN3	BIALLELIC	Waardenburg syndrome, type 4B, MIM# 613265
EDNRB	BOTH monoallelic and biallelic	Waardenburg syndrome, type 4A, MIM# 277580
EFL1	BIALLELIC	Shwachman-Diamond syndrome 2, MIM#617941
EIF2AK3	BIALLELIC	Wolcott-Rallison syndrome, MIM#226980
ELANE	MONOALLELIC	Neutropenia, congenital, MIM#202700
ENG	MONOALLELIC	Telangiectasia, hereditary hemorrhagic, type 1 MIM#187300
ENPP1	BIALLELIC	Arterial calcification, generalized, of infancy, 1, MIM# 208000;Hypophosphatemic rickets, autosomal recessive, 2, MIM# 613312
EPS8	BIALLELIC	Autosomal recessive nonsyndromic hearing loss 102, MIM#600205, MONDO:0014428
ERCC4	BIALLELIC	Fanconi anemia, complementation group Q, MIM# 615272
ESPN	BIALLELIC	Deafness, autosomal recessive 36, MIM# 609006
ESRRB	BIALLELIC	Deafness, autosomal recessive 35, MIM#608565
ETFA	BIALLELIC	Glutaric acidaemia IIA, MIM#231680
ETFB	BIALLELIC	Glutaric acidemia IIB, MIM#231680
ETFDH	BIALLELIC	Glutaric acidemia IIC, MIM#231680
ETHE1	BIALLELIC	Ethylmalonic encephalopathy, MIM#602473
F10	BIALLELIC	Factor X deficiency, MIM#227600
F13A1	BIALLELIC	Factor XIIIA deficiency, MIM#613225
F13B	BIALLELIC	Factor XIIIB deficiency, MIM#613235
F7	BIALLELIC	Factor VII deficiency MIM# 227500
F9	X-LINKED: hemizygous mutation in males	Haemophilia B, MIM#306900
FAH	BIALLELIC	Tyrosinaemia, type I, MIM#276700
FAM111A	MONOALLELIC	Kenny-Caffey syndrome, type 2, MIM# 127000
FANCA	BIALLELIC	Fanconi anaemia, complementation group A, MIM# 227650;MONDO:0009215
FANCB	X-LINKED: hemizygous mutation in males	Fanconi anaemia, complementation group B, MIM# 300514
FANCC	BIALLELIC	Fanconi anemia, complementation group C, MIM# 227645;MONDO:0009213
FANCD2	BIALLELIC	Fanconi anaemia, complementation group D2, MIM# 227646;MONDO:0009214
FANCG	BIALLELIC	Fanconi anaemia, MIM#614082

FANCI	BIALLELIC	Fanconi anaemia, MIM#609053
FBN1	MONOALLELIC	Marfan syndrome, MIM# 154700
FBP1	BIALLELIC	Fructose-1,6-bisphosphatase deficiency MIM# 229700
FCHO1	BIALLELIC	Immunodeficiency 76, MIM# 619164
FECH	BIALLELIC	Protoporphyria, erythropoietic, 1, MIM#177000
FERMT3	BIALLELIC	Leukocyte adhesion deficiency, type III, MIM#612840
FGA	BIALLELIC	Afibrinogenemia, congenital (MIM#202400)
FGB	BIALLELIC	Afibrinogenaemia, congenital, MIM# 202400
FGF23	BOTH monoallelic and biallelic	autosomal dominant hypophosphatemic rickets MONDO:0008660;familial hyperphosphatemic tumoral calcinosis/hyperphosphatemic hyperostosis syndrome MONDO:0100251
FGF3	BIALLELIC	Deafness, congenital with inner ear agenesis, microtia, and microdontia, MIM# 610706
FGFR3	MONOALLELIC	Achondroplasia MONDO:0007037
FGG	BIALLELIC	Afibrinogenemia, congenital, MIM# 202400
FH	BIALLELIC	Fumurase deficiency MIM# 606812
FKBP10	BIALLELIC	Osteogenesis imperfecta, type XI, OMIM:610968
FLAD1	BIALLELIC	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, MIM#255100
FOLR1	BIALLELIC	Neurodegeneration due to cerebral folate transport deficiency, MIM# 613068
FOXA2	MONOALLELIC	Hyperinsulinism MONDO:0002177
FOXE1	BIALLELIC	Bamforth-Lazarus syndrome MIM# 241850
FOXN1	BOTH monoallelic and biallelic	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, autosomal recessive MIM# 601705;T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, MIM#t 618806
FOXP3	X-LINKED: hemizygous mutation in males	IPEX syndrome, MIM#304790
FUCA1	BIALLELIC	Fucosidosis, MIM# 230000
G6PC	BIALLELIC	Glycogen storage disease Ia, MIM#232200
G6PC3	BIALLELIC	Neutropaenia, congenital, MIM#612541
G6PD	X-LINKED: hemizygous mutation in males	Glucose-6-phosphate dehydrogenase deficiency, MIM#300908
GAA	BIALLELIC	Glycogen storage disease II, Pompe disease, MIM# 232300
GALC	BIALLELIC	Krabbe disease, MIM#245200
GALE	BIALLELIC	Galactose epimerase deficiency, MIM#230350
GALK1	BIALLELIC	Galactokinase deficiency with cataracts, MIM#230200
GALM	BIALLELIC	Galactosemia IV MIM#618881
GALNS	BIALLELIC	Mucopolysaccharidosis IVA, MIM#253000
GALNT3	BIALLELIC	Tumoral calcinosis, hyperphosphatemic, familial, 1, MIM# 211900
GALT	BIALLELIC	Galactosaemia, MIM#230400
