



Screening Panels

Prices of the tests are in Euro, but can be converted to your local currency with the [currency converter](#).

Disease	Gene	Mutation	Price in Euro
Prenatal screening panel (Jewish diseases)			
ASKHENAZI JEWISH DISEASES (Tay-Sachs Disease, Bloom Syndrome, Canavan Disease, Niemann-Pick A, Familial Dysautonomia, Torsion Dystonia, Mucopolipidosis Type IV, Fanconi Anemia, Gaucher Disease, Factor XI Deficiency, Glycogen Storage Disease Type Ia, Maple Syrup Urine Disease, Non-Syndromic Sensorineural Hearing Loss, Familial Mediterranean Fever, Alpha 1-Anti-Trypsin Deficiency, NemaLine Myopathy, Usher Syndrome Type 1F, Familial Hyperinsulinemia, Lipoamide Dehydrogenase Deficiency and Glycogen Storage Disease Type III, Familial Hypercholesterolemia, Cystic Fibrosis)	Microarray Test with 77 mutations in 22 genes: HEX A, BLM, ASPA, SMPD1, IKBKAP, DYT1, MCOLN1, FANCC, F11, G6PC, BCKHDB, GJB2, MEFV, GDE, GBA, SERPINA1, NEB, DLD, PCDH15, ABCC8, LDLR and CFTR	77 Mutations in 22 Genes	600
ANTITRYPSINE DEFICIENCY	PI	E342K (Z Allele)	150
BLOOM SYNDROME	RECQL3	2281del6 / ins7	150
CANAVAN DISEASE	ASPA	Y231X and E285A	150
CYSTIC FIBROSIS	CF	DF508, G542X, W1282X, N1303K, 3849+10KbC>T, D1152H, 1717+1G->A	150
FAMILIAL DYSAUTONOMIA	IKBKAP	2507+6T>C	150
FANCONI ANEMIA TYPE C	FANCC	IVS4+4A>T	150
GAUCHER DISEASE	GBA	84GG, IVS2+1, N370S, L444P, V394L	150
GLYCOGENOSIS TYPE 1A	GSD1A	R83C	150
HEARING LOSS	GJB2 GJB6	35delG and 167delT 300 kb del	150
MAPLE SYRUP DISEASE	BCKDHB	R183P	150
MUCOLIPIDOSIS TYPE 4	MCOLN1	511-6944del and 5534A>G	150
NEMALINE MYOPATHY	NEB	2502del	150
NIEMANN-PICK DISEASE TYPE A/B	SMPD1	DelR608, FSP330, L302P R496L	150
TAY-SACHS DISEASE	HEXA	1277insTATC, IVS12+1G-C, G269S	150
USHER SYNDROME TYPE 1	PCDH15	R245X	150
Prenatal screening panel (standard)			
CYSTIC FIBROSIS CHROMOSOMES	CFTR	30 mutation kit	190
Prenatal screening panel MALES (extended)			
CYSTIC FIBROSIS SPINAL MUSCULAR ATROPHY CHROMOSOMES	CFTR SMN1	30 mutation kit Deletion	310
Prenatal screening panel FEMALES (extended)			
CYSTIC FIBROSIS SPINAL MUSCULAR ATROPHY FRAGILE X (only females) CHROMOSOMES	CFTR SMN1 FMR1	30 mutation kit Deletion Repeat	430
Thrombophilic panel			
THROMBOPHILIC PANEL	FACTOR 5 Leiden FACTOR 2 MTHFR	G1691A G20210A C677T, A1298C	250
Male infertility panel			
CBAVD (CONGENITAL BILATERAL VAS DEFERENS AGENESIS) Y DELETIONS	CFTR AZF	30 mutation kit Deletions AZFa,b,c	190

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