Pediatrics

Metabolic Diseases

Single Gene

1	Metabolic Diseases	Gene
1	17-hydroxylation activity deficiency	CYP17A1
1	2-aminoadipic 2-oxoadipic aciduria	DHTKD1
1	2-methylbutyrylglycinuria	ACADSB
1	3-beta-hydroxysteroid dehydrogenase deficiency type 2	HSD3B2
1	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL
1	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	HMGCS2
1	3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH
1	3-methylglutaconic aciduria type 1	AUH
1	3-methylglutaconic aciduria type 3	OPA3
1	3-methylglutaconic aciduria type 5	DNAJC19
1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	SERAC1
1	5-oxoprolinase deficiency	OPLAH
1	6q24-related transient neonatal diabetes mellitus type 1	UPD chr. 6
1	Abetalipoproteinemia	MTTP
1	Acetycholinesterase deficiency	ACHE
1	Acetyl-CoA carboxylase deficiency	ACACA
1	Acyl-CoA medium-chain dehydrogenase deficiency	ACADM
1	Acyl-CoA multiple dehydrogenase deficiency	ETFA
1	Acyl-CoA multiple dehydrogenase deficiency	ETFB
1	Acyl-CoA short-chain dehydrogenase deficiency	ACADS
1	Acyl-CoA very long-chain dehydrogenase deficiency	ACADVL
1	Adenine phosphoribosyltransferase deficiency	APRT
1	Adenylosuccinase deficiency	ADSL

1	Adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2
1	Adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	POR
1	Adrenal hyperplasia due to steroid 11-beta-hydroxylase deficiency	CYP11B1
1	Adrenal hypoplasia	NR0B1
1	Alkaptonuria	HGD
1	Alpha-2-macroglobulin deficiency	A2M
1	Alpha-ketoglutarate dehydrogenase deficiency	OGDH
1	Alpha-methylacyl CoA racemase deficiency	AMACR
1	Aminoacylase deficiency	ACY1
1	AMP deaminase deficiency, erythrocytic	AMPD3
1	Amyloidosis, familial visceral	APOA1
1	Andersen disease	GBE1
1	Anemia dyserythropoietic type 1A	CDAN1
1	Anemia dyserythropoietic type 2	SEC23B
1	Antitrypsin-alpha-1 deficiency	SERPINA1
1	Aplastic anemia	PRF1
1	Aplastic anemia	TERC
1	Aplastic anemia, SBDS related	SBDS
1	Apolipoprotein C-II deficiency	APOC2
1	Apparent mineralocorticoid excess	HSD11B2
1	Arginase deficiency	ARG1
1	Arginine-glycine amidinotransferase deficiency	GATM
1	Argininosuccinic aciduria	ASL
1	Aromatic L-amino acid decarboxylase deficiency	DDC
1	Asparaginesynthetase deficiency	ASNS
1	Aspartylglucosaminuria	AGA
1	Beta-Galactosamide alpha-2,6-Sialyltransferase 2 deficiency	ST6GAL2
1	Beta-ureidopropionase deficiency	UPB1
1	Bile acid malabsorption, primary	SLC10A2

1	Bile acid synthesis defect type 2, congenital	AKR1D1
1	Bile acid synthesis defect type 3, congenital	CYP7B1
1	Bile acid synthesis defect type 4, congenital	AMACR
1	Biotinidase deficiency	BTD
1	Bloom syndrome	BLM
1	Branched-chain aminotransferase 1 deficiency	BCAT1
1	Branched-chain aminotransferase 2 deficiency	BCAT2
1	Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK
1	Bronchiectasis with or without elevated sweat chloride type 2	SCNN1A
1	Butyrylcholinesterase deficiency	BCHE
1	Carbamoylphosphate synthetase I deficiency	CPS1
1	Carnitine deficiency	SLC22A5
1	Carnitine palmitoyltransferase 1A deficiency	CPT1A
1	Carnitine palmitoyltransferase 1B deficiency	CPT1B
1	Carnitine palmitoyltransferase 2 deficiency, infantile	CPT2
1	Carnitine palmitoyltransferase 2 deficiency, lethal neonatal	CPT2
1	Carnitine-acylcarnitine translocase deficiency	SLC25A20
1	Catechol-o-methyltransferase deficiency	COMT
1	Ceroid lipofuscinosis neuronal type 1	PPT1
1	Ceroid lipofuscinosis neuronal type 10	CTSD
1	Ceroid lipofuscinosis neuronal type 11	GRN
1	Ceroid lipofuscinosis neuronal type 2	TPP1
1	Ceroid lipofuscinosis neuronal type 3	CLN3
1	Ceroid lipofuscinosis neuronal type 4	DNAJC5
1	Ceroid lipofuscinosis neuronal type 5	CLN5
1	Ceroid lipofuscinosis neuronal type 6	CLN6
1	Ceroid lipofuscinosis neuronal type 7	MFSD8
1	Ceroid lipofuscinosis neuronal type 8	CLN8
1	Chanarin-Dorfman syndrome	ABHD5

1	Chloramphenicol resistance, MT-RNR2 related	MT-RNR2
1	Cholestasis benign recurrent intrahepatic type 2	ABCB11
1	Cholestasis intrahepatic, of pregnancy, type 3	ABCB4
1	Cholestasis progressive intrahepatic type 1	ATP8B1
1	Cholestasis progressive intrahepatic type 2	ABCB11
1	Cholestasis progressive intrahepatic type 3	ABCB4
1	Cholestasis, benign recurrent intrahepatic	ATP8B1
1	Cholestasis, intrahepatic, of pregnancy, type 1	ATP8B1
1	Cholesteryl ester storage disease	LIPA
1	Chylomicron retention disease	SAR1B
1	Citrin deficiency	SLC25A13
1	Citrullinemia	ASS1
1	CoA-2 4-dienoyl reductase 1 deficiency	DECR1
1	CoA-3-hydroxyacyl dehydrogenase deficiency	HADH
1	CoA-3-methylcrontonyl carboxylase 1 deficiency	MCCC1
1	CoA-3-methylcrontonyl carboxylase 2 deficiency	MCCC2
1	Colchicine resistance	ABCB1
1	Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1
1	Combined malonic and methylmalonic aciduria	ACSF3
1	Combined oxidative phosphorylation deficiency type 1	GFM1
1	Combined oxidative phosphorylation deficiency type 10	MTO1
1	Combined oxidative phosphorylation deficiency type 11	RMND1
1	Combined oxidative phosphorylation deficiency type 12	EARS2
1	Combined oxidative phosphorylation deficiency type 13	PNPT1
1	Combined oxidative phosphorylation deficiency type 14	FARS2
1	Combined oxidative phosphorylation deficiency type 15	MTFMT
1	Combined oxidative phosphorylation deficiency type 16	MRPL44
1	Combined oxidative phosphorylation deficiency type 17	ELAC2
1	Combined oxidative phosphorylation deficiency type 18	SFXN4

