BabyNEXT

BabyNEXT™ EXTENDED

Geni investigati e patologie associate

Gene	Disease	OMIM gene	OMIM Disease	Condition	RUSP
ABCC8	Familial hyperinsulinism ABCC8-related	600509	256450	Metabolic disorder - Inborn error of amino acid metabolism	
ABCD1	Adrenoleukodystrophy	300371	300100	Miscellaneous multisystem diseases	RUSP (C) *
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	603214	614857	Metabolic disorder - Inborn error of amino acid metabolism	
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	604773	611283	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S) **
ACAD9	acyl-CoA dehydrogenase-9 (ACAD9) deficiency	611103	611126	Metabolic Disorder - Inborn error of fatty acid metabolism	
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of	607008	201450	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	606885	201470	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ACADSB	2-methylbutyrylglycinuria	600301	610006	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
ACADVL	very long-chain acyl-CoA dehydrogenase deficiency	609575	201475	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
ACAT1	Alpha-methylacetoacetic aciduria	607809	203750	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
ACSF3	Combined malonic and methylmalonic aciduria	614245	614265	Metabolic Disorder - Inborn error of organic acid metabolism	

ADA	Severe combined immunodeficiency due to ADA deficiency	608958	102700	Primary Immunological deficiency	RUSP (S)
ADK	Hypermethioninemia due to adenosine kinase deficiency	102750	614300	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
AGL	Glycogen storage disease, type III	610860	232400	Other Disorders	
AGXT	Primary hyperoxaluria, type 1	604285	259900		
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	180960	613752	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
AK2	Reticular dysgenesis	103020	267500	Primary Immunological deficiency	
AKR1D1	Bile acid synthesis defect, congenital, 3	604741	235555		
ALDH4A1	Hyperprolinemia, type II	606811	239510	Metabolic disorder - Inborn error of amino acid metabolism	
ALDH7A1	Epilepsy, pyridoxine-dependent	107323	266100	Neurotransmitter Disorders	
ALDOB	Fructose intolerance, hereditary	612724	229600	Metabolic Disorder - Inborn error of organic acid metabolism	
ALPL	Hypophosphatasia	171760	241500		
ANK1	Spherocytosis, type 2	612641	182900		
AQP2	Diabetes insipidus, nephrogenic	107777	125800		

ARG1	Argininemia	608313	207800	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
ARSA	Metachromatic leukodystrophy	607574	250100	Lysosomal Storage Disorders (LSD)	
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	611542	253200	Lysosomal Storage Disorders (LSD)	
ASL	Argininosuccinic aciduria	608310	207900	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
ASS1	Citrullinemia Type 1	603470	215700	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
AUH	3-methylglutaconic aciduria, type I	600529	250950	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis / Nephrogenic diabetes insipidus AVPR2-related	300538	300539		
BCKDHA	Maple syrup urine disease, type Ia	608348	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
BCKDHB	Maple syrup urine disease, type Ib	248611	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
BTD	Biotinidase deficiency	609019	253260	Miscellaneous multisystem diseases	RUSP (C)
BTK	Agammaglobulinemia, X-linked 1	300300	300755	Primary Immunological deficiency	
CASR	Neonatal hyperparathyroidism / Autosomal dominant hypocalcemia	601199	601198	Endocrine Disorder	

CBS	Homocystinuria, B6-responsive and nonresponsive types	613381	236200	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
CD247	Immunodeficiency 25	186780	610163	Primary Immunological deficiency	
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	606475	613646	Metabolic Disorder - Inborn error of organic acid metabolism	
CD3D	Immunodeficiency 19	186790	615617	Primary Immunological deficiency	
CD3E	Immunodeficiency 18	186830	615615	Primary Immunological deficiency	
CFTR	Cystic fibrosis	602421	219700	Miscellaneous multisystem diseases	RUSP (C)
COL4A3	Alport syndrome COL4A3-related	120070	104200		
COL4A4	Alport syndrome, autosomal recessive	120131	203780		
COL4A5	Alport syndrome	303630	301050		
CPS1	Carbamoylphosphate synthetase I deficiency	608307	237300	Metabolic Disorder - Inborn error of amino acid metabolism	
CPT1A	Carnitine palmitoyltransferase type I deficiency	600528	255120	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
CPT2	Carnitine palmitoyltransferase type II deficiency	600650	255110	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
CTH	Cystathioninuria	607657	219500	Metabolic Disorder - Inborn error of amino acid metabolism	