GAMT	BIALLELIC	Cerebral creatine deficiency syndrome 2, MIM#612736
GATA2	MONOALLELIC	Immunodeficiency 21 MIM# 614172;Emberger syndrome MIM# 614038
GATA3	MONOALLELIC	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, MIM# 146255
GATA4	MONOALLELIC	Neonatal diabetes mellitus, MONDO:0016391, GATA4-related
GATM	BIALLELIC	Cerebral creatine deficiency syndrome 3 MIM#612718
GBA	BIALLELIC	Gaucher disease type 1, MIM#230800
GCDH	BIALLELIC	Glutaric aciduria, type I, MIM#231670
GCH1	BOTH monoallelic and biallelic	Hyperphenylalaninemia, BH4-deficient, B, MIM# 233910;Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, MIM# 128230
GCK	BOTH monoallelic and biallelic	Hyperinsulinemic hypoglycemia, familial, MIM#602485
GCM2	BOTH monoallelic and biallelic	Hyperparathyroidism 4, OMIM #617343;Hypoparathyroidism, familial isolated 2, OMIM #618883
GFI1	MONOALLELIC	Neutropenia, severe congenital 2, autosomal dominant, MIM# 613107
GGCX	BIALLELIC	Vitamin K-dependent clotting factors, combined deficiency of, 1 MIM# 277450
GH1	BOTH monoallelic and biallelic	Growth hormone deficiency, isolated, type IA, MIM# 262400;Growth hormone deficiency, isolated, type II, MIM# 173100;Kowarski syndrome, MIM# 262650
GHR	BOTH monoallelic and biallelic	Growth hormone insensitivity, partial, MIM# 604271;Laron dwarfism, MIM# 262500
GHRHR	BIALLELIC	Growth hormone deficiency, isolated, type IV, MIM# 618157
GIF	BIALLELIC	Intrinsic factor deficiency, MIM#261000

GIPC3	BIALLELIC	Deafness, autosomal recessive 15, MIM# 601869
GJB2	BIALLELIC	Deafness, autosomal recessive 1A, MIM# 220290
GLA	X-LINKED: hemizygous mutation in males	Fabry disease (MIM# 301500)
GLIS3	BIALLELIC	Diabetes mellitus, neonatal, with congenital hypothyroidism MIM#610199
GLRA1	BOTH monoallelic and biallelic	Hyperekplexia, hereditary 1, autosomal dominant or recessive, MIM#149400
GLUD1	MONOALLELIC	Hyperinsulinism, MIM#606762
GNAS	MONOALLELIC	Pseudopseudohypoparathyroidism MIM#612463;Pseudohypoparathyroidism MIM#612462, MIM#603233, MIM#103580
GOT2	BIALLELIC	Developmental and epileptic encephalopathy 82, MIM#618721
GPIHBP1	BIALLELIC	Hyperlipoproteinemia, type 1D MIM#615947;familial chylomicronemia syndrome
GREB1L	MONOALLELIC	Deafness, autosomal dominant 80MIM#619274
GRHPR	BIALLELIC	Hyperoxaluria, primary, type II, MIM# 260000
GRXCR1	BIALLELIC	Deafness, autosomal recessive 25, MIM#613285
GUSB	BIALLELIC	Mucopolysaccharidosis VII, MIM#253220
GYS2	BIALLELIC	Glycogen storage disease 0, liver (MIM#240600)
HADH	BIALLELIC	3-hydroxyacyl-CoA dehydrogenase deficiency, MIM# 231530
HADHA	BIALLELIC	Mitochondrial trifunctional protein deficiency, MIM#609015;LCHAD deficiency, MIM#609016
HADHB	BIALLELIC	Mitochondrial trifunctional protein deficiency, MIM#609015
HAX1	BIALLELIC	Neutropenia, severe congenital 3, autosomal recessive, MIM# 610738;Kostmann syndrome MONDO:0012548
HBB	BIALLELIC	Sickle cell anaemia, MIM# 603903
HELLS	BIALLELIC	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, MIM# 616911
HESX1	BOTH monoallelic and biallelic	Pituitary hormone deficiency, combined, 5, MIM# 182230
HGF	BIALLELIC	Deafness, autosomal recessive 39, MIM# 608265
HIBCH	BIALLELIC	3-hydroxyisobutryl-CoA hydrolase deficiency MIM#250620
HK1	MONOALLELIC	Hyperinsulinism MONDO:0002177, HK1-related
HLCS	BIALLELIC	Holocarboxylase synthetase deficiency, MIM#253270
HMGCL	BIALLELIC	3-hydroxy-3-methylglutaric aciduria, MIM#246450
HOGA1	BIALLELIC	Hyperoxaluria, primary, type III MIM#613616
HSD11B2	BIALLELIC	Apparent mineralocorticoid excess, MIM# 218030;MONDO:0009025
HSD3B2	BIALLELIC	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency MIM# 201810
HSD3B7	BIALLELIC	Bile acid synthesis defect, congenital, 1 MIM#607765
ICOS	BIALLELIC	Immunodeficiency, common variable, 1 MIM# 607594
IDS	X-LINKED: hemizygous mutation in males	Mucopolysaccharidosis II (MPS2, Hunter syndrome) 309900
IDUA	BIALLELIC	Mucopolysaccharidosis type 1, MONDO:0001586
IFITM5	MONOALLELIC	Osteogenesis imperfecta, type V MIM#610967
IGF1	BIALLELIC	Insulin-like growth factor I deficiency, MIM# 608747
