



New Molecular Tests (Ordered by Gene)

The molecular analysis includes analyses of all protein-coding sequences unless specified otherwise in the comment column.

If you don't find the disease (or gene) of interest in the alphabetical list, you can search the whole table with the Ctrl-F function:

1. Hold the Ctrl key, then press the F key
2. Type in the disease (gene) of interest
3. If the disease (gene) can be found, it is highlighted in black in the table

Information on mutations and genomic structure of genes can be found by clicking on [Mutation information](#).

Gene	Gene OMIM	Disease	Disease OMIM	Comment	Price in Euro
ACAD9 (ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 9)	611103	ACAD9 DEFICIENCY » ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 9, DEFICIENCY OF	611126		2300
ALG6 (ALG6, S. CEREVISIAE, HOMOLOG OF)	604566	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C, CDG1C » CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE 1, WITH DEFICIENT GLYCOSYLATION OF DOLICHOL-LINKED OLIGOSACCHARIDE, FORMERLY » CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE 5, FORMERLY	603147		1100
ALG8 (ALG8, S. CEREVISIAE, HOMOLOG OF)	608103	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1H, CDG1H	608104		1000
ALG9 (ALG9, S. CEREVISIAE, HOMOLOG OF)	606941	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1L, CDG1L	608776		1100
ALG12 (ALG12, S. CEREVISIAE, HOMOLOG OF; ASPARAGINE-LINKED GLYCOSYLATION 12, HOMOLOG OF; DOLICHYL-P-MANNOSE:MAN-7-GlcNAc-2-PP-DOLICHYL-ALPHA-6-MANNOSYLTRANSFERASE)	607144	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1G, CDG1G	607143		700
AMPD1 (AMP DEAMINASE)	102770	MYOADENYLATE DEAMINASE DEFICIENCY	102770	Whole Gene Sequencing	1600
AP1S2 (ADAPTOR-RELATED PROTEIN COMPLEX 1, SIGMA-2 SUBUNIT; CLATHRIN-ASSOCIATED/ASSEMBLY/ADAPTOR PROTEIN, SMALL 1-LIKE)	300629	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 59, MRX59	300630		480
ASAH1 (N-ACYLSPHINGOSINE AMIDOHYDROLASE 1; N-ACYLSPHINGOSINE DEACYLASE; ACID CERAMIDASE)	613468	FARBER LIPOGRANULOMATOSIS » CERAMIDASE DEFICIENCY » N-LAURYLSPHINGOSINE DEACYLASE DEFICIENCY	228000		Upon Request
ASPM (ABNORMAL SPINDLE-LIKE, MICROCEPHALY-ASSOCIATED; MCPH5)	605481	MICROCEPHALY, PRIMARY, TYPE 5 (AUTOSOMAL RECESSIVE), MCPH5	608716		2000
ATP1A3 (ATPase, Na ⁺ /K ⁺ TRANSPORTING, ALPHA-3 POLYPEPTIDE; SODIUM-POTASSIUM-ATPase)	182350	DYSTONIA, TYPE 12, DYT12	128235	Whole Gene Sequencing or 6 Exons	Whole Gene: 1700 Exons: 600
ATP6VOA2 (ATPase, H ⁺ TRANSPORTING, LYSOSOMAL, V0 SUBUNIT A2)	611716	WRINKLY SKIN SYNDROME CUTIS LAXA, TYPE 2A (AUTOSOMAL RECESSIVE) » CUTIS LAXA, DEBRE TYPE	278250 219200		1600 1600
B4GALT1 (UDP-GAL: BETA-GlcNAc BETA-1,4-GALACTOSYLTRANSFERASE, POLYPEPTIDE 1; BETA-1,4-GALACTOSYLTRANSFERASE 1)	137060	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2D, CDG2D	607091		700
CFHR5 (COMPLEMENT FACTOR H-RELATED 5)	608593	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, TYPE 6	612926		500
COG7 (COMPONENT OF OLIGOMERIC GOLGI COMPLEX 7)	606978	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2E, CDG2E	608779		1500
COG8 (COMPONENT OF OLIGOMERIC GOLGI COMPLEX 8)	606979	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2H, CDG2H	611182		550

