



Next Generation Sequencing Platforms

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

Heterogeneous diseases like Noonan syndrome, hypertrophic cardiomyopathy, dilated cardiomyopathy, deafness, Usher syndrome, retinitis pigmentosa, Brugada syndrome, Long QT syndrome, Arrhythmogenic Right Ventricular Cardiomyopathy, Polymorphic Ventricular Tachycardia, spastic paraplegia, Parkinson syndrome, Charcot-Marie-Tooth disease, mental retardation, etc can be caused by a multitude of genes.

Diagnostic molecular testing for these heterogeneous diseases is complicated and expensive if it follows a gene-after-gene classical approach.

Next generation sequencing platforms however allow the parallel sequencing of billions of nucleotides, so that large panels of genes can be sequenced in the same sequencing run. This new technology makes molecular testing for heterogeneous diseases cheap and fast.

Disease	Genes	Comment	Price in Euro
AORTIC ANEURYSM	<ul style="list-style-type: none"> • ACTA2 • CBS • FBN1 • FBN2 • MYH11 • COL3A1 • SLC2A10 • SMAD3 • TGFBR1 • TGFBR2 	10 genes	2900
ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC)	<ul style="list-style-type: none"> • Ryanodine receptor 2 (RYR2) for ARVC2 • Transmembrane protein 43 (TMEM43) for ARVC5 • Desmoplakin (DSP) for ARVC8 • Plakophilin-2 (PKP2) for ARVC9 • Desmoglein-2 (DSG2) for ARVC10 • Desmocollin-2 (DSC2) for ARVC11 • Junction plakoglobin (JUP) for ARVC12 	7 genes	2600
ATRIAL FIBRILLATION	<ul style="list-style-type: none"> • GJAS • KCNA5 • KCNE2 • KCNQ1 • NPPA 	5 genes	2275
AUTOSOMAL RECESSIVE AND SPORADIC RETINITIS PIGMENTOSA	<ul style="list-style-type: none"> • USH2A (Usherin) • EYS (Eyes Shut Homolog) • ABCA4 (ATP-binding cassette, subfamily A, member 4) • CRB1 (Crumbs homologue 1) • PDE6A (Phosphodiesterase 6A, cGMP-specific, rod alpha subunit) • PDE6B (Phosphodiesterase 6B, cGMP-specific, rod beta subunit) • RPE65 (Retinal pigment epithelium-specific protein, 65-kd) 	7 genes	2800

BRUGADA SYNDROME	<ul style="list-style-type: none"> • SCN5A (Brugada Syndrome, Type 1) • GPD1L (Brugada syndrome, Type 2) • CACNA1C (Brugada syndrome, Type 3) • CACNB2 (Brugada syndrome, Type 4) • SCN1B (Brugada syndrome, Type 5) 	5 genes	2600
CANCER	<ul style="list-style-type: none"> • BRCA1 • BRCA2 • CHEK2 • PALB2 • BRIP1 • BARD1 • CDH1 • ATM • TP53 • PTEN • STK11 • RAD50 • RAD51C • MRE11A • NBN 	15 cancer genes	2900
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)	<ul style="list-style-type: none"> • Cardiac ryanodine receptor channel (RYR2 gene with 105 exons) for CPVT1 • Calsequestrin (CASQ2 gene with 11 exons) for CPVT2 	2 genes	2600
COMPLEX IV GENE PANEL	<ul style="list-style-type: none"> • COX6B1 • COX10 • COX15 • SCO1 • SCO2 • SURF1 • TACO1 	7 genes	2600
CONGENITAL DISORDER OF GLYCOSYLATION (CDG)	<ul style="list-style-type: none"> • ALG2 • ALG3 • ALG6 • ALG8 • ALG9 • ALG12 • ATP6V0A2 • B4GALT1 • COG1 • COG7 • COG8 • DK1(DOLK) • DPAGT1 • DPM1 • GNE • LEC2(SLC35A1) • MGAT2 • MOGS(GCS1) • MPDU1 • MP1 • PMM2 • RFT1 • SLC35C1 • TUSC3 	24 genes	3600
CONGENITAL MUSCULAR DYSTROPHIES	<ul style="list-style-type: none"> • COL6A1 • COL6A2 • COL6A3 • FKR1 • FKTN • ITGA7 • LAMA2 • LARGE • POMGNT1 • POMT1 • POMT2 • SEPN1 	12 genes	3900
DEMENTIA AND AMYOTROPHIC LATERAL SCLEROSIS	<ul style="list-style-type: none"> • ALS2 • ANG • APOE • APP • CHMP2B • DCTN1 • FIG4 • FUS • MAPT • OPTN • PRGN • PSEN1 • PSEN2 • SETX • SOD • SORL1 • TARDBP • VABP • VCP 	19 genes	3000