IGHM	BIALLELIC	Agammaglobulinaemia 1, MIM#601495
IGLL1	BIALLELIC	Agammaglobulinaemia 2, MIM#613500
IGSF1	X-LINKED: hemizygous mutation in males	Hypothyroidism, central, and testicular enlargement, MIM# 300888
IKBKB	BIALLELIC	Immunodeficiency 15B, MIM#615592
IKZF1	MONOALLELIC	Immunodeficiency, common variable, 13 MIM# 616873
IL10	BIALLELIC	Autoinflammatory syndrome, MONDO:0019751, IL10-related
IL10RA	BIALLELIC	Inflammatory bowel disease 28, early onset, autosomal recessive, MIM# 613148
IL10RB	BIALLELIC	Inflammatory bowel disease 25, early onset, autosomal recessive, MIM#612567
IL1RN	BIALLELIC	Interleukin 1 receptor antagonist deficiency, MIM# 612852
IL21R	BIALLELIC	Immunodeficiency 56, MIM# 615207
IL2RA	BIALLELIC	Immunodeficiency 41 with lymphoproliferation and autoimmunity, MIM# 606367
IL2RB	BIALLELIC	Immunodeficiency 63 with lymphoproliferation and autoimmunity, MIM#618495
IL2RG	X-LINKED: hemizygous mutation in males	Severe combined immunodeficiency, X-linked, MIM#312863
IL36RN	BIALLELIC	Psoriasis 14, pustular, MIM# 614204
IL7R	BIALLELIC	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type MIM#608971

ILDR1	BIALLELIC	Deafness, autosomal recessive 42, MIM# 609646
INS	BOTH monoallelic and biallelic	Diabetes mellitus, insulin-dependent, 2, MIM# 125852;Diabetes mellitus, permanent neonatal 4, MIM# 618858;Maturity-onset diabetes of the young, type 10, MIM# 613370
IRAK4	BIALLELIC	Immunodeficiency 67, MIM#607676
IRF8	BIALLELIC	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, MIM# 226990
IRS4	X-LINKED: hemizygous mutation in males	Hypothyroidism, congenital, nongoitrous, 9, MIM# 301035
ITGA2B	BIALLELIC	Glanzmann thrombasthaenia 1, MIM# 273800
ITGB2	BIALLELIC	Leukocyte adhesion deficiency, MIM#116920
ITGB3	BIALLELIC	Glanzmann thrombasthenia 2, MIM#619267
ITK	BIALLELIC	Lymphoproliferative syndrome 1, MIM#613011
IVD	BIALLELIC	Isovaleric acidemia, MIM#243500
IYD	BIALLELIC	Thyroid dyshormonogenesis 4, MIM# 274800
JAGN1	BIALLELIC	Neutropenia, severe congenital, 6, autosomal recessive, MIM# 616022
JAK3	BIALLELIC	SCID, autosomal recessive, T-negative/B-positive type, MIM#600802
KCNH2	MONOALLELIC	Long QT syndrome 2, MIM# 613688
KCNJ1	BIALLELIC	Bartter syndrome, type 2, 241200
KCNJ11	BOTH monoallelic and biallelic	Diabetes mellitus, transient neonatal, 3 610582;Diabetes, permanent neonatal, with or without neurologic features 606176;Hyperinsulinemic hypoglycemia, familial, 2 601820
KCNJ2	MONOALLELIC	Andersen syndrome MIM#170390
KCNQ1	BOTH monoallelic and biallelic	Jervell and Lange-Nielsen syndrome MIM#220400;Long QT syndrome 1, MIM# 192500
KDELR2	BIALLELIC	Osteogenesis imperfecta 21, MIM# 619131
KLHL3	BOTH monoallelic and biallelic	Pseudohypoaldosteronism, type IID, MIM# 614495
LAMA2	BIALLELIC	Muscular dystrophy, congenital, merosin deficient or partially deficient, MIM# 607855
LAT	BIALLELIC	Immunodeficiency 52, MIM# 617514
LDLR	BIALLELIC	Hypercholesterolemia, familial, 1, MIM# 143890
LEP	BIALLELIC	Obesity, morbid, due to leptin deficiency (MIM#614962)
LEPR	BIALLELIC	Obesity, morbid, due to leptin receptor deficiency (MIM#614963)
LHFPL5	BIALLELIC	Deafness, autosomal recessive 67, MIM# 610265
LHX3	BIALLELIC	Pituitary hormone deficiency, combined, MIM#221750
LHX4	MONOALLELIC	Pituitary hormone deficiency, combined, 4, MIM#262700
LIG1	BIALLELIC	Immunodeficiency 96, MIM#619774
LIG4	BIALLELIC	LIG4 syndrome, MIM# 606593
LIPA	BIALLELIC	Wolman syndrome, MIM#278000
LMBRD1	BIALLELIC	Methylmalonic aciduria and homocystinuria, MIM#277380
LOXHD1	BIALLELIC	Deafness, autosomal recessive 77, MIM# 613079
LPL	BIALLELIC	Lipoprotein lipase deficiency, MIM# 238600
LRBA	BIALLELIC	Immunodeficiency, common variable, 8, with autoimmunity MIM# 614700
LRP5	BIALLELIC	Osteoporosis-pseudoglioma syndrome, MIM# 259770
LRTOMT	BIALLELIC	Deafness, autosomal recessive 63, MIM# 611451
LYST	BIALLELIC	Chediak-Higashi syndrome, MIM#214500
MAFB	MONOALLELIC	Multicentric carpotarsal osteolysis syndrome (MIM#166300)