DLD (DIHYDROLIPOAMIDE DEHYDROGENASE; BRANCHED CHAIN ALPHA-KETO ACID DEHYDROGENASE COMPLEX, E3 COMPONENT; PYRUVATE DEHYDROGENASE COMPONENT E3; GLYCINE CLEAVAGE SYSTEM L PROTEIN)	238331	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY » LIPOAMIDE DEHYDROGENASE DEFICIENCY, LACTIC ACIDOSIS DUE TO	248600		1800
DPAQT1 (DOLICHYL-PHOSPHATE N-ACETYLGLUCOSAMINE PHOSPHOTRANSFERASE; UDP-GlcNAc: DOLICHYL-PHOSPHATE N-ACETYLGLUCOSAMINE-PHOSPHOTRANSFERASE)	191350	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1J, CDG1J	608093		950
DPM1 (DOLICHYL-PHOSPHATE MANNOSYLTRANSFERASE 1, CATALYTIC SUBUNIT; DOLICHO-PHOSPHATE MANNOSYLTRANSFERASE)	603503	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1E, CDG1E	608799		800
EHMT1 (EUCHROMATIC HISTONE METHYLTRANSFERASE 1; EUHMTASE1)	607001	KLEEFSTRA SYNDROME » CHROMOSOME 9q34.3 DELETION SYNDROME	610253		2200
EP300 (E1A-BINDING PROTEIN, 300-KD)	602700	RUBINSTEIN-TAYBI SYNDROME » BROAD THUMBS AND GREAT TOES, CHARACTERISTIC FACIES, AND MENTAL RETARDATION » BROAD THUMB-HALLUX SYNDROME	180849		900
ESCO2 (ESTABLISHMENT OF COHESION 1, S. CEREVISIAE, HOMOLOG OF, 2)	609353	ROBERTS SYNDROME SC PHOCOMELIA SYNDROME » SC PSEUDOTHALIDOMIDE SYNDROME	268300 269000		1300 1300
GALE (UDP-GALACTOSE-4-EPIMERASE; GALACTOSE EPIMERASE)	606953	GALACTOSE EPIMERASE DEFICIENCY » GALE DEFICIENCY » GALACTOSEMIA, TYPE 3 » UDP-GALACTOSE-4-EPIMERASE DEFICIENCY	230350		850
GAS1 (GROWTH ARREST-SPECIFIC 1)	139185	HOLOPROSENCEPHALY 1, HPE1	236100		500
GCS1 (GLUCOSIDASE 1)	601336	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2B, CDG2B » GLUCOSIDASE 1 DEFICIENCY	606056		640
IL1RAPL1 (INTERLEUKIN 1 RECEPTOR ACCESSORY PROTEIN-LIKE 1; INTERLEUKIN 1 RECEPTOR 8)	300206	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 21, MRX21 » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 34, MRX34	300143		850
ITGA7 (INTEGRIN, ALPHA-7)	600536	MUSCULAR DYSTROPHY, CONGENITAL, DUE TO INTEGRIN ALPHA-7 DEFICIENCY » MYOPATHY, CONGENITAL, DUE TO INTEGRIN ALPHA-7 DEFICIENCY	613204		1900
LARGE (ACETYLGLUCOSAMINYLTRANSFERASE-LIKE PROTEIN -LIKE-GLYCOSYLTRANSFERASE)	603590	MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1D WALKER-WARBURG SYNDROME » HYDROCEPHALUS, AGYRIA, AND RETINAL DYSPLASIA » HARD SYNDROME » PAGON SYNDROME	608840 236670		1100 1100
MAN2B1 (MANNOSIDASE, ALPHA, CLASS 2B, MEMBER 1; MANB)	609458	ALPHA MANNOSIDOSIS	248500		2000
MGAT2 (ALPHA-1,6-@MANNOSYL-GLYCOPROTEIN BETA-1,2-N-ACETYLGLUCOSAMINYLTRANSFERASE)	602616	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2A, CDG2A	212066		300
MLYCD (MALONYL-CoA DECARBOXYLASE)	606761	MALONYL-CoA DECARBOXYLASE DEFICIENCY	248360		600
MPDU1 (MANNOSE-P-DOLICHO UTILIZATION DEFECT 1)	609458	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1F, CDG1F	609180		550
MPI (MANNOSEPHOSPHATE ISOMERASE; PHOSPHOMANNOSE ISOMERASE 1)	604041	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B, CDG1B » SAGUENAY-LAC SAINT-JEAN SYNDROME, SLSJ SYNDROME » MANNOSEPHOSPHATE ISOMERASE DEFICIENCY » PROTEIN-LOSING ENTEROPATHY-HEPATIC FIBROSIS SYNDROME	602579		600
NPHP1, NPHP2, NPHP3, NPHP4		NEPHRONOPHTHISIS		4 Genes	3000
NR5A1 (NUCLEAR RECEPTOR SUBFAMILY 5, GROUP A, MEMBER 1; STEROIDOGENIC FACTOR 1; SF1)	184757	46,XY GONADAL DYSGENESIS, COMPLETE OR PARTIAL, WITH OR WITHOUT ADRENAL FAILURE PREMATURE OVARIAN FAILURE 7, POF7	612965 612964		800 800