1	Combined oxidative phosphorylation deficiency type 19	LYRM4
1	Combined oxidative phosphorylation deficiency type 2	MRPS16
1	Combined oxidative phosphorylation deficiency type 20	VARS2
1	Combined oxidative phosphorylation deficiency type 21	TARS2
1	Combined oxidative phosphorylation deficiency type 22	ATP5A1
1	Combined oxidative phosphorylation deficiency type 23	GTPBP3
1	Combined oxidative phosphorylation deficiency type 24	NARS2
1	Combined oxidative phosphorylation deficiency type 25	MARS2
1	Combined oxidative phosphorylation deficiency type 26	TRMT5
1	Combined oxidative phosphorylation deficiency type 3	TSFM
1	Combined oxidative phosphorylation deficiency type 4	TUFM
1	Combined oxidative phosphorylation deficiency type 5	MRPS22
1	Combined oxidative phosphorylation deficiency type 6	AIFM1
1	Combined oxidative phosphorylation deficiency type 7	C12ORF65
1	Combined oxidative phosphorylation deficiency type 8	AARS2
1	Combined oxidative phosphorylation deficiency type 9	MRPL3
1	Congenital disorder of glycosylation, type Ip	ALG11
1	Congenital disorder of glycosylation, type Iq	SRD5A3
1	Congenital disorder of glycosylation, type Iw	STT3A
1	Coproporphyria	CPOX
1	Coumarin resistance	VKORC1
1	Coumarin/Warfarin resistance due to CYP2C9 variants	CYP2C9
1	Creatine deficiency syndrome X-linked	SLC6A8
1	CYP2C19 related poor drug metabolism	CYP2C19
1	Cystathioninuria	CTH
1	Cystic fibrosis	CFTR
1	Cystic fibrosis, SLC6A14 related	SLC6A14
1	Cystinosis, nephropathic	CTNS
1	Cytochrome P450 deficiency	CYP1A2

1	D-2-hydroxyglutaric aciduria type 1	D2HGDH
1	D-2-hydroxyglutaric aciduria type 2	IDH2
1	D-bifunctional protein deficiency	HSD17B4
1	D-glyceric aciduria	GLYCTK
1	Diabetes insipidus, nephrogenic, X-linked	AVPR2
1	Diabetes insipidus, neurohypophyseal	AVP
1	Diabetes mellitus type 1	INS
1	Diabetes mellitus, insulin-dependent type 20	HNF1A
1	Diabetes mellitus, insulin-resistant with acanthosis nigricans	INSR
1	Diabetes mellitus, neonatal	GLIS3
1	Diabetes mellitus, noninsulin-dependent	AKT2
1	Diabetes mellitus, noninsulin-dependent	KCNJ11
1	Diabetes mellitus, noninsulin-dependent	ABCC8
1	Diabetes mellitus, permanent neonatal	ABCC8
1	Diabetes mellitus, transient neonatal type 2	ABCC8
1	Diabetes, IGF2 related	IGF2
1	Diarrhea type 1, secretory chloride, congenital	SLC26A3
1	Diarrhea type 4, malabsorptive, congenital	NEUROG3
1	Dihydropyrimidine dehydrogenase deficiency	DPYD
1	Dihydropyrimidinuria	DPYS
1	Dimethylglycine dehydrogenase deficiency	DMGDH
1	Dyggve-Melchior-Clausen disease	DYM
1	Efavirenz, poor metabolism of	CYP2B6
1	Enterokinase deficiency	TMPRSS15
1	Erythrocyte lactate transporter defect	SLC16A1
1	Fabry disease	GLA
1	Factor II deficiency	F2
1	Factor V deficiency	F5
1	Factor XIIIB deficiency	F13B

1	Fanconi anemia type A	FANCA
1	Fanconi anemia type B	FANCB
1	Fanconi anemia type C	FANCC
1	Fanconi anemia type D1	BRCA2
1	Fanconi anemia type D2	FANCD2
1	Fanconi anemia type E	FANCE
1	Fanconi anemia type F	FANCF
1	Fanconi anemia type G	FANCG
1	Fanconi anemia type I	FANCI
1	Fanconi anemia type J	BRIP1
1	Fanconi anemia type L	FANCL
1	Fanconi anemia type M	FANCM
1	Fanconi anemia type N	PALB2
1	Fanconi anemia type P	SLX4
1	Fanconi anemia, XRCCR2 related	XRCC2
1	Fanconi-Bickel syndrome	SLC2A2
1	Farber disease	ASAH1
1	Favism, susceptibility to	G6PD
1	Fish eye disease	LCAT
1	Folate malabsorption, hereditary	SLC46A1
1	Fructose intolerance	ALDOB
1	Fructose uptake deficiency, SLC2A5 related	SLC2A5
1	Fructose-1,6-bisphosphatase deficiency	FBP1
1	Fructosuria essential	KHK
1	Fucosidosis	FUCA1
1	Fumarase deficiency	FH
1	GABA-transaminase deficiency	ABAT
1	Galactokinase deficiency	GALK1
1	Galactose epimerase deficiency	GALE

1	Galactosemia	GALT
1	Galactosialidosis	CTSA
1	Gallbladder disease type 1	ABCB4
1	Gaucher disease type 1	GBA
1	Gaucher disease type 2	GBA
1	Gaucher disease type 3	GBA
1	Gaucher disease type 3C	GBA
1	Gaucher disease, atypical	PSAP
1	Gaucher disease, perinatal lethal	GBA
1	Glucocorticoid deficiency type 1	MC2R
1	Glucocorticoid deficiency type 2	MRAP
1	Glucose/Galactose malabsorption	SLC5A1
1	Glutamate formiminotransferase deficiency	FTCD
1	Glutamine deficiency, congenital	GLUL
1	Glutaric acidemia type 1	GCDH
1	Glutaric acidemia type 2C	ETFDH
1	Glutaric aciduria type 3	SUGCT
1	Glutathione S-transferase theta-1 defficiency	GSTT1
1	Glutathione synthetase deficiency	GSS
1	Glycerol kinase deficiency	GK
1	Glycogen storage disease of heart (lethal)	PRKAG2
1	Glycogen storage disease type 0	GYS2
1	Glycogen storage disease type 0 muscle	GYS1
1	Glycogen storage disease type 10	PGAM2
1	Glycogen storage disease type 11	LDHA
1	Glycogen storage disease type 12	ALDOA
1	Glycogen storage disease type 13	ENO3
1	Glycogen storage disease type 14	PGM1
1	Glycogen storage disease type 15	GYG1

1	Glycogen storage disease type 1A	G6PC
1	Glycogen storage disease type 1B	SLC37A4
1	Glycogen storage disease type 1C	SLC37A4
1	Glycogen storage disease type 2	GAA
1	Glycogen storage disease type 3	AGL
1	Glycogen storage disease type 4	GBE1
1	Glycogen storage disease type 5	PYGM
1	Glycogen storage disease type 6B	PYGL
1	Glycogen storage disease type 7	PFKM
1	Glycogen storage disease type 9A	PHKA2
1	Glycogen storage disease type 9B	PHKB
1	Glycogen storage disease type 9C	PHKG2
1	Glycosylation disorder type 1A	PMM2
1	Glycosylation disorder type 1B	MPI
1	Glycosylation disorder type 1C	ALG6
1	Glycosylation disorder type 1D	ALG3
1	Glycosylation disorder type 1E	DPM1
1	Glycosylation disorder type 1F	MPDU1
1	Glycosylation disorder type 1G	ALG12
1	Glycosylation disorder type 1H	ALG8
1	Glycosylation disorder type 1I	ALG2
1	Glycosylation disorder type 1J	DPAGT1
1	Glycosylation disorder type 1K	ALG1
1	Glycosylation disorder type 1L	ALG9
1	Glycosylation disorder type 1M	DOLK
1	Glycosylation disorder type 1N	RFT1
1	Glycosylation disorder type 1O	DPM3
1	Glycosylation disorder type 1S	ALG13
1	Glycosylation disorder type 1U	DPM2

