Screen4Care TREAT Panel: Gene List

Symbol	Disease
Cardiological disorders	
CACNA1C	Timothy Syndrome (long QT syndrome)
KCNH2	long QT syndrome, short QT syndrome
KCNQ1	Jervell and Lange-Nielsen syndrome, long QT syndrome
SCN5A	long QT syndrome, Brugada syndrome, dilated cardiomyopathy
Neurologic/neurodegenerative and neuromuscular disorders	
AGRN	congenital myasthenic syndrome
ALDH7A1	pyridoxine-dependent Epilepsy
ALG14	congenital myasthenic syndrome
ALG2	congenital myasthenic syndrome
ARSA	juvenile metachromatic leukodystrophy
ATP7A	Menkes diseases
CHAT	congenital myasthenic syndrome
CHRNA1	congenital myasthenic syndrome
CHRNB1	congenital myasthenic syndrome
CHRND	congenital myasthenic syndrome
CHRNE	congenital myasthenic syndrome
COL13A1	congential myasthenic syndrome
COLQ	congenital myasthenic syndrome
CYP27A1	cerebrotendinous xanthomatosis
DDC	aromatic L-amino acid decarboxylase deficiency
DMD	Duchenne muscular dystrophy
DOK7	congential myasthenic syndrome
DPAGT1	congenital myasthenic syndrome
GCDH	glutaryl-CoA dehydrogenase deficiency
GCH1	DOPA-responsive dystonia
GFPT1	congential myasthenia
GMPPB	congenital myasthenic syndrome
LRP4	congenital myasthenic syndrome
MUSK	congenital myasthenic syndrome
PLPBP	vitamin B6 responsive epilepsy
PNPO	pyridoxal phosphate-responsive seizures
PTS	BH4-deficient hyperphenylalaninemia A
QDPR	dihydropteridine reductase deficiency
RAPSN	congenital myastenic syndrome
SCN4A	congenital myastenic syndrome, hypokalemic periodic paralysis, paramyotonia, congenita of Von Eulenburg, autosomal recessive SCN4A-related myopathy
SLC18A3	congenital myasthenic syndrome
SLC19A3	biotin-responsive basal ganglia disease
SLC25A1	congential myasthenic syndrome
SLC2A1	encephalopathy due to GLUT1 deficiency
SLC52A2	Brown-Vialetto-Van Laere syndrome 2
SLC52A3	Brown-Vialetto-Van Laere syndrome 1
SLC5A7	congenital myasthenic syndrome
SMN1	spinal muscular atrophy
SPR	dopa-responsive dystonia
SYT2	congential myasthenic syndrome

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TH	dopa-responsive dystonia
TPP1	neuronal ceroid lipofuscinosis
VAMP1	congenital myasthenic syndrome
Metabolic	
ABCD4	methylmalonic acidemia with homocystinuria
ACAD8	isobutyryl-CoA dehydrogenase deficiency
ACADM	medium chain acyl-CoA dehydrogenase deficiency
ACADVL	very long chain acyl-CoA dehydrogenase deficiency
ACAT1	beta-ketothiolase deficiency
AGXT	primary hyperoxaluria
ALDOB	hereditary fructose intolerance
ARG1	hyperargininemia
ARSB	mucopolysaccharidosis
ASL	argininosuccinic aciduria
ASS1	citrullinemia
ATP7B	Wilson disease
AUH	3-methylglutaconic aciduria
BCKDHA	maple syrup urine disease
BCKDHB	maple syrup urine disease
BCKDK	branched chain keto acid dehydrogenase kinase defect
BTD	biotinidase deficiency
CBS	classic homocystinuria
COQ2	coenzyme Q10 deficiency
CPS1	carbamoyl phosphate synthetase I deficiency disease
CPT1A	carnitine palmitoyl transferase 1A deficiency
CPT2	carnitine palmitoyltransferase II deficiency
CTNS	cystinosis
CYP27B1	vitamin D-dependent rickets, type 1A
DBT	maple syrup urine disease type 2, maple syrup urine disease
ETFA	multiple acyl-CoA dehydrogenase deficiency
ETFB	multiple acyl-CoA dehydrogenase deficiency
ETFDH	multiple acyl-CoA dehydrogenase deficiency
FAH	tyrosinemia type I
G6PC1	glycogen storage disease
GAA	glycogen storage disease
GALC	Krabbe disease
GALK1	galactokinase deficiency
GALNS	mucopolysaccharidosis
GALT	galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GATM	L-Arginine:glycine amidinotransferase (AGAT) deficiency
GBA1	Gaucher disease
GUSB	mucopolysaccharidosis
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHB	mitochondrial trifunctional protein deficiency
HLCS	holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaric aciduria
HPD	tyrosinemia
IDS	mucopolysaccharidosis