		ADRENOCORTICAL INSUFFICIENCY			800
OPA3	606580	OPTIC ATROPHY, TYPE 3, OPA3 (AUTOSOMAL RECESSIVE) > OPTIC ATROPHY PLUS SYNDROME > COSTEFF SYNDROME > 3-METHYLGLOUTACONIC ACIDURIA, TYPE 3	258501		650
		OPTIC ATROPHY, TYPE 3, OPA3 (AUTOSOMAL DOMINANT)	165300		650
PALB2 (PARTNER AND LOCALIZER OF BRCA2; FANCN)	610355	FANCONI ANEMIA, COMPLEMENTATION GROUP N, FANCN	610832		880
		BREAST CANCER			880
		PANCREATIC CANCER, SUSCEPTIBILITY TO, TYPE 3	613348		880
PHF6 (PHD FINGER PROTEIN 6)	300414	BORJESON-FORSSMAN-LEHMANN SYNDROME	301900		800
PMM2 (PHOSPHOMANNOMUTASE 2)	601785	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, CDG1A > JAEKEN SYNDROME > CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE 1A > PHOSPHOMANNOMUTASE 2 DEFICIENCY	212065		850
PRKCSH (PROTEIN KINASE C SUBSTRATE, 80-KD, HEAVY CHAIN; GLUCOSIDASE II, BETA SUBUNIT; HEPATOCYSTIN)	177060	POLYCYSTIC LIVER DISEASE	174050		1200
RECQL3 (RECQ2)	604610	BLOOM SYNDROME > (See also Molecular Screening Tests)	210900	Whole Gene Sequencing	2400
RFT1 (RFT1, S. CEREVISIAE, HOMOLOG OF)	611908	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1N, CDG1N	612015		1600
SEC63 (SEC63, S. CEREVISIAE, HOMOLOG OF)	608648	POLYCYSTIC LIVER DISEASE	174050		1500
SEPN1 (SELENOPROTEIN N, 1; SELN)	606210	RIGID SPINE MUSCULAR DYSTROPHY, TYPE 1 > MULTICORE MYOPATHY, SEVERE CLASSIC FORM > MINICORE MYOPATHY, SEVERE CLASSIC FORM > DESMIN-RELATED MYOPATHY WITH MALLORY BODIES > MUSCULAR DYSTROPHY, CONGENITAL, EICHSFELD TYPE	602771		1000
SLC35A1 (SOLUTE CARRIER FAMILY 35 (CMP-SIALIC ACID TRANSPORTER), MEMBER 1; CYTIDINE MONOPHOSPHATE-SIALIC ACID TRANSPORTER)	605634	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2F, CDG2F	603585		850
SLC35C1 (SOLUTE CARRIER FAMILY 35, MEMBER C1; GDP-FUCOSE TRANSPORTER 1)	605881	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2C, CDG2C > LEUKOCYTE ADHESION DEFICIENCY, TYPE 2C > RAMBAM-HASHARON SYNDROME	266265		400
THBD (THROMBOMODULIN)	188040	THROMBOPHILIA DUE TO THROMBOMODULIN DEFECT	188040		500
		HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, TYPE 6	612926		500
TMEM126A (TRANSMEMBRANE PROTEIN 126A)	612988	OPTIC ATROPHY, TYPE 7, OPA7	612989		700
TRIM32 (TRIPARTITE MOTIF-CONTAINING PROTEIN 32)	602290	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2H, LGMD2H	254110		550
TUBA1A (TUBULIN, ALPHA-1A)	602529	LISSENCEPHALY 3, LIS3	611603		800
TUSC3 (TUMOR SUPPRESSOR CANDIDATE 3)	601385	MENTAL RETARDATION (AUTOSOMAL RECESSIVE), TYPE 7, MRT7	611093		950
VPS33B (VACUOLAR PROTEIN SORTING 33, YEAST, HOMOLOG OF, B)	608552	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 1, ARC SYNDROME	208085		800
WNT10A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10A)	606268	ODONTOONYCHODERMAL DYSPLASIA	257980		750
		SCHOPF-SCHULZ-PASSARGE SYNDROME > KERATOSIS PALMOPLANTARIS WITH CYSTIC EYELIDS, HYPODONTIA, AND HYPOTRICHOSIS	224750		750

Top

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