CTNS	Cystinosis	606272	219800	Lysosomal Storage Disorders (LSD)	
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA	608508	233690		
CYBB	Chronic granulomatous disease CYBB-related	300481	306400		
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	610613	202010	Endocrine Disorder	
CYP11B2	Corticosterone methyloxidase deficiency	124080	610600		
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	613815	201910	Endocrine Disorder	RUSP (C)
CYP27A1	Cerebrotendinous xanthomatosis	606530	213700	Other Disorders	
DBT	Maple syrup urine disease, type II	248610	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
DCLRE1C	Omenn syndrome / Severe combined immunodeficiency, Athabaskan-type	605988	603554	Primary Immunological deficiency	
DECR1	2,4-dienoyl-CoA reductase deficiency	222745	-	Metabolic Disorder - Inborn error of fatty acid metabolism	
DLD	Dihydrolipoamide dehydrogenase deficiency	238331	246900	Metabolic Disorder - Organic Acidemias	
DNAJC19	Hyperphenylalaninemia, mild, non-BH4-deficient	606060	617384	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
DUOX2	Thyroid dyshormonogenesis 6	606759	607200	Endocrine Disorder	

DUOXA2	Thyroid dyshormonogenesis 5	612772	274900	Endocrine Disorder	
EPB42	Spherocytosis, type 6	177070	612690		
ETFA	Glutaric acidemia IIA	608053	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ETFB	Glutaric acidemia IIB	130410	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ETFDH	Glutaric acidemia IIC	231675	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ETHE1	Ethylmalonic encephalopathy	608451	602473	Metabolic Disorder - Organic Acidemias	
F9	Factor IX deficiency	300746	300807		
FAH	Tyrosinemia, type I	613871	276700	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
FBN1	Marfan syndrome and other FBN1-related disorders	134797	154700		
FBP1	Fructose-1,6-bisphosphatase deficiency	611570	229700	Metabolic Disorder - Organic Acidemias	
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	136430	613068	Other Disorders	
FTCD	Glutamate formiminotransferase deficiency	606806	229100	Metabolic Disorder - Organic Acidemias	
G6PC	Glycogen storage disease, type Ia	613742	232200	Metabolic Disorder - Inborn error of organic acid metabolism	
G6PD	Hemolytic anemia, G6PD deficient (favism)	305900	300908	Miscellaneous multisystem diseases	

GAA	Glycogen storage disease II - Pompe disease	606800	232300	Lysosomal Storage Disorders (LSD)	RUSP (C)
GALC	Krabbe disease	606890	245200	Lysosomal Storage Disorders (LSD)	
GALE	Galactose epimerase deficiency	606953	230350	Miscellaneous multisystem diseases	RUSP (S)
GALK1	Galactokinase deficiency with cataracts	604313	230200	Miscellaneous multisystem diseases	RUSP (S)
GALNS	Mucopolysaccharidosis IVA	612222	253000	Lysosomal Storage Disorders (LSD)	
GALT	Galactosemia	606999	230400	Miscellaneous multisystem diseases	RUSP (C)
GAMT	Cerebral creatine deficiency syndrome 2	601240	612736		
GATM	Cerebral creatine deficiency syndrome	602360	612718		
GBA	Gaucher disease, type I	606463	230800	Lysosomal Storage Disorders (LSD)	
GCDH	Glutaricaciduria, type I	608801	231670	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
GCH1	Hyperphenylalaninemia, BH4-deficient, B	600225	233910	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
GJB2	Deafness, autosomal recessive 1A	121011	220290	Deafness	RUSP (C)
GJB3	Deafness, digenic, GJB2/GJB3	603324	220290	Deafness	RUSP (C)
GJB6	Deafness, digenic GJB2/GJB6	604418	220290	Deafness	RUSP (C)
GLA	Fabry disease	300644	301500	Lysosomal Storage Disorders (LSD)	
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	610192	610199	Endocrine Disorder	

GLUD1	congenital hyperinsulinic hyperammonemia (HI/HA) syndrome	138130	606762	Endocrine Disorders	
GNAS	Pseudohypoparathyroidism Ia	139320	103580	Endocrine Disorders	
GNAS	Pseudohypoparathyroidism Ib	139320	603233	Endocrine Disorders	
GNAS	Pseudohypoparathyroidism Ic	139320	612462	Endocrine Disorders	
GNAS	Pseudopseudohypoparathyroidism	139320	612463	Endocrine Disorders	
GNMT	Glycine N-methyltransferase deficiency	606628	606664	Metabolic Disorder - Inborn error of amino acid metabolism	
GRHPR	Hyperoxaluria, primary, type II	604296	260000	Metabolic Disorder - Inborn error of amino acid metabolism	
GSS	Glutathione synthetase deficiency - 5-oxoprolinuria	601002	266130	Metabolic Disorder - Inborn error of amino acid metabolism	
GYS2	Glycogen storage disease 0, liver	138571	240600	Metabolic Disorder - Inborn error of organic acid metabolism	
HADH	Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency	601609	231530	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
HADHA	long-chain hydroxyacyl-CoA dehydrogenase deficiency	600890	609016	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
HADHA	Trifunctional protein deficiency	600890	609015	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
HADHB	Trifunctional protein deficiency	143450	609015	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)