1	Glycosylation disorder type 2A	MGAT2
1	Glycosylation disorder type 2B	MOGS
1	Glycosylation disorder type 2C	SLC35C1
1	Glycosylation disorder type 2D	B4GALT1
1	Glycosylation disorder type 2E	COG7
1	Glycosylation disorder type 2F	SLC35A1
1	Glycosylation disorder type 2G	COG1
1	Glycosylation disorder type 2H	COG8
1	Glycosylation disorder type 2I	COG5
1	Glycosylation disorder type 2J	COG4
1	Glycosylation disorder type 2K	TMEM165
1	Glycosylation disorder type 2M	SLC35A2
1	Glycosylation disorder type 3	COG6
1	Glycosylation disorder type IR	DDOST
1	Glycosylation disorder x-linked	SSR4
1	GM1-gangliosidosis type 1	GLB1
1	GM1-gangliosidosis type 2	GLB1
1	GM2-gangliosidosis type 2	HEXB
1	Guanidinoacetate methyltransferase deficiency	GAMT
1	Hartnup disorder	SLC6A19
1	Hawkinsinuria	HPD
1	HDL deficiency, type 2	ABCA1
1	Hemochromatosis classical	HFE
1	Hemochromatosis type 2A	HFE2
1	Hemochromatosis type 2B	HAMP
1	Hemochromatosis type 3	TFR2
1	Hemochromatosis type 4	SLC40A1
1	Hemolytic anemia due to G6PD deficiency	G6PD
1	Hemophilia A	F8

1	Holocarboxylase synthetase deficiency	HLCS
1	Hurler syndrome	IDUA
1	Hurler-Scheie syndrome	IDUA
1	Hypercalcemia infantile type	CYP24A1
1	Hypercholanemia	BAAT
1	Hypercholanemia	TJP2
1	Hypercholesterolemia autosomal dominant type 3	PCSK9
1	Hypercholesterolemia autosomal recessive	LDLRAP1
1	Hypercholesterolemia due to LDL-receptor-disorder autosomal dominant	LDLR
1	Hypercholesterolemia type B autosomanl dominant	APOB
1	Hyperchylomicronemia type 5	APOA5
1	Hyperinsulinaemia, association with, G6PC2 related	G6PC2
1	Hyperinsulinemic hypoglycemia type 1	ABCC8
1	Hyperinsulinemic hypoglycemia type 2	KCNJ11
1	Hyperinsulinemic hypoglycemia type 3	GCK
1	Hyperinsulinemic hypoglycemia type 6	GLUD1
1	Hyperinsulinemic hypoglycemia type 7	SLC16A1
1	Hyperinsulinism, UCP2 related	UCP2
1	Hyperlipidemia, familial combined, susceptibility to	USF1
1	Hyperlipoproteinemia type 1	LPL
1	Hyperlysinemia type 1	AASS
1	Hypermanganesemia with dystonia, polycythemia and cirrhosis	SLC30A10
1	Hypermethioninemia due to adenosine kinase deficiency	ADK
1	Hyperornithinemia- Hyperammonemia - Homocitrullinuria syndrome	SLC25A15
1	Hyperoxaluria type 1	AGXT
1	Hyperoxaluria type 2	GRHPR
1	Hyperoxaluria type 3	HOGA1
1	Hyperoxaluria, SLC26A6 related	SLC26A6
1	Hyperphenylalaninemia, BH4 deficient, type C	QDPR

1	Hyperphenylalaninemia, BH4 deficient, type D	PCBD1
1	Hyperphenylalaninemia, BH4-deficient, type A	PTS
1	Hyperprolinemia type 1	PRODH
1	Hypertriglyceridemia, susceptibility to	LIPI
1	Hypoaldosteronism congenital due to CMO I deficiency	CYP11B2
1	Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2
1	Hypoalphalipoproteinemia	APOA1
1	Hypobetalipoproteinemia type 1	APOB
1	Hypocalcemia, autosomal dominant 2	GNA11
1	Hypocalciuric hypercalcemia, familial type 3	AP2S1
1	Hypoglycemia of infancy, leucine-sensitive	ABCC8
1	Hypoinsulinemic hypoglycemia with hemihypertrophy	AKT2
1	Hypomagnesemia type 1	TRPM6
1	Hypomagnesemia type 2	FXYD2
1	Hypomagnesemia type 3	CLDN16
1	Hypomagnesemia type 4	EGF
1	Hypomagnesemia type 5	CLDN19
1	Hypomagnesemia type 6	CNNM2
1	Hypophosphatasia, adult	ALPL
1	Hypophosphatasia, childhood	ALPL
1	Hypophosphatasia, infantile	ALPL
1	Hypophosphatemic rickets with hypercalciuria	SLC34A3
1	Hypophosphatemic rickets, autosomal dominant	FGF23
1	Hypouricemia, renal type 1	SLC22A12
1	Hypouricemia, renal type 2	SLC2A9
1	Insulin-like growth factor resistance	IGF1R
1	Isobutyryl-CoA dehydrogenase deficiency	ACAD8
1	Isovaleric acidemia	IVD
1	Krabbe disease	GALC

1	Krabbe disease, atypical	PSAP
1	L-2-hydroxyglutaric aciduria	L2HGDH
1	Lactase deficiency, congenital	LCT
1	Lactate dehydrogenase-B deficiency	LDHB
1	Lacticacidemia due to PDX1 deficiency	PDHX
1	Lactose intolerance, adult type	MCM6
1	LCAD deficiency	ACADL
1	LCAT DEFICIENCY	LCAT
1	Leukocyte adhesion deficiency	ITGB1
1	Leukocyte adhesion deficiency	ITGB2
1	Lipodystrophy generalized type 1	AGPAT2
1	Lipodystrophy generalized type 2	BSCL2
1	Lipodystrophy generalized type 4	PTRF
1	Lipodystrophy type 2, familial partial	LMNA
1	Lipodystrophy, familial partial, type 3	PPARG
1	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA
1	Lung alpha-beta hydrolase deficiency type 1	ABHD1
1	Lysosomal acid phosphatase deficiency	ACP2
1	Malonyl-CoA decarboxylase deficiency	MLYCD
1	Mannose-binding protein deficiency	MBL2
1	Mannosidosis, beta A, lysosomal-like	MANBAL
1	Mannosidosis-alpha	MAN2B1
1	Mannosidosis-beta	MANBA
1	Maple syrup urine disease type 1a	BCKDHA
1	Maple syrup urine disease type 1b	BCKDHB
1	Maple syrup urine disease type 2	DBT
1	Maple syrup urine disease type 3	DLD
1	Maple syrup urine disease, mild variant	PPM1K
1	Maturity-onset diabetes of the young type 1	HNF4A