MAGT1	X-LINKED: hemizygous mutation in males	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia (MIM# 300853)
MALT1	BIALLELIC	Immunodeficiency 12 MIM# 615468
MAN2B1	BIALLELIC	Mannosidosis, alpha-, types I and II, MIM# 248500
MARVELD2	BIALLELIC	Deafness, autosomal recessive 49, MIM# 610153
MC2R	BIALLELIC	Glucocorticoid deficiency, due to ACTH unresponsiveness, MIM#202200
MCEE	BIALLELIC	Methylmalonyl-CoA epimerase deficiency MIM#251120
MEFV	BIALLELIC	Familial Mediterranean fever MIM# 249100
MESD	BIALLELIC	Osteogenesis imperfecta, type XX, MIM#618644
MITF	BOTH monoallelic and biallelic	Waardenburg syndrome, type 2A, MIM# 193510;Deafness
MLH1	BIALLELIC	Mismatch repair cancer syndrome 1, MIM# 276300
MLYCD	BIALLELIC	Malonyl-CoA decarboxylase deficiency, MIM# 248360
MMAA	BIALLELIC	Methylmalonic aciduria, vitamin B12-responsive, MIM#251100
MMAB	BIALLELIC	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, MIM#251110

MMACHC	BIALLELIC	Methylmalonic aciduria and homocystinuria, cblC type, MIM#277400
MMADHC	BIALLELIC	Methylmalonic aciduria and homocystinuria, cblD type, MIM#277410
MNX1	BIALLELIC	Permanent neonatal diabetes mellitus, MONDO:0100164, MNX1-related
MOCS1	BIALLELIC	Molybdenum cofactor deficiency, MIM#252150
MPI	BIALLELIC	Congenital disorder of glycosylation, type Ib, MIM# 602579
MPL	BIALLELIC	Thrombocytopenia, congenital amegakaryocytic, MIM# 604498
MRAP	BIALLELIC	Glucocorticoid deficiency 2, MIM#607398
MSH2	BIALLELIC	Mismatch repair cancer syndrome 2, MIM# 619096
MSH6	BIALLELIC	Mismatch repair cancer syndrome 3, MIM# 619097
MTHFD1	BIALLELIC	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinaemia MIM # 617780
MTR	BIALLELIC	Homocystinuria-megaloblastic anaemia, cblG complementation type, MIM# 250940
MT-RNR1	MITOCHONDRIAL	Aminoglycoside sensitivity
MTRR	BIALLELIC	Methylmalonic aciduria and homocystinuria, MIM#236270
MTTP	BIALLELIC	Abetalipoproteinemia, MIM# 200100
MUSK	BIALLELIC	Congenital myasthenic syndrome, MIM#616325
MUT	BIALLELIC	Methylmalonic aciduria, mut(0) type, MIM#251000
MVK	BIALLELIC	Mevalonic aciduria, MIM# 610377
MYD88	BIALLELIC	Immunodeficiency 68, MIM# 612260
MYH7	BIALLELIC	Cardiomyopathy, hypertrophic, 1, MIM# 192600
MYO15A	BIALLELIC	Deafness, autosomal recessive 3, MIM# 600316
MYO3A	BIALLELIC	Deafness, autosomal recessive 30, MIM:607101
MYO6	BIALLELIC	Deafness, autosomal recessive 37, MIM# 607821
MYO7A	BIALLELIC	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, MIM# 276900
MYSM1	BIALLELIC	Bone marrow failure syndrome 4, MIM#618116
NAGLU	BIALLELIC	Mucopolysaccharidosis type IIIB (Sanfilippo B), MIM# 252920
NAGS	BIALLELIC	N-acetylglutamate synthetase deficiency, MIM#237310
NCF2	BIALLELIC	Chronic granulomatous disease, MIM#233710
NCF4	BIALLELIC	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III MIM#613960
NEUROG3	BIALLELIC	Diarrhoea 4, malabsorptive, congenital, MIM# 610370
NFKBIA	MONOALLELIC	Ectodermal dysplasia and immunodeficiency 2 MIM# 612132
NHEJ1	BIALLELIC	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, MIM#611291
NIPAL4	BIALLELIC	Ichthyosis, congenital, autosomal recessive 6, MIM# 612281
NKX2-1	MONOALLELIC	Choreoathetosis, hypothyroidism, and neonatal respiratory distress MIM#610978
NKX2-5	MONOALLELIC	Atrial septal defect 7, with or without AV conduction defects, MIM#108900
NNT	BIALLELIC	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, MIM#614736
NPC1	BIALLELIC	Niemann-Pick disease type C1, MIM#257220
NPC2	BIALLELIC	Niemann-Pick disease type C2, MIM#607625
NR0B1	X-LINKED: hemizygous mutation in males	Adrenal hypoplasia, congenital (MIM# 300200)
NR3C2	MONOALLELIC	Pseudohypoaldosteronism type I, autosomal dominant, MIM#177735
NR5A1	MONOALLELIC	Adrenocortical insufficiency, (MIM#612964)
OAS1	MONOALLELIC	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinaemia, MIM#618042
OAT	BIALLELIC	Gyrate atrophy of choroid and retina with or without ornithinemia MIM#258870
ORAI1	BIALLELIC	Immunodeficiency 9, MIM# 612782
OTC	X-LINKED: hemizygous mutation in males	Ornithine transcarbamylase deficiency, MIM#311250
