	Congenital phenotypes				Top targets	Orthologous genes						
	GM2-ganglioside accumulation		П	-	GM2-gangliosidosis, AB variant	alternatively activated macrophage	GM2A	LOC713375	Gm2a	gm2a		
	Histiocytosis	-	Ш	-	Histiocytosis-lymphadenopathy plus syndrome	alternatively activated macrophage	SLC29A3	SLC29A3	Slc29a3	LOC101887173		
	Short telomere length	_		-	Dyskeratosis congenita, autosomal dominant 6	T cell	ACD	ACD	Acd			
	Spinocerebellar atrophy	-	Ш	-	Boucher–Neuhauser syndrome	alternatively activated macrophage	PNPLA6		Pnpla6	LOC560986		
	Neonatal sepsis		p		T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	chondrocyte	IL7R	IL7R	117r			
	Profound static encephalopathy		ofound		Hypotonia, infantile, with psychomotor retardation and characterist	fibroblast	UNC80	UNC80	Unc80	unc80	CG18437	
	Reduced beta-hexosaminidase activity				Sandhoff disease	extravillous trophoblast	HEXB	HEXB	Hexb	hexa		hex-1
	·					·						nex-1
	Subependymal giant-cell astrocytoma		111		Tuberous sclerosis complex	endocrine cell	TSC2	TSC2	Tsc2	tsc2	gig	
	Abnormality of amino acid metabolism $\ensuremath{\mathfrak{Q}}$				Tyrosinemia type 2	epithelial cell	TAT	TAT	Tat	tat	CG1461	tatn-1
	Decreased activity of mitochondrial complex I	-	Ш	1	Isolated complex I deficiency	inflammatory cell	NDUFB10		Ndufb10	ndufb10	Pdsw	CELE_F59C6.5
	O HbS hemoglobin	-		-	Hereditary persistence of fetal hemoglobin–sickle cell disease synd	erythroid lineage cell	HBG1					
	Decreased CSF biopterin level	-		-	Alacrimia-choreoathetosis-liver dysfunction syndrome	mature neutrophil	NGLY1	NGLY1	Ngly1	ngly1	Pngl	png-1
	Decreased mitochondrial number	-		-	DNA2-related mitochondrial DNA deletion syndrome	enterocyte	DNA2	DNA2	Dna2	dna2	CG2990	dna-2
	Profound global developmental delay	-		-	Developmental and epileptic encephalopathy 101	GABAergic neuron	GRIN1	GRIN1	Grin1		Nmdar1	nmr–1
	Increased circulating very long-chain fatty acid concentration	-	sev	-	Peroxisome biogenesis disorder 4A (Zellweger)	epithelial cell of proximal tubule	PEX6	PEX6	Pex6	pex6		
	Loss of Purkinje cells in the cerebellar vermis	-	/ere	-	Spinocerebellar ataxia 42	embryonic stem cell	CACNA1G		Cacna1g	cacna1g		
	Absence of bactericidal oxidative respiratory burst in phagocytes	-		-	Granulomatous Disease, Chronic, Autosomal Recessive, Cytochrome B-	N.monocyte	СҮВА	LOC696748	Cyba	cyba		
	Defective production of NFKB1-dependent cytokines	-		-	Ectodermal dysplasia and immunodeficiency 2	stromal cell	NFKBIA	NFKBIA	Nfkbia		cact	
	Progressive spastic paraparesis	-		-	Adrenomyeloneuropathy	stratified epithelial cell	ABCD1	ABCD1	Abcd1	abcd1		J
	Recurrent Aspergillus infections			-	Autoimmune enteropathy and endocrinopathy–susceptibility to chronic	monocyte	STAT1	STAT1	Stat1	stat1a		
			on-congenital phenotypes									
	Cerebellar medulloblastoma		Ш	-	Turcot syndrome with polyposis	epithelial cell	APC		Арс	apc		7
	Vegetative state	1	profound	1	Mitochondrial DNA depletion syndrome 4A (Alpers type)	kidney loop of Henle epithelial cell	POLG	POLG	Polg	polg	tam	
	Malnutrition			-	Adult-onset autosomal dominant leukodystrophy	smooth muscle cell	LMNB1	LMNB1	Lmnb1	lmnb1		lmn-1
	Hypoglycemic encephalopathy			-	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase	inflammatory cell	HADH	HADH	Hadh	hadh		
	Decerebrate rigidity			-	Krabbe disease	alternatively activated macrophage	GALC	GALC	Galc	galcb	(CELE_C29E4.10
	Acute myeloid leukemia	-		-	Tumor predisposition syndrome 2	mature neutrophil	MBD4	MBD4	Mbd4			
Occurrence	Pallidal degeneration		Ш	-	Glutaryl-CoA dehydrogenase deficiency	primordial germ cell	GCDH	GCDH	Gcdh		CG9547	CELE_F54D5.7
never rarely	Cerebral cortical neurodegeneration			-	Mitochondrial DNA depletion syndrome 4A (Alpers type)	kidney loop of Henle epithelial cell	POLG	POLG	Polg	polg	tam	
often	Neuronal loss in central nervous system	-		-	Mitochondrial complex IV deficiency, nuclear type 2	alternatively activated macrophage	SCO2	LOC720679	Sco2	sco2		
always	Cerebral vasculitis	-		-	Immunodeficiency due to purine nucleoside phosphorylase deficiency	myeloid cell	PNP	LOC710245			CG16758	K02D7.1
Severity score	Acute myelomonocytic leukemia		П	ĺ	Ataxia-pancytopenia syndrome	alternatively activated macrophage	SAMD9L	LOC701572	Samd9l			
33	B Acute Lymphoblastic Leukemia	-		-	Thrombocytopenia 5	hematopoietic stem cell	ETV6	ETV6	Etv6	etv6		
30 27	Late-onset spinocerebellar degeneration	-	111	-	Gamma–Glutamylcysteine synthetase deficiency, hemolytic anemia dueto	erythroid progenitor cell	GCLC	GCLC	Gclc	gclc	Gclc	gcs-1
24	Increased CSF protein concentration		Ш	-	Familial or sporadic hemiplegic migraine	astrocyte	PRRT2		Prrt2			
	Progressive leukoencephalopathy	-		-	Myoclonus, intractable, neonatal	neuron	KIF5A	KIF5A	Kif5a	kif5aa		
	Progressive psychomotor deterioration	-	severe	-	Sandhoff disease	extravillous trophoblast	HEXB	HEXB	Hexb	hexa		hex-1
	Esophageal carcinoma	-		-	Autoimmune enteropathy and endocrinopathy-susceptibility to chronic	alternatively activated macrophage	STAT1	STAT1	Stat1	stat1a		
	Glucocortocoid-insensitive primary hyperaldosteronism	-		-	Generalized pseudohypoaldosteronism type 1	taste receptor cell	SCNN1A	SCNN1A	Scnn1a			
	Anorexia		Ш	-	Methylmalonic acidemia with homocystinuria, type cblD	mature neutrophil	MMADHC	MMADHC	Mmadhc	zgc:92335		Y76A2B.5
	Kernicterus	1		-	Crigler–Najjar syndrome type 1	enterocyte	UGT1A1	UGT1A1	Ugt1a1		CG10178	
	Necrotizing myopathy	1		-	Hereditary myopathy with early respiratory failure	cell of skeletal muscle	TTN	LOC703527	Ttn	ttna		
		degistaling lindess gious elisacy les findes			Disease	ell type	Homo sapiens	Macaca mulatta	Mus musculus	Danio rerio	Drosophila melanogaster	Caenorhabditis elegans
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