1	Maturity-onset diabetes of the young type 10	INS
1	Maturity-onset diabetes of the young type 11	BLK
1	Maturity-onset diabetes of the young type 2	GCK
1	Maturity-onset diabetes of the young type 3	HNF1A
1	Maturity-onset diabetes of the young type 4	PDX1
1	Maturity-onset diabetes of the young type 5	HNF1B
1	Maturity-onset diabetes of the young type 6	NEUROD1
1	Maturity-onset diabetes of the young type 7	KLF11
1	Maturity-onset diabetes of the young type 8	CEL
1	Maturity-onset diabetes of the young type 9	PAX4
1	Maturity-onset diabetes of the young, NKX2-2 related	NKX2-2
1	Maturity-onset diabetes of the young, RFX6 related	RFX6
1	Maturity-onset diabetes of the young, ZFP57 related	ZFP57
1	Mediterranean fever	MEFV
1	MELAS syndrome, MT-TL1 related	MT-TL1
1	Metachromatic Leukodystrophy	ARSA
1	Methylacetoacetic aciduria	ACAT1
1	Methylcobalamin deficiency CblG type	MTR
1	Methylmalonate semialdehyde dehydrogenase deficiency	ALDH6A1
1	Methylmalonic aciduria CblA type	MMAA
1	Methylmalonic aciduria CblB type	MMAB
1	Methylmalonic aciduria CblC type	MMACHC
1	Methylmalonic aciduria CblD type	MMADHC
1	Methylmalonic aciduria CblF type	LMBRD1
1	Methylmalonic aciduria CblJ type	ABCD4
1	Methylmalonic aciduria CblR type	CD320
1	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	MUT
1	Methylmalonyl-CoA epimerase deficiency	MCEE
1	Mevalonic aciduria	MVK

1	Microvascular complications of diabetes type 1	VEGFA
1	Microvascular complications of diabetes type 6, susceptibility to	SOD2
1	Mitchell-Riley syndrome	RFX6
1	Mitochondrial complex III deficiency, nuclear type 7	UQCC2
1	Mitochondrial pyruvate carrier deficiency	MPC1
1	Molybdenum cofactor deficiency type A	MOCS1
1	Molybdenum cofactor deficiency type B	MOCS2
1	Molybdenum cofactor deficiency type C	GPHN
1	Monocarboxylate transporter 1 deficiency	SLC16A1
1	Mucolipidosis type 2 alpha/beta	GNPTAB
1	Mucolipidosis type 3	GNPTAB
1	Mucolipidosis type 3 gamma	GNPTG
1	Mucolipidosis type 4	MCOLN1
1	Mucopolysaccharidosis type 2	IDS
1	Mucopolysaccharidosis type 3A	SGSH
1	Mucopolysaccharidosis type 3B	NAGLU
1	Mucopolysaccharidosis type 3C	HGSNAT
1	Mucopolysaccharidosis type 3D	GNS
1	Mucopolysaccharidosis type 4A	GALNS
1	Mucopolysaccharidosis type 4B	GLB1
1	Mucopolysaccharidosis type 6	ARSB
1	Mucopolysaccharidosis type 7	GUSB
1	Mucopolysaccharidosis type 9	HYAL1
1	Mucopolysaccharidosis type IH	IDUA
1	Muscle glycogenosis	PHKA1
1	Myopathy due to myoadenylate deaminase deficiency	AMPD1
1	N-acetylglutamate synthase deficiency	NAGS
1	Neuraminidase deficiency	NEU1
1	Niemann-Pick disease type A/B	SMPD1

1	Niemann-Pick disease type C1	NPC1
1	Niemann-Pick disease type C2	NPC2
1	Odontohypophosphatasia	ALPL
1	Ornithine transcarbamoylase deficiency	OTC
1	Orotic aciduria	UMPS
1	Pancreatic agenesis type 2	PTF1A
1	Pancreatic and cerebellar agenesis	PTF1A
1	Pentosuria	DCXR
1	Periodic fever autosomal dominant	TNFRSF1A
1	Phenylketonuria	PAH
1	Phenylketonuria modifier, SLC7A5 related	SLC7A5
1	Phosphoenolpyruvate carboxykinase deficiency, cytosolic	PCK1
1	Phosphoenolpyruvate carboxykinase deficiency, mitochondrial	PCK2
1	Phosphoglycerate dehydrogenase deficiency	PHGDH
1	Phosphoribosylpyrophosphate synthetase superactivity	PRPS1
1	Phosphoserine aminotransferase deficiency	PSAT1
1	Phosphoserine phosphatase deficiency	PSPH
1	Pituitary stalk interruption syndrome, GPR161 related	GPR161
1	Pompe disease	GAA
1	Porphyria acute intermittent	HMBS
1	Porphyria congenital erythropoietic	UROS
1	Porphyria variegata	PPOX
1	Prolidase deficiency	PEPD
1	Propionic acidemia	PCCB
1	Propionic acidemia	PCCA
1	Prosaposin deficiency	PSAP
1	Protoporphyria, erythropoietic, X-linked	ALAS2
1	Pseudohermaphroditism with gynecomastia	HSD17B3
1	Pyridoxamine 5'-phosphate oxidase deficiency	PNPO

1	Pyruvate carboxylase deficiency	PC
1	Pyruvate dehydrogenase E1-alpha deficiency	PDHA1
1	Pyruvate dehydrogenase E1-beta deficiency	PDHB
1	Pyruvate dehydrogenase E2 deficiency	DLAT
1	Pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS
1	Pyruvate dehydrogenase phosphatase deficiency	PDP1
1	Pyruvate kinase deficiency with hemolytic anemia	PKLR
1	Refsum disease	PEX7
1	Refsum disease	PHYH
1	Riboflavin deficiency	SLC52A1
1	Rickets, vitamin D 25-hydroxylation-deficient, type 1B	CYP2R1
1	Rickets, vitamin D dependent, type 1	CYP27B1
1	Saccharopinuria	AASS
1	Sandhoff disease	HEXB
1	Sarcosinemia	SARDH
1	Scheie syndrome	IDUA
1	Schindler disease	NAGA
1	Serine hydrolase deficiency, SERHL2 related	SERHL2
1	Succinic semialdehyde dehydrogenase deficiency	ALDH5A1
1	Succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1
1	Sucrase-isomaltase deficiency	SI
1	Sulfatase deficiency	SUMF1
1	Sulfite oxidase deficiency	SUOX
1	Surfactant metabolism dysfunction	SFTPD
1	Surfactant metabolism dysfunction type 1	SFTPB
1	Surfactant metabolism dysfunction type 2	SFTPC
1	Surfactant metabolism dysfunction type 3	ABCA3
1	Surfactant metabolism dysfunction type 4	CSF2RA
1	Surfactant metabolism dysfunction type 5	CSF2RB

1	Tangier disease	ABCA1
1	Tay-Sachs disease	HEXA
1	Tay-Sachs disease AB variant	GM2A
1	Thiamine metabolism dysfunction syndrome type 5	TPK1
1	TJP1 deficiency	TJP1
1	TPMT deficiency	TPMT
1	Transaldolase deficiency	TALDO1
1	Transcobalamin II deficiency	TCN2
1	Trifunctional protein deficiency	HADHA
1	Trimethylaminuria	FMO3
1	Triosephosphate isomerase deficiency	TPI1
1	Tyrosine kinase 2 deficiency	TYK2
1	Tyrosinemia type 1	FAH
1	Tyrosinemia type 1B	GSTZ1
1	Tyrosinemia type 2	TAT
1	Tyrosinemia type 3	HPD
1	Urbach-Wiethe disease	ECM1
1	Von-Gierke disease	G6PC
1	Wilson disease	ATP7B
1	Wolman disease	LIPA
1	Xanthinuria type 1	XDH