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IDUA	mucopolysaccharidosis (Scheie syndrome)
IVD	isovaleric acidemia
LIPA	lysosomal acid lipase deficiency
LMBRD1	methylmalonic aciduria and homocystinuria
MAN2B1	α-mannosidosis
MLYCD	malonic aciduria
MMAA	methylmalonic aciduria
MMAB	methylmalonic aciduria
MMACHC	methylmalonic aciduria and homocystinuria
MMADHC	methylmalonic aciduria and homocystinuria inborn disorder of cobalamin metabolism and transport
MMUT	methylmalonic aciduria
MPI	MPI-congenital disorder of glycosylation SRD5A3-congenital disorder of glycosylation
MTHFR	homocystinuria
MTR	methylcobalamin deficiency
MTRR	methylcobalamin deficiency
NAGS	hyperammonemia due to N-acetylglutamate synthase deficiency
OTC	ornithine carbamoyltransferase deficiency
PAH	phenylketonuria
PCCA	propionic acidemia
PCCB	propionic acidemia
PDSS2	coenzyme Q10 deficiency
PHKA2	glycogen storage disease
PHKB	glycogen storage disease
PHKG2	glycogen storage disease
PSAT1	phosphoserine aminotransferase deficiency
SLC16A1	ketoacidosis due to monocarboxylate transporter-1 deficiency
SLC22A5	systemic primary carnitine deficiency disease
SLC25A13	citrin deficiency
SLC25A15	ornithine translocase deficiency
SLC25A20	carnitine-acylcarnitine translocase deficiency
SLC37A4	glycogen storage disease
SLC46A1	hereditary folate malabsorption
SLC6A8	creatine transporter deficiency
SLC7A7	lysinuric protein intolerance
SMPD1	Niemann-Pick disease type A
TAT	tyrosinemia
TK2	mitochondrial DNA depletion syndrome
TMEM70	mitochondrial complex V (ATP synthase) deficiency nuclear
Endocrinological disorders	
ABCC8	neonatal diabetes mellitus
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP11B1	congenital adrenal hyperplasia due to 11-β-hydroxylase deficiency
CYP21A2	classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency
DUOX2	thyroid dyshormonogenesis
DUOXA2	congenital hypothyroidism
GCK	monogenic diabetes congenital glucokinase-related hyperinsulinism

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GLUD1	hyperinsulinism-hyperammonemia syndrome
GNAS	Pseudohypoparathyroidism, ACTH-independent macronodular adrenal hyperplasia McCune-Albright syndrome
IGF1	growth delay due to insulin-like growth factor type 1 deficiency
IGSF1	X-linked central congenital hypothyroidism with late-onset testicular enlargement
INS	isolated permanent neonatal diabetes mellitus
KCNJ11	hyperinsulinemic hypoglycemia, monogenic diabetes mellitus
LHX3	non-acquired combined pituitary hormone deficiency with spine abnormalities
PHEX	X-linked Hypophosphataemia
POU1F1	multiple pituitary hormone deficiency
PROP1	combined pituitary hormone deficiency
RET	multiple endocrine neoplasia
SLC5A5	familial thyroid dyshormonogenesis 1
STAR	congenital lipoid adrenal hyperplasia
TG	thyroid dyshormonogenesis
THRA	congenital nongoitrous hypothryoidism
TPO	thyroid dyshormonogenesis
TSHB	isolated thyroid-stimulating hormone deficiency
TSHR	hypothyroidism due to TSH receptor mutations, familial gestational hyperthyroidism
Kidney diseases	
ADA	severe combined immunodeficiency
AGL	glycogen storage disease
ATP6V0A4	distal renal tubular acidosis
ATP6V1B1	distal renal tubular acidosis
COQ6	familial steroid-resistant nephrotic syndrome with sensorineural deafness
COQ8B	nephrotic syndrome
SLC12A3	Gitelman syndrome
SCNN1B	bronchiectasis with or without elevated sweat chloride pseudohypoaldosteronism Liddle syndrome
SCNN1G	Liddle syndrome
Blood and coagulation disorders	
ADAMTS13	congenital thrombotic thrombocytopenic purpura
ELANE	neutropenia
F13A1	FXIII A subunit deficiency
F2	thrombophilia due to thrombin defect congenital prothrombin deficiency
F7	FVII deficiency - bleeding disorder
F8	hemophilia A
F9	hemophilia B
FANCA	Fanconi anemia complementation group A
FANCB	Fanconi anemia complementation group B
FANCC	Fanconi anemia complementation group C
FANCD2	Fanconi anemia complementation group D2
FANCE	Fanconi anemia complementation group E
FANCF	Fanconi anemia complementation group F
FANCG	Fanconi anemia complementation group G
FANCI	Fanconi anemia complementation group I