OTOA	BIALLELIC	Deafness, autosomal recessive 22, MIM#607039
OTOF	BIALLELIC	Deafness, autosomal recessive 9, MIM#601071
OTOG	BIALLELIC	Deafness, autosomal recessive 18B - MIM#614945
OTOGL	BIALLELIC	Deafness, autosomal recessive 84B, MIM# 614944
OTULIN	BIALLELIC	Autoinflammation, panniculitis, and dermatosis syndrome, MIM# 617099
OTX2	MONOALLELIC	Pituitary hormone deficiency, combined, 6, MIM# 613986
OXCT1	BIALLELIC	Succinyl CoA:3-oxoacid CoA transferase deficiency, MIM#245050
P3H1	BIALLELIC	Osteogenesis imperfecta, type VIII, (MIM# 610915)

PAH	BIALLELIC	Phenylketonuria, MIM#261600
PALB2	BIALLELIC	Fanconi anemia, complementation group N, MIM# 610832
PAX3	MONOALLELIC	Waardenburg syndrome, type 1, OMIM 193500
PAX8	MONOALLELIC	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, MIM#218700
PC	BIALLELIC	Pyruvate carboxylase deficiency, MIM# 266150
PCBD1	BIALLELIC	Hyperphenylalaninemia, BH4-deficient, D, MIM#264070
PCCA	BIALLELIC	Propionic acidaemia, MIM#606054
PCCB	BIALLELIC	Propionicacidaemia, MIM#606054
PCDH15	BIALLELIC	Usher syndrome, type 1F 602083, Deafness, autosomal recessive 23 609533
PCSK9	MONOALLELIC	Hypercholesterolaemia, familial, 3, MIM# 603776
PDHA1	X-LINKED: hemizygous mutation in males	Pyruvate dehydrogenase E1-alpha deficiency, MIM# 312170
PDHB	BIALLELIC	Pyruvate dehydrogenase E1-beta deficiency, MIM#614111
PDHX	BIALLELIC	Lactic acidaemia due to PDX1 deficiency, MIM# 245349
PDP1	BIALLELIC	Pyruvate dehydrogenase phosphatase deficiency, MIM# 608782
PDX1	BIALLELIC	Pancreatic agenesis, MIM# # 260370
PDZD7	BIALLELIC	Deafness, autosomal recessive 57, MIM# 618003;Usher syndrome, type IIC, GPR98/PDZD7 digenic, MIM# 605472
PGM1	BIALLELIC	Congenital disorder of glycosylation, type It, MIM#614921
PGM3	BIALLELIC	Immunodeficiency 23, MIM#615816
PHEX	X-LINKED: hemizygous mutation in males	Hypophosphatemic rickets, X-linked dominant, MIM#307800
PHGDH	BIALLELIC	Phosphoglycerate dehydrogenase deficiency, MIM#601815
PHKA2	X-LINKED: hemizygous mutation in males	Glycogen storage disease, type IXa1 and a2, MIM# 306000
PHKB	BIALLELIC	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive 261750;Glycogen storage disease IXb, MONDO:0009868
PHKG2	BIALLELIC	Glycogen storage disease IXc, MIM# 613027
PIK3CD	BOTH monoallelic and biallelic	Immunodeficiency 14B, autosomal recessive, MIM# 619281;Immunodeficiency 14A, autosomal dominant, MIM# 615513
PIK3R1	BOTH monoallelic and biallelic	Agammaglobulinemia 7, autosomal recessive, MIM# 615214;Immunodeficiency 36, MIM#616005
PKLR	BIALLELIC	Pyruvate kinase deficiency, MIM#266200
PLG	BIALLELIC	Plasminogen deficiency, type I, MIM# 217090
PLPBP	BIALLELIC	Epilepsy, early-onset, vitamin B6-dependent, MIM#617290
PLS3	X-LINKED: hemizygous mutation in males	Bone mineral density QTL18, osteoporosis - MIM#300910
PNP	BIALLELIC	Immunodeficiency due to purine nucleoside phosphorylase deficiency MIM#613179
PNPO	BIALLELIC	Pyridoxamine 5'-phosphate oxidase deficiency, MIM# 610090
POLE	BIALLELIC	IMAGE-I syndrome, MIM#618336
POMC	BIALLELIC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency MIM#609734
POR	BIALLELIC	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, MIM#201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, MIM# 613571
POU1F1	BOTH monoallelic and biallelic	Pituitary hormone deficiency, combined, 1 MIM# 613038
POU3F4	X-LINKED: hemizygous mutation in males	Deafness, X-linked 2, MIM#304400
PPOX	BIALLELIC	Variegate porphyria, childhood-onset, MIM# 620483
PRDX1	Other	Methylmalonic aciduria and homocystinuria, cblC type, digenic MIM#277400
PRF1	BIALLELIC	Haemophagocytic lymphohistiocytosis, familial, 2, MIM#603553
PRKAR1A	MONOALLELIC	Carney complex, type 1, MIM# 160980
PRKDC	BIALLELIC	Immunodeficiency 26, with or without neurologic abnormalities, MIM#615966
PROP1	BIALLELIC	Pituitary hormone deficiency, combined, 2, MIM#262600
PSTPIP1	MONOALLELIC	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, MIM# 604416
PTCH1	MONOALLELIC	Basal cell nevus syndrome, MIM# 109400
