Congenital phenotypes

Top targets

Orthologous genes

HPO phenotype

Congenital phenotypes		Top targets		Orthologous genes					
GM2−ganglioside accumulation	profound	GM2−gangliosidosis, AB variant	alternatively activated macrophage	GM2A	LOC713375	Gm2a	gm2a		
Histiocytosis	profound	Histiocytosis−lymphadenopathy plus syndrome	alternatively activated macrophage	SLC29A3	SLC29A3	Slc29a3	LOC101887173		
Short telomere length	profound	Dyskeratosis congenita, autosomal dominant 6	T cell	ACD	ACD	Acd			
Spinocerebellar atrophy	profound	Boucher−Neuhauser syndrome	alternatively activated macrophage	PNPLA6		Pnpla6	LOC560986		
Neonatal sepsis	profound	T−B+ severe combined immunodeficiency due to IL−7Ralpha deficiency	chondrocyte	IL7R	IL7R	Il7r			
Profound static encephalopathy	profound	Hypotonia, infantile, with psychomotor retardation and characterist...	fibroblast	UNC80	UNC80	Unc80	unc80	CG18437	
Reduced beta−hexosaminidase activity	profound	Sandhoff disease	extravillous trophoblast	HEXB	HEXB	Hexb	hexa		hex−1
Subependymal giant−cell astrocytoma	profound	Tuberous sclerosis complex	endocrine cell	TSC2	TSC2	Tsc2	tsc2	gig	
Abnormality of amino acid metabolism	profound	Tyrosinemia type 2	epithelial cell	TAT	TAT	Tat	tat	CG1461	tatn−1
Decreased activity of mitochondrial complex I	profound	Isolated complex I deficiency	inflammatory cell	NDUFB10		Ndufb10	ndufb10	Pdsw	CELE_F59C6.5
HbS hemoglobin	severe	Hereditary persistence of fetal hemoglobin−sickle cell disease synd...	erythroid lineage cell	HBG1					
Decreased CSF biopterin level	severe	Alacrimia−choreoathetosis−liver dysfunction syndrome	mature neutrophil	NGLY1	NGLY1	Ngly1	ngly1	Pngl	png−1
Decreased mitochondrial number	severe	DNA2−related mitochondrial DNA deletion syndrome	enterocyte	DNA2	DNA2	Dna2	dna2	CG2990	dna−2
Profound global developmental delay	severe	Developmental and epileptic encephalopathy 101	GABAergic neuron	GRIN1	GRIN1	Grin1		Nmdar1	nmr−1
Increased circulating very long−chain fatty acid concentration	severe	Peroxisome biogenesis disorder 4A (Zellweger)	epithelial cell of proximal tubule	PEX6	PEX6	Pex6	pex6		
Loss of Purkinje cells in the cerebellar vermis	severe	Spinocerebellar ataxia 42	embryonic stem cell	CACNA1G		Cacna1g	cacna1g		
Absence of bactericidal oxidative respiratory burst in phagocytes	severe	Granulomatous Disease, Chronic, Autosomal Recessive, Cytochrome B−N...	monocyte	CYBA	LOC696748	Cyba	cyba		
Defective production of NFKB1−dependent cytokines	severe	Ectodermal dysplasia and immunodeficiency 2	stromal cell	NFKBIA	NFKBIA	Nfkbia		cact	
Progressive spastic paraparesis	severe	Adrenomyeloneuropathy	stratified epithelial cell	ABCD1	ABCD1	Abcd1	abcd1		
Recurrent Aspergillus infections	severe	Autoimmune enteropathy and endocrinopathy−susceptibility to chronic...	monocyte	STAT1	STAT1	Stat1	stat1a		
Cerebellar medulloblastoma	profound	Turcot syndrome with polyposis	epithelial cell	APC		Apc	apc		
Vegetative state	profound	Mitochondrial DNA depletion syndrome 4A (Alpers type)	kidney loop of Henle epithelial cell	POLG	POLG	Polg	polg	tam	
Malnutrition	profound	Adult−onset autosomal dominant leukodystrophy	smooth muscle cell	LMNB1	LMNB1	Lmnb1	lmnb1		lmn−1
Hypoglycemic encephalopathy	profound	Hyperinsulinism due to short chain 3−hydroxylacyl−CoA dehydrogenase...	inflammatory cell	HADH	HADH	Hadh	hadh		
Decerebrate rigidity	profound	Krabbe disease	alternatively activated macrophage	GALC	GALC	Galc	galcb		CELE_C29E4.10
Acute myeloid leukemia	profound	Tumor predisposition syndrome 2	mature neutrophil	MBD4	MBD4	Mbd4			
Pallidal degeneration	profound	Glutaryl−CoA dehydrogenase deficiency	primordial germ cell	GCDH	GCDH	Gcdh		CG9547	CELE_F54D5.7
Cerebral cortical neurodegeneration	profound	Mitochondrial DNA depletion syndrome 4A (Alpers type)	kidney loop of Henle epithelial cell	POLG	POLG	Polg	polg	tam	
Neuronal loss in central nervous system	profound	Mitochondrial complex IV deficiency, nuclear type 2	alternatively activated macrophage	SCO2	LOC720679	Sco2	sco2		
Cerebral vasculitis	profound	Immunodeficiency due to purine nucleoside phosphorylase deficiency	myeloid cell	PNP	LOC710245			CG16758	K02D7.1
Acute myelomonocytic leukemia	severe	Ataxia−pancytopenia syndrome	alternatively activated macrophage	SAMD9L	LOC701572	Samd9l			
B Acute Lymphoblastic Leukemia	severe	Thrombocytopenia 5	hematopoietic stem cell	ETV6	ETV6	Etv6	etv6		
Late−onset spinocerebellar degeneration	severe	Hemolytic anemia due to gamma−glutamylcysteine synthetase deficiency	erythroid progenitor cell	GCLC	GCLC	Gclc	gclc	Gclc	gcs−1
Increased CSF protein concentration	severe	Familial or sporadic hemiplegic migraine	astrocyte	PRRT2		Prrt2			
Progressive leukoencephalopathy	severe	Myoclonus, intractable, neonatal	neuron	KIF5A	KIF5A	Kif5a	kif5aa		
Progressive psychomotor deterioration	severe	Sandhoff disease	extravillous trophoblast	HEXB	HEXB	Hexb	hexa		hex−1
Esophageal carcinoma	severe	Autoimmune enteropathy and endocrinopathy−susceptibility to chronic...	alternatively activated macrophage	STAT1	STAT1	Stat1	stat1a		
Glucocortocoid−insensitive primary hyperaldosteronism	severe	Generalized pseudohypoaldosteronism type 1	taste receptor cell	SCNN1A	SCNN1A	Scnn1a			
Anorexia	severe	Methylmalonic acidemia with homocystinuria, type cblD	mature neutrophil	MMADHC	MMADHC	Mmadhc	zgc:92335		Y76A2B.5
Kernicterus	severe	Crigler−Najjar syndrome type 1	enterocyte	UGT1A1	UGT1A1	Ugt1a1		CG10178	
Necrotizing myopathy	severe	Hereditary myopathy with early respiratory failure	cell of skeletal muscle	TTN	LOC703527	Ttn	ttna		

Non−congenital phenotypes

death
intellectual disability
impaired mobility
blindness
physical malformations
sensory impairments
immunodeficiency
cancer
reduced fertility
congenital onset

Disease

Cell type

Homo sapiens
Macaca mulatta
Mus musculus
Danio rerio
Drosophila melanogaster
Caenorhabditis elegans

Occurrence
never
rarely
often
always

Severity score
33
30
27
24