Panel

	Product name Clinical Exome Whole exome Solo	TAT-day
01	Brain iron accumulation syndromes panel	25
02	Ceroid lipofuscinosis panel	25
03	Congenital glycosylation disease panel	25
04	Diabetes neonatal panel	25
05	Diamond-Blackfan anemia panel	25
06	Familial hypercholesterolemia panel	25
07	Fatty acid oxidation disorder panel	25
08	Glycogen storage disease panel (advanced)	25
09	Glycogen storage disease panel (basic)	25
10	Hyperinsulinemic hypoglycemia panel	25
11	Leigh syndrome and mitochondrial encephalopathy panel	25
12	Lipodystrophy panel	25
13	Lysosomal storage disease panel	25
14	Methylmalonic acidemia panel (advanced)	25
15	Methylmalonic acidemia panel (basic)	25
16	MODY panel	25
17	Mucopolysaccharidosis panel	25
18	Non ketotic hyperglycinemia panel	25
19	Obesity panel	25
20	Refsum disease panel	25
21	Renal tubular acidosis panel	25
22	Surfactant metabolism dysfunction panel	25
23	Urea cycle disorder panel	25
24	Zellweger syndrome panel	25

	Biochemical Genetics Panel	
25	CentoSphingo Enzyme Panel	
26	CentoSphingo Enzyme Panel X-TRA	
27	CentoMPS Enzyme Panel	
28	CentoMPS Enzyme Panel X-TRA	
29	CentoNCL Enzyme Panel	
30	CentoNCL Enzyme Panel X-TRA	
31	CentoLSD Enzyme Panel	
32	CentoLSD Enzyme Panel X-TRA	

Malformation and Retardation Syndromes

Single Gene

	Malformation and Retardation Syndromes	Gene
1	Achondrogenesis type 2	COL2A1
1	Acrodysostosis type 1, with or without hormone resistance	PRKAR1A
1	Acromelic frontonasal dysostosis	ZSWIM6
1	Acromesomelic dysplasia, Maroteaux type	NPR2
1	Acromicric dysplasia	FBN1
1	Adams-Oliver syndrome type 6	DLL4
1	ADULT syndrome, split hand-foot malformation	TP63
1	Alacrima, achalasia and mental retardation syndrome	GMPPA
1	Alagille syndrome type 1	JAG1
1	Alagille syndrome type 2	NOTCH2
1	Alazami syndrome	LARP7
1	Alpha-thalassemia/mental retardation syndrome	ATRX
1	Ankyloblepharon-ectodermal defects-cleft lip/palate	TP63
1	Anterior segment mesenchymal dysgenesis	PITX3
1	Antley-Bixler syndrome	FGFR2
1	Apert syndrome	FGFR2
1	Athabaskan brainstem dysgenesis syndrome	HOXA1
1	Atrial septal defect type 2	GATA4
1	Atrial septal defect with atrioventricular conduction defects	NKX2-5
1	Atrioventricular septal defect, partial with heterotaxy syndrome	CRELD1
1	Auriculocondylar syndrome type 1	GNAI3
1	Auriculocondylar syndrome type 2	PLCB4
1	Axenfeld-Rieger syndrome type 1	PITX2
1	Axenfeld-Rieger syndrome type 3	FOXC1
1	Bainbridge-Ropers syndrome	ASXL3

1	Baller-Gerold syndrome	RECQL4
1	Band-like calcification with simplified gyration and polymicrogyria	OCLN
1	Baraitser-Winter syndrome type 1	ACTB
1	Baraitser-Winter syndrome type 2	ACTG1
1	Basal cell nevus syndrome	SUFU
1	Basal cell nevus syndrome	PTCH1
1	Basal ganglia calcification type 1, ideopathic	SLC20A2
1	Basal ganglia calcification type 4	PDGFRB
1	Basal ganglia calcification type 5, idiopathic	PDGFB
1	Basal ganglia calcification type 6, idiopathic	XPR1
1	Beta-ureidopropionase deficiency	UPB1
1	Bifid nose	FREM1
1	Birt-Hogg-Dube syndrome	FLCN
1	Bjornstad syndrome	BCS1L
1	Blau syndrome	NOD2
1	Bohring-Opitz syndrome	ASXL1
1	Bone marrow failure syndrome type 2	ERCC6L2
1	Brachydactyly type A1C	GDF5
1	Brachydactyly type A2	BMP2
1	Brachydactyly type A2	BMPR1B
1	Brachydactyly type B1	ROR2
1	Brachydactyly type E1	HOXD13
1	Brachydactyly-mental retardation syndrome	HDAC4
1	Brachydactyly-syndactyly syndrome	HOXD13
1	Branchiooculofacial syndrome	TFAP2A
1	C syndrome	CD96
1	Campomelic dysplasia	SOX9
1	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	PRG4
1	Camurati-Engelmann disease	TGFB1
1	Cantu syndrome	ABCC9

1	Carpenter syndrome	RAB23
1	Carpenter syndrome type 2	MEGF8
1	Cartilage-hair hypoplasia	RMRP
1	Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B
1	Central hypoventilation syndrome, congenital	ECE1
1	Central hypoventilation syndrome, congenital	MECP2
1	Central hypoventilation syndrome, congenital	ZEB2
1	Central hypoventilation syndrome, congenital	GFRA1
1	Central hypoventilation syndrome, congenital	PHOX2A
1	Central hypoventilation syndrome, congenital	ASCL1
1	Central hypoventilation syndrome, congenital	EDN3
1	Central hypoventilation syndrome, congenital	BDNF
1	Central hypoventilation syndrome, congenital	GDNF
1	Central hypoventilation syndrome, congenital	RET
1	Cerebral dysgenesis, neuropathy, ichthyosis, palmoplantar keratoderma syndrome	SNAP29
1	Cerebrooculofacioskeletal syndrome type 1	ERCC6
1	Cerebrooculofacioskeletal syndrome type 4	ERCC1
1	CHARGE syndrome	CHD7
1	CHIME syndrome	PIGL
1	Chondrodysplasia with joint dislocations, GPAPP type	IMPAD1
1	Chondrodysplasia, Blomstrand type	PTH1R
1	Choroideremia	CHM
1	Chrondrodysplasia, acromesomelic, with genital anomalies	BMPR1B
1	Chudley-McCullough syndrome	GPSM2
1	Club foot	PITX1
1	Cockayne syndrome type A	ERCC8
1	Cockayne syndrome type B	ERCC6
1	CODAS syndrome	LONP1
1	Coffin-Siris syndrome, SMARCE1 related	SMARCE1
1	Cold-induced sweating syndrome	CRLF1