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FANCL	Fanconi anemia complementation group L
FGA	congenital fibrinogen deficiency
FGB	congenital fibrinogen deficiency
FGG	congenital fibrinogen deficiency
G6PD	nonspherocytic hemolytic anemia
GATA1	GATA1-Related X-Linked cytopenia
HBA1	α-thalassemia
HBA2	familial erythrocytosis
HBB	β-thalassemia sickle cell anemia
PKLR	pyruvate kinase deficiency of red cells
RPL11	Diamond-Blackfan anemia
RPL35A	Diamond-Blackfan anemia
RPL5	Diamond-Blackfan anemia
RPS10	Diamond-Blackfan anemia
RPS17	Diamond-Blackfan anemia
RPS19	Diamond-Blackfan anemia
RPS24	Diamond-Blackfan anemia
RPS26	Diamond-Blackfan anemia
VWF	Type 3 VWD - lack of VWF
Immunological disorders	
ADA	severe combined immunodeficiency
ADA2	vasculitis-autoinflammation-immunodeficiency- hematologic defects syndrome
BTK	Bruton-type agammaglobulinemia
CD40LG	hyper-IgM syndrome
DCLRE1C	severe combined immunodeficiency due to DCLRE1C deficiency
DOCK8	combined immunodeficiency due to DOCK8 deficiency
FERMT3	leukocyte adhesion deficiency 3
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy
FOXP3	immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome
IL2RG	severe combined immunodeficiency
IL7R	immunodeficiency 104
ITGB2	leukocyte adhesion deficiency 1
JAK3	severe combined immunodeficiency
LYST	Chediak-Higashi syndrome
NHEJ1	Cernunnos-XLF deficiency
PIK3CD	autosomal recessive immunodeficiency
PIK3R1	autosomal recessive agammaglobulinemia SHORT syndrome
PRF1	hemophagocytic lymphohistiocytosis
RAB27A	Griscelli syndrome type 2
RAG1	Omenn syndrome recombinase activating gene 1 deficiency
RAG2	recombinase activating gene 2 deficiency
SH2D1A	X-linked lymphoproliferative syndrome
STX11	familial hemophagocytic lymphohistiocytosis
STXBP2	familial hemophagocytic lymphohistiocytosis
UNC13D	familial hemophagocytic lymphohistiocytosis
WAS	Wiskott-Aldrich syndrome x-linked severe congenital neutropenia

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XIAP	X-linked lymphoproliferative disease
ZAP70	combined immunodeficiency
Syndromic	
CFTR	cystic fibrosis
NBN	Nijmegen breakage syndrome
NKX2-1	hereditary progressive chorea without dementia brain-lung-thyroid syndrome
PMM2	congenital disorder of glycosylation SRD5A3-congenital disorder of glycosylation
SBDS	Shwachman-Diamond syndrome 1, Shwachman-Diamond syndrome
Others	
MVK	porokeratosis of Mibelli
RB1	hereditary retinoblastoma
RPE65	Leber congenital amaurosis RPE65-related recessive retinopathy
SLC26A3	congenital secretory chloride diarrhea 1
SLC26A4	autosomal recessive nonsyndromic hearing loss, Pendred syndrome
ACVR1	fibrodysplasia ossificans progressiva
ALPL	infantile hypophosphatasia
CA2	autosomal recessive osteopetrosis
COL1A1	osteogenesis imperfecta
COL1A2	osteogenesis imperfecta
FGFR3	achondroplasia
TCIRG1	autosomal recessive osteopetrosis

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