HAL	Histidinemia	609457	235800	Metabolic Disorder - Inborn error of amino acid metabolism	
HAX1	Neutropenia, severe congenital 3, autosomal recessive	605998	610738		
HBA1	Thalassemia, alpha-	141850	604131	Hemoglobinopathies	RUSP (S)
HBA2	Thalassemia, alpha-	141850	604131	Hemoglobinopathies	RUSP (S)
HBB	Sickle cell anemia	141900	603903	Hemoglobinopathies	RUSP (C)
HBB	Thalassemias, beta-	141900	613985	Hemoglobinopathies	RUSP (C)
HGD	Alkaptonuria	607474	203500	Metabolic Disorder - Inborn error of amino acid metabolism	
HLCS	Holocarboxylase synthetase deficiency	609018	253270	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
HMGCL	HMG-CoA lyase deficiency	613898	246450	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
HMGCS2	HMG-CoA synthase-2 deficiency	600234	605911	Metabolic Disorder - Inborn error of fatty acid metabolism	
HOGA1	Hyperoxaluria, primary, type III	613597	613616		
HPD	Tyrosinemia, type III	609695	276710	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
HSD17B10	17-beta-hydroxysteroid dehydrogenase X (HSD10) deficiency	300256	300438	Metabolic Disorder - Organic Acidemias	
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	613890	201810	Endocrine Disorder	

HSD3B7	Bile acid synthesis defect, congenital, 2	607764	607765		
IDS	Mucopolysaccharidosis II	300823	309900	Lysosomal Storage Disorders (LSD)	
IDUA	Mucopolysaccharidosis type Ih	252800	607014	Lysosomal Storage Disorders (LSD)	
IL2RG	Severe combined immunodeficiency, X-linked	308380	300400	Primary Immunological deficiency	RUSP (C)
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	146661	608971	Primary Immunological deficiency	
INS	Diabetes mellitus, permanent neonatal	176730	606176	Endocrine Disorder	
IVD	Isovaleric acidemia	607036	243500	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
IYD	Thyroid dyshormonogenesis 5	612025	274800	Endocrine Disorder	
JAG1	Alagille syndrome 1 / Tetralogy of Fallot	601920	118450	Endocrine Disorder	
JAK3	Severe combined immunodeficiency	600173	600802	Primary Immunological deficiency	
KCNJ11	Familial hyperinsulinism	600937	601820	Endocrine Disorder	
KCNQ2	Early Infantile epileptic encephalopathy 7 / Benign neonatal seizures 2	602235	613720	Other Disorders	
LDLR	Familial hypercholesterolemia	606945	143890	Endocrine Disorder	
LHX3	Combined pituitary hormone deficiency 4	600577	221750	Endocrine Disorder	

LIG4	LIG4 syndrome	601837	606593	Primary Immunological deficiency	
LIPA	Wolman disease / Cholesteryl ester storage disease	613497	278000	Lysosomal Storage Disorders (LSD)	
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	612625	277380	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
LPL	Lipoprotein lipase deficiency	609708	238600		
MAT1A	Hypermethioninemia, due to methionine adenosyltransferase I/III deficiency	610550	250850	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	609010	210200	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	609014	210210	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MCEE	Methylmalonyl-CoA epimerase deficiency	608419	251120	Metabolic Disorder - Organic Acidemias	
MLYCD	Malonyl-CoA decarboxylase deficiency	606761	248360	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MMAA	Methylmalonic aciduria, vitamin B12-responsive	607481	251100	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	607568	251110	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	609831	277400	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)

MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	611935	277410	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MPI	Congenital disorder of glycosylation, type Ib	154550	602579		
MPL	Congenital amegakaryocytic thrombocytopenia	159530	604498		
MTHFR	Homocystinuria due to MTHFR deficiency	607093	236250	Metabolic Disorder - Inborn error of amino acid metabolism	
MTR	Homocystinuria-megaloblastic anemia, cobalamin G type	156570	250940	Metabolic Disorder - Inborn error of amino acid metabolism	
MTRR	Homocystinuria, cobalamin E type	602568	236270	Metabolic Disorder - Inborn error of amino acid metabolism	
MTTP	Abetalipoproteinemia	157147	200100		
MUT	Methylmalonic aciduria, mut(0) type	609058	251000	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MVK	Mevalonic aciduria	251170	610377	Metabolic Disorder - Inborn error of organic acid metabolism	
NADK2	2,4-dienoyl-CoA reductase deficiency	615787	616034	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
NAGS	N-acetylglutamate synthase deficiency	608300	237310	Metabolic Disorder - Inborn error of amino acid metabolism	
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611290	611291	Primary Immunological deficiency	

NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	600635	610978	Endocrine Disorder	
NKX2-5	Hypothyroidism, congenital nongoitrous, 5	600584	225250	Endocrine Disorder	
NPC1	Niemann-Pick disease, type C1	607623	257220	Lysosomal Storage Disorders (LSD)	
NPC2	Niemann-pick disease, type C2	601015	607625	Lysosomal Storage Disorders (LSD)	
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	613349	258870	Metabolic Disorder - Inborn error of amino acid metabolism	
OPA3	3-methylglutaconic aciduria, type III	606580	258501	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
OTC	Ornithine transcarbamylase deficiency	300461	311250	Metabolic Disorder - Inborn error of amino acid metabolism	
PAH	Phenylketonuria	612349	261600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	167415	218700	Endocrine Disorder	RUSP (C)
PC	Pyruvate carboxylase deficiency	608786	266150		
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	126090	264070	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
PCCA	Propionic acidemia	232000	606054	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
PCCB	Propionic acidemia	232050	606054	Metabolic Disorder - Inborn error of	RUSP (C)

				organic acid metabolism	
PHGDH	3-phosphoglycerate dehydrogenase deficiency	606879	601815	Metabolic Disorder - Inborn error of amino acid metabolism	
PHKB	Glycogen storage disease, type IXb	172490	261750	Other Disorders	
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	164050	613179	Primary Immunological deficiency	
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	603287	610090		
POU1F1	Combined pituitary hormone deficiency 1	173110	613038	Endocrine Disorder	
PPARG	peroxisome proliferator–activated receptorgamma (PPAR-g) ligand resistance syndrome (PLRS) or familial partial lipodystrophy type 3	601487	604367		
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	170280	603553	Primary Immunological deficiency	
PRODH	Hyperprolinemia, type I	606810	239500	Metabolic Disorder - Inborn error of amino acid metabolism	
PROP1	Combined pituitary hormone deficiency 2	601538	262600	Endocrine Disorder	
PRRT2	Familial infantile convulsions with paroxysmal choreoathetosis	614386	602066	Other Disorders	
PTPRC	Severe combined immunodeficiency PTPRC-related	151460	608971	Primary Immunological deficiency	
PTS	Hyperphenylalaninemia, BH4-deficient, A	612719	261640	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)

PYGL	Glycogen storage disease, type VI	613741	232700	Other Disorders	
QDPR	Hyperphenylalaninemia, BH4-deficient, C	612676	261630	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
RAG1	Omenn syndrome and other RAG1-related disorders	179615	603554	Primary Immunological deficiency	
RAG2	Omenn syndrome RAG2-related	179616	603554	Primary Immunological deficiency	
RB1	Retinoblastoma	614041	180200	Other Disorders	
SCN2A	Early infantile epileptic encephalopathy 11 / Benign familial infantile seizures 3	182390	613721	Other Disorders	
SCN8A	Early infantile epileptic encephalopathy 13 / Benign familial infantile seizures 5	600702	614558	Other Disorders	
SLC22A5	Carnitine deficiency, systemic primary	603377	212140	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
SLC25A13	Citrullinemia, type II, adult-onset - neonatal-onset	603859	603471	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	603861	238970	Metabolic Disorder - Inborn error of amino acid metabolism	
SLC25A20	Carnitine-acylcarnitine translocase deficiency	613698	212138	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
SLC26A4	Pendred syndrome	605646	274600	Deafness	
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	605646	600791	Deafness	