1	Cold-induced sweating syndrome type 2	CLCF1
1	Congenital heart disease and transposition of the great arteries	FOXH1
1	Congenital short-bowel syndrome	CLMP
1	Corpus callosum, agenesis of, with abnormal genitalia	ARX
1	Cortical malformations, occipital	LAMC3
1	Costello syndrome	HRAS
1	Cousin syndrome	TBX15
1	Craniodiaphyseal dysplasia, autosomal dominant	SOST
1	Cranioectodermal dysplasia type 1	IFT122
1	Cranioectodermal dysplasia type 2	WDR35
1	Cranioectodermal dysplasia type 3	IFT43
1	Cranioectodermal dysplasia type 4	WDR19
1	Craniofacial and neuro-developmental abnormalities	DISP1
1	Craniofacial and neuro-developmental abnormalities, JAG2 related	JAG2
1	Craniofacial anomalies and anterior segment dysgenesis syndrome	VSX1
1	Craniofrontonasal syndrome	EFNB1
1	Craniometaphyseal dysplasia	ANKH
1	Craniosynostosis and dental anomalies	IL11RA
1	Craniosynostosis type 1	TWIST1
1	Craniosynostosis type 2	MSX2
1	Craniosynostosis type 3	TCF12
1	Craniosynostosis type 4	ERF
1	Craniosynostosis type 6	ZIC1
1	Craniosynostosis, FGFR1 related	FGFR1
1	Craniosynostosis, nonspecific	FGFR2
1	Crouzon syndrome	FGFR2
1	Currarino syndrome	MNX1
1	Czech dysplasia	COL2A1
1	Dandy-Walker malformation and occipital cephaloceles, LAMC1 related	LAMC1
1	D-bifunctional protein deficiency	HSD17B4

1	De Sanctis-Cacchione syndrome	ERCC6
1	Desbuquois dysplasia type 1	CANT1
1	Desbuquois dysplasia type 2	XYLT1
1	Desmosterolosis	DHCR24
1	Diamond Blackfan anemia type 15 with mandibulofacial dysostosis	RPS28
1	Diamond-Blackfan anemia type 14 with mandibulofacial dysostosis	TSR2
1	DiGeorge syndrome	TBX1
1	Donnai-Barrow syndrome	LRP2
1	DOOR syndrome	TBC1D24
1	Duane Retraction syndrome	SALL4
1	Dysmorphism, HMG20B related	HMG20B
1	Ectodactyly, ectodermal dysplasia, and cleft lip/palate syndrome type 3	TP63
1	Ellis-van Creveld syndrome	EVC
1	Ellis-van Creveld syndrome	EVC2
1	Epiphyseal dysplasia, multiple, with myopia and deafness	COL2A1
1	Faciogenital dysplasia	FGD1
1	Fanconi anemia, complementation group Q	ERCC4
1	Feingold syndrome	MYCN
1	Fetal akinesia deformation sequence	DOK7
1	Fetal akinesia deformation sequence	RAPSN
1	Fibrochondrogenesis type 1	COL11A1
1	Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly	WNT7A
1	FILS syndrome	POLE
1	Fraser syndrome	FRAS1
1	Fraser syndrome	FREM2
1	Fraser syndrome	GRIP1
1	Frontonasal dysplasia type 1	ALX3
1	Frontonasal dysplasia type 2	ALX4
1	Fumarase deficiency	FH
1	Galloway-Mowat syndrome	WDR73

1	Geleophysic dysplasia type 2	FBN1
1	Genitopatellar syndrome	KAT6B
1	Glass syndrome	SATB2
1	Goldberg-Shprintzen megacolon syndrome	KIF1BP
1	GRACILE syndrome	BCS1L
1	Greenberg skeletal dysplasia	LBR
1	Greig cephalopolysyndactyly syndrome	GLI3
1	Growth hormone insensitivity, partial	GHR
1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	IGF1
1	Guttmacher syndrome	HOXA13
1	Hamamy syndrome	IRX5
1	Hand-foot-uterus syndrome	HOXA13
1	Hartsfield syndrome	FGFR1
1	Heart-hand syndrome, Slovenian type	LMNA
1	Helsmoortel-van der Aa syndrome	ADNP
1	Hennekam lymphangiectasia-lymphedema syndrome type 2	FAT4
1	Heterotaxy, visceral type 1	ZIC3
1	Heterotaxy, visceral type 2	CFC1
1	Heterotaxy, visceral type 4	ACVR2B
1	Heterotaxy, visceral type 5	NODAL
1	Heterotaxy, visceral type 6	CFAP53
1	Heterotaxy, visceral type 7	MMP21
1	Heterotaxy, visceral, BCL9L related	BCL9L
1	Hirschsprung disease	ZEB2
1	Hirschsprung disease	NRTN
1	Hirschsprung disease	RET
1	Hirschsprung disease	ECE1
1	Hirschsprung disease	NRG1
1	Hirschsprung disease	KIF1BP
1	Hirschsprung disease	EDNRB

1	Hirschsprung disease	EDN3
1	Hirschsprung disease, type 3, susceptibility to	GDNF
1	Holoprosencephaly type 11	CDON
1	Holoprosencephaly type 2	SIX3
1	Holoprosencephaly type 3	SHH
1	Holoprosencephaly type 4	TGIF1
1	Holoprosencephaly type 5	ZIC2
1	Holoprosencephaly-type 9	GLI2
1	Holt-Oram syndrome	TBX5
1	Hutchinson-Gilford progeria	LMNA
1	Hydranencephaly, Fowler type	FLVCR2
1	Hydrolethalus syndrome	HYLS1
1	Hypermethioninemia due to adenosine kinase deficiency	ADK
1	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AHCY
1	Hyperphosphatasia with mental retardation syndrome type 1	PIGV
1	Hyperphosphatasia with mental retardation syndrome type 2	PIGO
1	Hyperphosphatasia with mental retardation syndrome type 3	PGAP2
1	Hyperphosphatasia with mental retardation syndrome type 4	PGAP3
1	Hyperphosphatasia with mental retardation syndrome type 5	PIGW
1	Hypogonadism, alopecia, Diabetes mellitus, mental retardation and extrapyramidal syndrome	DCAF17
1	Hypogonadotropic hypogonadism type 6 with or without anosmia	FGF8
1	Hypospadias type 1, X-linked	AR
1	Hypospadias type 2, X-linked	MAMLD1
1	IMAGE syndrome	CDKN1C
1	Immunodeficiency-centromeric instability-facial anomalies syndrome type 2	ZBTB24
1	Intestinal atresia, multiple	TTC7A
1	IVIC syndrome	SALL4
1	Jackson-Weiss syndrome	FGFR1
1	Jackson-Weiss syndrome	FGFR2

1	Jawad syndrome	RBBP8
1	Kabuki syndrome type 1	KMT2D
1	Kabuki syndrome type 2	KDM6A
1	Kagami-Ogata syndrome	paternal UPD chr. 14
1	Kallmann syndrome type 2	FGFR1
1	Kallmann syndrome type 4	PROK2
1	Kallmann syndrome type 5	CHD7
1	KBG syndrome	ANKRD11
1	Keutel syndrome	MGP
1	Kleefstra syndrome	EHMT1
1	Klippel-Feil syndrome type 1, autosomal dominant	GDF6
1	Klippel-Feil syndrome type 2, autosomal dominant	MEOX1
1	Klippel-Feil syndrome type 3, autosomal dominant	GDF3
1	Klippel-Feil syndrome type 4, autosomal dominant, with myopathy and facial dysmorphism	MYO18B
1	Kniest dysplasia	COL2A1
1	Koolen syndrome	KANSL1
1	LADD syndrome	FGF10
1	LADD syndrome	FGFR2
1	Langer-Giedion syndrome	EXT1
1	Langer-Giedion syndrome	TRPS1
1	Larsen syndrome	FLNB
1	Left-right axis malformations	LEFTY2
1	Lenz-Majewski hyperostotic dwarfism	PTDSS1
1	LEOPARD syndrome type 3	BRAF
1	Lethal congenital contracture syndrome type 1	GLE1
1	Lethal congenital contracture syndrome type 4	MYBPC1
1	Limb-mammary syndrome	TP63
1	Lissencephaly type 1	PAFAH1B1