PTF1A	BIALLELIC	Pancreatic and cerebellar agenesis, MIM#609069;Pancreatic agenesis 2, MIM#615935
PTPRC	BIALLELIC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive MIM# 608971
PTPRQ	BOTH monoallelic and biallelic	Deafness, autosomal recessive 84A, MIM# 613391;Deafness, autosomal dominant 73, MIM# 617663
PTS	BIALLELIC	Hyperphenylalaninemia, BH4-deficient, A, MIM#261640
PYGL	BIALLELIC	Glycogen storage disease VI, MIM# 232700

QDPR	BIALLELIC	Dihydropteridine reductase deficiency, MIM#261630
RAB27A	BIALLELIC	Griscelli syndrome, MIM#607624
RAC2	MONOALLELIC	Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia MIM# 618986
RAG1	BIALLELIC	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity MIM# 609889;Combined cellular and humoral immune defects with granulomas MIM# 233650;Omenn syndrome MIM# 603554;Severe combined immunodeficiency, B cell-negative MIM# 601457
RAG2	BIALLELIC	Omenn syndrome MIM# 603554;Severe combined immunodeficiency, B cell-negative MIM# 601457;Combined cellular and humoral immune defects with granulomas MIM# 233650
RAPSN	BIALLELIC	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency (MIM#616326)
RASGRP1	BIALLELIC	Immunodeficiency 64 (MIM#618534)
RB1	MONOALLELIC	Retinoblastoma, MIM# 180200
RDX	BIALLELIC	Deafness, autosomal recessive 24, MIM# 611022
REST	MONOALLELIC	{Wilms tumor 6, susceptibility to}, MIM# 616806
RET	MONOALLELIC	Multiple endocrine neoplasia IIB;Multiple endocrine neoplasia IIA
RFX5	BIALLELIC	Bare lymphocyte syndrome, type II, complementation group C MIM# 209920;Bare lymphocyte syndrome, type II, complementation group E MIM# 209920
RFXANK	BIALLELIC	MHC class II deficiency, complementation group B, MIM#209920
RFXAP	BIALLELIC	Bare lymphocyte syndrome, type II, complementation group D MIM# 209920
RMRP	BIALLELIC	Cartilage-hair hypoplasia MIM#250250
RNPC3	BIALLELIC	Pituitary hormone deficiency, combined or isolated, 7, MIM# 618160
RPE65	BIALLELIC	Leber congenital amaurosis 2 MIM#204100;Retinitis pigmentosa 20 MIM#613794
RPL11	MONOALLELIC	Diamond-Blackfan anaemia, MIM#612562
RPL15	MONOALLELIC	Diamond-Blackfan anaemia 12 , MIM#615550
RPL35A	MONOALLELIC	Diamond-Blackfan anaemia 5, MIM#612528
RPL5	MONOALLELIC	Diamond-Blackfan anaemia, MIM#612561
RPS10	MONOALLELIC	Diamond-Blackfan anaemia 9, MIM#613308
RPS17	MONOALLELIC	Diamond-Blackfan anaemia, MIM#612527
RPS19	MONOALLELIC	Diamond-Blackfan anaemia, MIM#105650
RPS24	MONOALLELIC	Diamond-Blackfan anaemia, MIM#610629
RPS26	MONOALLELIC	Diamond-Blackfan anaemia, MM#613309
RPS7	MONOALLELIC	Diamond-Blackfan anaemia 8, MIM#612563
RUNX1	MONOALLELIC	Platelet disorder, familial, with associated myeloid malignancy, MIM# 601399
RYR1	MONOALLELIC	{Malignant hyperthermia susceptibility 1} MIM#145600
RYR2	MONOALLELIC	Arrhythmogenic right ventricular dysplasia 2;Ventricular tachycardia, catecholaminergic polymorphic
S1PR2	BIALLELIC	Deafness, autosomal recessive 68, MIM# 610419
SAMD9	MONOALLELIC	MIRAGE syndrome, MIM#617053
SAMD9L	MONOALLELIC	Ataxia-pancytopenia syndrome, MIM# 159550
SAR1B	BIALLELIC	Chylomicron retention disease, MIM# 246700
SBDS	BIALLELIC	Shwachman-Diamond syndrome, MIM# 260400
SCNN1A	BIALLELIC	Pseudohypoaldosteronism, type I, MIM# 264350
SCNN1B	BIALLELIC	Pseudohypoaldosteronism, type I MIM# 264350
SCNN1G	BIALLELIC	Pseudohypoaldosteronism, type I, MIM#264350
SERPINF1	BIALLELIC	Osteogenesis imperfecta, type VI, MIM# 613982
SERPINH1	BIALLELIC	Osteogenesis imperfecta, type X, MIM#613848
SGPL1	BIALLELIC	Nephrotic syndrome, type 14 MIM#617575
SH2D1A	X-LINKED: hemizygous mutation in males	Lymphoproliferative syndrome, X-linked, 1, MIM# 308240
SI	BIALLELIC	Sucrase-isomaltase deficiency, congenital, MIM#222900
SLC12A1	BIALLELIC	Bartter syndrome, type 1, MIM# 601678
SLC18A2	BIALLELIC	Parkinsonism-dystonia, infantile, 2, MIM# 618049
SLC18A3	BIALLELIC	Myasthenic syndrome, congenital, 21, presynaptic, MIM#617239
SLC19A2	BIALLELIC	Thiamine-responsive megaloblastic anemia syndrome, MIM# 249270