SLC2A1	Glucose transporter 1 deficiency syndrome and other SLC2A1-related disorders	138140	606777	
SLC37A4	Glycogen storage disease Ib	602671	232220	Lysosomal Storage Disorders (LSD)
SLC37A4	Glycogen storage disease Ic	602671	232240	Lysosomal Storage Disorders (LSD)
SLC39A4	Acrodermatitis enteropathica	607059	201100	
SLC3A1	Cystinuria	104614	220100	Metabolic Disorder - Inborn error of amino acid metabolism
SLC4A1	Distal renal tubular acidosis and other SLC4A1-related disorders	109270	179800	
SLC5A5	Thyroid dyshormonogenesis 1	601843	274400	Endocrine Disorder
SLC7A7	Lysinuric protein intolerance	603593	222700	
SLC7A9	Cystinuria	604144	220100	Metabolic Disorder - Inborn error of amino acid metabolism
SMPD1	Niemann-Pick disease, type A	607608	257200	Lysosomal Storage Disorders (LSD)
SMPD1	Niemann-Pick disease, type B	607608	607616	Lysosomal Storage Disorders (LSD)
SPR	Sepiapterin reductase deficiency	182125	612716	Metabolic Disorder - Inborn error of amino acid metabolism
STAR	Lipoid adrenal hyperplasia	600617	201710	
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	605014	603552	Primary Immunological deficiency

SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	603921	612073	Metabolic Disorder - Organic Acidemias	
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	611224	245400	Metabolic Disorder - Organic Acidemias	
TAT	Tyrosinemia, type II	613018	276600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
TAZ	3-methylglutaconic aciduria, type II - Barth syndrome	300394	302060	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
TCIRG1	Osteopetrosis 1	604592	259700		
TG	Thyroid dyshormonogenesis 4	188450	274700	Endocrine Disorder	
TH	Segawa syndrome	191290	605407		
THRA	Congenital nongoitrous hypothryoidism 6	190120	614450	Endocrine Disorder	
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	612418	614052	Metabolic Disorder - Organic Acidemias	
TPO	Thyroid dyshormonogenesis 2A	606765	274500	Endocrine Disorder	
TRHR	Generalized thyrotropin-releasing hormone resistance	188545	-		
TRMU	Acute infantile liver failure	610230	613070		

TSHB	Congenital nongoitrous hypothryoidism 4	188540	275100	Endocrine Disorder	
TSHR	Hypothyroidism, congenital, nongoitrous, 1	603372	275200	Endocrine Disorder	RUSP (C)
TTPA	Ataxia with isolated vitamin E deficiency	600415	277460	Other Disorders	
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	608897	608898	Primary Immunological deficiency	
WT1	Wilms tumor, type 1 and other WT1-related disorders	607102	194070		
ZAP70	Immunodeficiency 48	176947	269840	Primary Immunological deficiency	

* RUSP (C): Recommended Uniform Screening Panel - Core Conditions ACOG Committee Opinion 616 Jan 2015

** RUSP (S): Recommended Uniform Screening Panel - Secondary Conditions ACOG Committee Opinion 616 Jan 2015

BabyNEXT™ Farmacogenetica

Lista dei geni analizzati e dei farmaci metabolizzati dai rispettivi enzimi

Gene	Drugs	OMIM gene
CYP1A2	Phenacetin	124060
CYP2C19	Amitriptyline, Citalopram, Clomipramine, Doxepin, Escitalopram, Sertraline, Trimipramine, Clopidogrel, Voriconazole	124020
CYP2C9	Celecoxib, Warfarin, Fosphenytoin, Phenytoin	601130
CYP2D6	Codeine, Hydrocodone, Oxycodone, Tramadol, Ondansetron, Amitriptyline, Clomipramine, Desipramine, Doxepin, Fluoxetine, Fluvoxamine, Imipramine, Notriptyline, Paroxetine, Trimipramine, Aripiprazole, Iloperidone, Pimozide, Eliglustat, Atomoxetine	124030
CYP3A4	Imipramine, Amitriptyline, Sertraline, Venlafaxine, Nefazodone, Alprazolam, Triazolam, Midazolam, Ketoconazole, Itraconazole, Fluconazole, Astemizole, Ritonavir, Indinavir, Nelfinavir, Saquinavir, Carbamazepine, Dexamethasone, Phenobarbital, Phenytoin, Rifampicin, Terfenadine, Verapamil, Testosterone, Theophylline, Carbamazepine, Cisapride, Dexamethasone, Eritromicina, Ethinyl estradiol, Glyburide, Cyclosporin, Lovastatin	124010
CYP3A5	Tacrolimus	605325
CYP3A7	responsible for the metabolism of more than 50% of all clinically used drugs	605340
DPYD	Capecitabine	612779
SLCO1B1	Simvastatin	604843
TPMT	Mercaptopurine, Thioguanine, Azathioprine	187680
UGT1A1	Atazanavir	191740
VKORC1	Warfarin	608547