1	Lissencephaly type 2 (Norman-Roberts type)	RELN
1	Lissencephaly type 3	TUBA1A
1	Lissencephaly type 5	LAMB1
1	Lissencephaly, X-linked type 1	DCX
1	Lissencephaly, X-linked type 2	ARX
1	Lujan-Fryns syndrome	MED12
1	Lymphedema-distichiasis syndrome	FOXC2
1	Macrocephaly, alopecia, cutis laxa, and scoliosis	RIN2
1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	POLD1
1	Mandibuloacral dysplasia	LMNA
1	Mandibulofacial dysostosis with microcephaly	EFTUD2
1	Marden-Walker syndrome	PIEZO2
1	Marfan lipodystrophy syndrome	FBN1
1	Marfan syndrome	FBN1
1	Martsolf syndrome	RAB3GAP2
1	MASS syndrome	FBN1
1	McLeod syndrome with or without chronic granulomatous disease	XK
1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	PIK3R2
1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 2	AKT3
1	Meier-Gorlin syndrome 4	CDT1
1	Meier-Gorlin syndrome type 1	ORC1
1	Mental retardation and distinctive facial features with or without cardiac defects	MED13L
1	Mental retardation, autosomal dominant type 13	DYNC1H1
1	Mental retardation, truncal obesity, retinal dystrophy, and micropenis	INPP5E
1	Mental retardation, X-linked syndromic, Turner type	HUWE1
1	Metaphyseal anadysplasia type 1	MMP13
1	Metaphyseal anadysplasia type 2	MMP9
1	Metaphyseal dysplasia without hypotrichosis	RMRP
1	Microcephalic osteodysplastic primordial dwarfism type 1	RNU4ATAC
1	Microcephalic osteodysplastic primordial dwarfism type 2	PCNT

1	Microcephaly and chorioretinopathy with or without mental retardation	TUBGCP6
1	Microcephaly with cortical malformations, autosomal recessive type 2	WDR62
1	Microcephaly with epilepsy and diabetes syndrome	IER3IP1
1	Microcephaly with or without chorioretinopathy, Lymphedema, or Mental retardation, MCLMR	KIF11
1	Microcephaly with symplified gyral pattern and insulin-dependant diabetes	GFM2
1	Microcephaly, Amish type	SLC25A19
1	Microcephaly, AP4M1 related	AP4M1
1	Microcephaly, autosomal recessive type 1	MCPH1
1	Microcephaly, autosomal recessive type 11	PHC1
1	Microcephaly, autosomal recessive type 12	CDK6
1	Microcephaly, autosomal recessive type 13	CENPE
1	Microcephaly, autosomal recessive type 3	CDK5RAP2
1	Microcephaly, autosomal recessive type 4	CASC5
1	Microcephaly, autosomal recessive type 5	ASPM
1	Microcephaly, autosomal recessive type 6	CENPJ
1	Microcephaly, autosomal recessive type 7	STIL
1	Microcephaly, autosomal recessive type 8	CEP135
1	Microcephaly, autosomal recessive type 9	CEP152
1	Microcephaly, CEP63 related	CEP63
1	Microcephaly, MRE11A related	MRE11A
1	Microcephaly, MSMO1 related	MSMO1
1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	QARS
1	Microcephaly, short stature, and polymicrogyria with seizures	RTTN
1	Microcephaly, TUBB2B related	TUBB2B
1	Miller Dieker lissencephaly syndrome	YWHAE
1	Mitochondrial complex III deficiency, nuclear type 7	UQCC2
1	Mitochondrial myopathy and sideroblastic anemia type 1	PUS1
1	Mosaic variegated aneuploidy syndrome type 2	CEP57
1	Mulibrey nanism	TRIM37

1	Multiple congenital anomalies-hypotonia-seizures syndrome type 1	PIGN
1	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	B3GAT3
1	Myhre syndrome	SMAD4
1	Native American myopathy	STAC3
1	Neuroaxonal neurodegeneration, infantile, with facial dysmophism	NALCN
1	Neuroblastoma type 3, susceptibility to, familial	ALK
1	Nicolaides Baraitser syndrome	SMARCA2
1	Nijmegen breakage syndrome	NBN
1	Noonan syndrom like	SHOC2
1	Noonan syndrome type 1	PTPN11
1	Noonan syndrome type 10	LZTR1
1	Noonan syndrome type 3	KRAS
1	Noonan syndrome type 4	SOS1
1	Noonan syndrome type 5	RAF1
1	Noonan syndrome type 6	NRAS
1	Noonan syndrome type 7	BRAF
1	Noonan syndrome type 8	RIT1
1	Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia	CBL
1	Oculodentodigital dysplasia	GJA1
1	Ogden syndrome	NAA10
1	Olmsted syndrome	TRPV3
1	Opitz-Kaveggia syndrome	MED12
1	Orofacial cleft type 10	SUMO1
1	Orofacial cleft type 11	BMP4
1	Orofacial cleft type 5	MSX1
1	Orofacial cleft type 6	IRF6
1	Orofacial cleft type 7	PVRL1
1	Orofaciodigital syndrome type 14	C2CD3
1	Orofaciodigital syndrome type 4	TCTN3

1	Orofaciodigital syndrome type 5	DDX59
1	Orofaciodigital syndrome type 6	C5orf42
1	Osteoglophonic dysplasia	FGFR1
1	Otofaciocervical syndrome	EYA1
1	Otospondylomegaepiphyseal dysplasia	COL2A1
1	Otospondylomegaepiphyseal dysplasia	COL11A2
1	Pallister-Hall syndrome	GLI3
1	Papillorenal syndrome	PAX2
1	Parietal foramina type 1	MSX2
1	Pelger-Huet anomaly	LBR
1	Pelvic organ prolapse, LAMC1 related	LAMC1
1	Perlman Syndrome	DIS3L2
1	Pfeiffer syndrome	FGFR1
1	Pfeiffer syndrome	FGFR2
1	Phelan-McDermid syndrome	SHANK3
1	Phelan-McDermid syndrome	chr. 22q13.3
1	Pitt-Hopkins syndrome	NRXN1
1	Pitt-Hopkins syndrome	TCF4
1	Pituitary hormone deficiency type 1	POU1F1
1	Pituitary hormone deficiency type 2	PROP1
1	Platyspondylic skeletal dysplasia, Torrance type	COL2A1
1	Polymicrogyria asymmetric	TUBB2B
1	Polymicrogyria bilateral frontoparietal	ADGRG1
1	Polymicrogyria bilateral occipital	NR2E1
1	Polymicrogyria with optic nerve hypoplasia	TUBA8
1	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	PI4KA
1	Popliteal pterygium syndrome type 1	IRF6
1	Popliteal pterygium syndrome, lethal type	RIPK4
1	Porencephaly, familial	COL4A1
1	Poretti-Boltshauser syndrome	LAMA1