SLC19A3	BIALLELIC	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), MIM# 607483

SLC22A5	BIALLELIC	Carnitine deficiency, systemic primary, MIM# 212140, MONDO:0008919
SLC25A13	BIALLELIC	Citrullinemia, type II, neonatal-onset, MIM# 605814
SLC25A15	BIALLELIC	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, MIM#238970
SLC25A19	BIALLELIC	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), MIM#613710
SLC25A20	BIALLELIC	Carnitine-acylcarnitine translocase deficiency, MIM#212138
SLC25A38	BIALLELIC	Anemia, sideroblastic, 2, pyridoxine-refractory, MIM# 205950
SLC26A3	BIALLELIC	Diarrhoea 1, secretory chloride, congenital, MIM# 214700
SLC26A4	BIALLELIC	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct 600791;Pendred syndrome 274600
SLC26A7	BIALLELIC	Congenital hypothyroidism, MONDO:0018612, SLC26A7-related
SLC2A1	BOTH monoallelic and biallelic	GLUT1 deficiency syndrome 2, childhood onset, 612126;{Epilepsy, idiopathic generalized, susceptibility to, 12}, MIM#614847;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777
SLC30A10	BIALLELIC	Hypermanganesemia with dystonia 1, MIM# 613280
SLC34A3	BIALLELIC	Hypophosphatemic rickets with hypercalciuria, MIM#241530
SLC35A2	X-LINKED: hemizygous mutation in males	Congenital disorder of glycosylation, type IIm, MIM #300896
SLC37A4	BOTH monoallelic and biallelic	Glycogen storage disease Ib, MIM# 232220;Glycogen storage disease Ic, MIM# 232240;Congenital disorder of glycosylation, type IIw, MIM# 619525
SLC39A4	BIALLELIC	Acrodermatitis enteropathica, MIM# 201100
SLC39A7	BIALLELIC	Agammaglobulinaemia 9, autosomal recessive, MIM# 619693
SLC39A8	BIALLELIC	Congenital disorder of glycosylation, type IIn , MIM#16721
SLC46A1	BIALLELIC	Folate malabsorption, hereditary, MIM# 229050
SLC4A1	BIALLELIC	Distal renal tubular acidosis 4 with haemolytic anaemia MIM# 611590
SLC52A2	BIALLELIC	Brown-Vialetto-Van Laere syndrome 2, MIM# 614707
SLC52A3	BIALLELIC	Brown-Vialetto-Van Laere syndrome 1, MIM# 211530
SLC5A1	BIALLELIC	Glucose/galactose malabsorption, MIM# 606824
SLC5A5	BIALLELIC	Thyroid dyshormonogenesis 1, MIM# 274400
SLC5A6	BIALLELIC	Neurodegeneration, infantile-onset, biotin-responsive, MIM# 618973
SLC5A7	BIALLELIC	Myasthenic syndrome, congenital, 20, presynaptic, MIM# 617143
SLC7A7	BIALLELIC	Lysinuric protein intolerance, MIM# 222700
SLITRK6	BIALLELIC	Deafness and myopia MIM#221200
SLX4	BIALLELIC	Fanconi anaemia, complementation group P, MIM# 613951
SMAD2	MONOALLELIC	Loeys-Dietz syndrome 6, MIM# 619656
SMAD3	MONOALLELIC	Loeys-Dietz syndrome 3, MIM# 613795
SMARCD2	BIALLELIC	Specific granule deficiency 2 MIM#617475
SMN1	BIALLELIC	Spinal muscular atrophy type 1, MIM#253300
SMPD1	BIALLELIC	Niemann-Pick disease, type A, MIM# 257200;Niemann-Pick disease, type B, MIM# 607616
SNX10	BIALLELIC	Osteopetrosis, autosomal recessive 8 MIM#615085
SP110	BIALLELIC	Hepatic veno-occlusive disease with immunodeficiency MIM#235550
SPR	BIALLELIC	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, MIM# 612716
SRP54	MONOALLELIC	Neutropenia, severe congenital, 8, autosomal dominant, MIM# 618752
STAR	BIALLELIC	Congenital lipoid adrenal hyperplasia, MIM#201710
STAT1	BIALLELIC	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive MIM#613796
STAT3	MONOALLELIC	Autoimmune disease, multisystem, infantile-onset, 1 MIM# 615952
STIM1	BIALLELIC	Immunodeficiency 10 MIM612783
STK4	BIALLELIC	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations MIM#614868
STX11	BIALLELIC	Haemophagocytic lymphohistiocytosis, familial, 4, MIM#603552
STX16	MONOALLELIC, imprinted	Pseudohypoparathyroidism, type IB MIM#603233
STXBP2	BIALLELIC	Hemophagocytic lymphohistiocytosis, familial, 5, MIM# 613101
SYT2	BIALLELIC	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive MIM#619461
TANGO2	BIALLELIC	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, MIM# 616878
TAT	BIALLELIC	Tyrosinemia, type II, MIM#276600
TBL1X	X-LINKED: hemizygous mutation in males	Hypothyroidism, congenital, nongoitrous, 8 MIM#301033

TBX19	BIALLELIC	Adrenocorticotropic hormone deficiency, MIM#201400