1	Postaxial acrofacial dysostosis	DHODH
1	Prolidase deficiency	PEPD
1	Prune belly syndrome	CHRM3
1	RAPADILINO syndrome	RECQL4
1	Rapp-Hodgkin syndrome	TP63
1	Rett syndrome	MECP2
1	Rett syndrome, congenital variant	FOXG1
1	Rhizomelic chondrodysplasia punctata type 2	GNPAT
1	Rhizomelic chondrodysplasia punctata type 3	AGPS
1	Rhizomelic chondrodysplasia punctata type 5	PEX5
1	Ritscher-Schinzel syndrome type 1	KIAA0196
1	RNA processing related disorders	HNRNPU
1	Roberts syndrome	ESCO2
1	Robinow syndrome, autosomal dominant type 1	WNT5A
1	Robinow syndrome, autosomal dominant type 2	DVL1
1	Robinow syndrome, autosomal recessive	ROR2
1	Robinow-Sorauf syndrome	TWIST1
1	Rothmund-Thomson syndrome	RECQL4
1	Rubinstein-Taybi syndrome	CREBBP
1	Rubinstein-Taybi syndrome	EP300
1	Saethre-Chotzen syndrome	TWIST1
1	Saethre-Chotzen syndrome	FGFR2
1	SC Phocomelia syndrome	ESCO2
1	Scaphocephaly, maxillary retrusion, and mental retardation	FGFR2
1	Schaaf-Yang syndrome	MAGEL2
1	Schinzel-Giedion midface retraction syndrome	SETBP1
1	Schizencephaly	EMX2
1	Schneckenbecken dysplasia	SLC35D1
1	Sclerosteosis type 1	SOST
1	Seckel syndrome	ATRIP

1	Seckel syndrome type 1	ATR
1	Seckel syndrome type 2	RBBP8
1	Seckel syndrome type 4	CENPJ
1	Seckel syndrome type 5	CEP152
1	Seckel syndrome type 6	CEP63
1	Seckel syndrome type 7	NIN
1	SED congenital	COL2A1
1	Seizures, scoliosis, and macrocephaly syndrome	EXT2
1	Septooptic dysplasia	HESX1
1	SERKAL syndrome	WNT4
1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	NHEJ1
1	Short stature syndrome	SHOX
1	Short stature, microcephaly, and endocrine dysfunction	XRCC4
1	Short stature, optic nerve atrophy, and Pelger-Huet anomaly	NBAS
1	SHORT syndrome	PIK3R1
1	Short-rib thoracic dysplasia type 10 with or without polydactyly	IFT172
1	Short-rib thoracic dysplasia type 11 with or without polydactyly	WDR34
1	Short-rib thoracic dysplasia type 2 with or without polydactyly	IFT80
1	Short-rib thoracic dysplasia type 3 with or without polydactyly	DYNC2H1
1	Short-rib thoracic dysplasia type 4 with or without polydactyly	TTC21B
1	Short-rib thoracic dysplasia type 5 with or without polydactyly	WDR19
1	Short-rib thoracic dysplasia type 6 with or without polydactyly	NEK1
1	Short-rib thoracic dysplasia type 7 with or without polydactyly	WDR35
1	Short-rib thoracic dysplasia type 8 with or without polydactyly	WDR60
1	Shprintzen-Goldberg syndrome	SKI
1	Silver-Russell syndrome	chr. 11p15
1	Silver-Russell syndrome	maternal UPD chr. 7
1	Skeletal abnormalities, CBFB related	CBFB

1	SMED Strudwick type	COL2A1
1	Smith-Lemli-Opitz syndrome	DHCR7
1	Sotos syndrome type 1	NSD1
1	Sotos-like syndrome	NFIX
1	Speech-language disorder type 1	FOXP2
1	Spina bifida folate sensitive	MTRR
1	Split-hand/foot malformation type 1 with sensorineural hearing loss	DLX5
1	Split-hand/foot malformation type 6	WNT10B
1	Spondylocostal dysostosis type 5	TBX6
1	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	B3GALT6
1	Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2
1	Spondyloperipheral dysplasia	COL2A1
1	Stiff skin syndrome	FBN1
1	Stuve-Wiedemann syndrome	LIFR
1	Syndactyly type 1	HOXD13
1	Syndactyly type 5	HOXD13
1	Syndactyly, mesoaxial synostotic, with phalangeal reduction	BHLHA9
1	TANC2 related brain disorders	TANC2
1	Temple syndrome	Maternal UPD chr. 14
1	Temple-Baraitser syndrome	KCNH1
1	Temtamy syndrome	C12orf57
1	Tetraamelia, autosomal recessive	WNT3
1	Three M syndrome type 1	CUL7
1	Three M syndrome type 2	OBSL1
1	Three M syndrome type 3	CCDC8
1	Toe syndactyly, telecanthus, and anogenital and renal malformations	FAM58A
1	Tooth agenesis, selective type 1	MSX1
1	Tooth agenesis, selective type 3	PAX9
1	Townes-Brocks syndrome	SALL1

1	Transposition of great arteries, dextro-looped 3	GDF1
1	Transposition of the great arteries, dextro-looped 1	MED13L
1	Treacher Collins syndrome type 1	TCOF1
1	Treacher Collins syndrome type 2	POLR1D
1	Treacher Collins syndrome type 3	POLR1C
1	Trigonocephaly type 1	FGFR1
1	Ulna and fibula, absence of, with severe limb deficiency	WNT7A
1	Ulnar-Mammary syndrome	TBX3
1	Urofacial syndrome	LRIG2
1	Van den Ende-Gupta syndrome	SCARF2
1	van der Woude syndrome type 1	IRF6
1	van der Woude syndrome type 2	GRHL3
1	Van Maldergem syndrome type 2	FAT4
1	Vater association	HOXD13
1	Vici syndrome	EPG5
1	Visceral myopathy	ACTG2
1	Warburg micro syndrome type 1	RAB3GAP1
1	Warburg micro syndrome type 2	RAB3GAP2
1	Warsaw breakage syndrome	DDX11
1	Weaver syndrome	EZH2
1	Webb-Dattani syndrome	ARNT2
1	Weill-Marchesani syndrome type 3	LTBP2
1	Weill-Marchesani syndrome, dominant type 2	FBN1
1	Werner syndrome	WRN
1	Wiedemann-Steiner syndrome	KMT2A
1	Williams-Beuren syndrome	chr. 7q11.23
1	Witkop syndrome	MSX1
1	XFE progeroid syndrome	ERCC4
1	ZIC5 related brain disorders	ZIC5

Panel

	Product name	TAT-day
	NIPT Clinical Exome Whole exome Solo	
33	Arthrogryposis panel	25
34	Ashkenazi panel (advanced)	25
35	Ashkenazi panel (basic)	25
36	Central hypoventilation syndrome panel	25
37	Cerebral cavernous malformations panel	25
38	Cleft lip/palate panel	25
39	Coffin-Siris syndrome panel	25
40	Craniosynostosis and craniofacial disorders panel	25
41	Heterotaxy panel	25
42	Hirschsprung disease panel	25
43	Holoprosencephaly panel	25
44	Klippel-feil syndrome panel	25
45	Lissencephaly and brain malformation panel	25
46	Marfan, Loeys-Dietz syndrome and related disorders panel	25
47	Metaphyseal dysplasia panel	25
48	Micro syndrome panel	25
49	Microcephaly panel	25
50	Multiple epiphyseal dysplasia panel	25
51	Noonan - CFC syndrome panel	25
52	Seckel syndrome panel	25
53	Skeletal dysplasia ciliopathy panel	25
54	Skeletal dysplasia extended panel	25
55	Tuberous sclerosis panel	25