TCF3	BOTH monoallelic and biallelic	Agammaglobulinaemia 8, autosomal dominant, MIM# 616941;Agammaglobulinaemia 8B, autosomal recessive, MIM# 619824
TCIRG1	BIALLELIC	Osteopetrosis, autosomal recessive 1, MIM# 259700
TCN2	BIALLELIC	Transcobalamin II deficiency MIM# 275350
TECTA	BOTH monoallelic and biallelic	Deafness, autosomal recessive 21 603629;Deafness, autosomal dominant 8/12 601543
TF	BIALLELIC	Atransferrinemia MIM#209300
TG	BIALLELIC	Thyroid dyshormonogenesis 3, MIM# 274700
TGFB2	MONOALLELIC	Loeys-Dietz syndrome 4, MIM#614816
TGFB3	MONOALLELIC	Loeys-Dietz syndrome 5 , MIM#615582
TGFBR1	MONOALLELIC	Loeys-Dietz syndrome 1, MIM# 609192
TGFBR2	MONOALLELIC	Loeys-Dietz syndrome 2, MIM# 610168
TH	BIALLELIC	Tyrosine hydroxylase deficiency, MIM#605407
THRA	MONOALLELIC	Hypothyroidism, congenital, nongoitrous, 6, MIM# 614450
TK2	BIALLELIC	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMC1	BIALLELIC	Deafness, autosomal recessive 7 MIM#600974
TMEM38B	BIALLELIC	Osteogenesis imperfecta, type XIV , MIM#615066
TMIE	BIALLELIC	Deafness, autosomal recessive 6 MIM#600971
TMPRSS3	BIALLELIC	deafness, autosomal recessive MIM#601072
TNFRSF11A	BIALLELIC	Osteopetrosis, autosomal recessive 7 - MIM# 612301
TP53	MONOALLELIC	Li-Fraumeni syndrome MIM#151623
TPK1	BIALLELIC	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) MIM#614458
TPO	BIALLELIC	Thyroid dyshormonogenesis 2A MIM#274500
TPP1	BIALLELIC	Ceroid lipofuscinosis, neuronal, 2 MIM#204500 (Batten disease)
TPRN	BIALLELIC	Deafness, autosomal recessive 79, MIM# 613307
TRHR	BIALLELIC	Hypothyroidism, congenital, nongoitrous, 7, MIM# 618573
TRIM28	MONOALLELIC	Wilms tumour, MONDO:0006058, TRIM28-related
TRIOBP	BIALLELIC	Deafness, autosomal recessive 28, MIM#609823
TRMU	BIALLELIC	Liver failure, transient infantile MIM# 613070
TRPM6	BIALLELIC	Hypomagnesemia 1, intestinal MIM#602014
TSHB	BIALLELIC	Hypothyroidism, congenital, nongoitrous 4, MIM#275100
TSHR	BOTH monoallelic and biallelic	Hypothyroidism, congenital, nongoitrous, 1 - MIM#275200
TTPA	BIALLELIC	Ataxia with isolated vitamin E deficiency MIM#277460
TUBB1	MONOALLELIC	Congenital hypothyroidism, MONDO:0018612, TUBB1-related;Macrothrombocytopenia, autosomal dominant, TUBB1-related, OMIM # 613112
UBE2T	BIALLELIC	Fanconi anaemia, complementation group T, MIM# 616435
UGT1A1	BIALLELIC	Crigler-Najjar syndrome, type I, MIM#218800
UMPS	BIALLELIC	Orotic aciduriaMIM#258900
UNC13D	BIALLELIC	Haemophagocytic lymphohistiocytosis, familial, 3, MIM#608898
UROS	BIALLELIC	Porphyria, congenital erythropoietic MIM#263700
USH1C	BIALLELIC	Usher syndrome type 1 MIM#276904
USH1G	BIALLELIC	Usher syndrome type 1 MIM#606943
USH2A	BIALLELIC	Usher Syndrome Type II MIM#276901
VAMP1	BIALLELIC	Myasthenic syndrome, congenital, 25, MIM# 618323
VDR	BIALLELIC	Rickets, vitamin D-resistant, type IIA MIM#277440
VHL	MONOALLELIC	von Hippel-Lindau syndrome MIM#193300
VKORC1	BIALLELIC	Vitamin K-dependent clotting factors, combined deficiency of, 2 MIM#607473
VPS45	BIALLELIC	Neutropenia, severe congenital, 5, autosomal recessive, MIM#615285
WAS	X-LINKED: hemizygous mutation in males	Neutropenia, severe congenital, X-linked , MIM#300299;Thrombocytopaenia, X-linked, MIM# 313900;Wiskott-Aldrich syndrome, MIM# 301000
WDR1	BIALLELIC	Periodic fever, immunodeficiency, and thrombocytopenia syndrome MIM#150550
WDR72	BIALLELIC	Amelogenesis imperfecta, type IIA3, MIM# 613211;Distal RTA MONDO:0015827
WHRN	BIALLELIC	Usher syndrome, type 2D, MIM# 611383;Deafness, autosomal recessive 31, MIM# 607084
WIPF1	BIALLELIC	Wiskott-Aldrich syndrome 2 MIM#614493
WNK1	MONOALLELIC	Pseudohypoaldosteronism 2C (PHA2C), MIM#614492

WNK4	MONOALLELIC	Pseudohypoaldosteronism, type IIB MIM#614491
WT1	MONOALLELIC	Wilms tumor, type 1, MIM#194070
XIAP	X-LINKED: hemizygous mutation in males	Lymphoproliferative syndrome, X-linked, 2, MIM# 300635
XPA	BIALLELIC	Xeroderma pigmentosum, group A MIM#278700
XPC	BIALLELIC	Xeroderma pigmentosum, group C MIM#278720
ZAP70	BIALLELIC	Immunodeficiency MIM#176947