DILATED CARDIOMIOPATHY (DCM)	<ul style="list-style-type: none"> • Myosin heavy chain (MYH7) • Troponin T (TNNT2) • Myosin-binding protein C (MYBPC3) • Troponin I (TNNI3) • Tropomyosin 1 (TPM1) • Actin (ACTC) • Lamin A/C (LMNA) • Z-band alternatively spliced PDZ motif-containing protein (ZASP) • Tafazzin (TAZ) • Phospholamban (PLN) • Lysosome-associated membrane protein 2 (LAMP2) • Transthyretin (TTR) • Delta-sarcoglycan (SGCD) • Desmin (DES) • 10 Mitochondrial transfer RNA's for leucine1/ leucine2 / glutamine /histidine / aspartic acid / isoleucine/ methionine/ lysine / serine1 / serine2 (MTTL1, MTTL2, MTTQ, MTHH, MTTD, MTTI, MTTM MTTK, MTTT1, MTTT2) • 3 Mitochondrial complex 1 subunits ND1, ND5 and ND6 (MTND1, MTND5, and MTND6) 	27 DCM genes	2800
FAMILIAL AORTOPATHIES	<ul style="list-style-type: none"> • FBN1 gene encoding fibrillin 1 for Marfan syndrome, MASS (mitral valve, aorta, skeleton, and skin) syndrome, isolated ectopia lentis, Weill-Marchesani syndrome, and Shprintzen-Goldberg syndrome • TGFBR2 gene encoding transforming growth factor-beta receptor, type 2 for Loey's-Dietz Syndrome, Marfan-like syndrome (Marfan syndrome type 2), Ehlers-Danlos syndrome type 4, and Thoracic Aortic Aneurysms • FBN2 gene encoding fibrillin 2 for Beals syndrome (congenital contractural arachnodactyly) 	3 genes	2275
FAMILIAL ARRHYTHMIA	<ul style="list-style-type: none"> • AKAP9 • ANK2 • CACNA1C • CACNB2 • CASQ2 • CAV3 • DSC2 • DSG2 • DSP • GPD1L • JUP • KCNA5 • KCNE1 • KCNE2 • KCNE3 • KCNH2 • KCNJ2 • KCNQ1 • NPPA • PKP2 • PLN • RYR2 • SCN1B • SCN3B • SCN4B • SCN5A • SNTA1 • TGFB3 • TMEM43 	29 genes	2275

FAMILIAL CARDIOMYOPATHY	<ul style="list-style-type: none"> • ABC9 • ACTC1 • ACTN2 • CALR3 • CAV3 • CSRP3 • DES • DSG2 • DTNA • EYA4 • FKTN • JPH2 • LAMP2 • LDB3 • LMNA • MIOZ2 • MYBPC3 • MYH6 • MYH7 • MYL2 • MYL3 • MYLK2 • NEXN • PLN • PRKAG2 • PSEN1 • PSEN2 • RBM20 • SCN5A • SGCD • SLC25A4 • TAZ • TCAP • TMPO • TNNC1 • TNNT2 • TPM1 • TNNI3 • TTN • VCL 	40 genes	2275
HYPERTROPHIC CARDIOMIOPATHY (HCM)	<ul style="list-style-type: none"> • Myosin-binding protein C (MYBPC3) • Regulatory and Essential light chains (MYL2, MYL3) • Myosin heavy chain (MYH7) • Actin (ACTC) • Tropomyosin 1 (TPM1) • Troponin I (TNNI3) • Troponin C (TNNC1) • Troponin T (TNNT2) • Caveolin 3 (CAV3) • Noncatalytic AMP-activated protein kinase gamma 2 (PRKAG2) • Lysosome-associated membrane protein 2 (LAMP2) • Alpha galactosidase (GLA) • Transthyretin (TTR) 	17 HCM genes with a detection rate of at least 60% in HCM	2600
LEIGH SYNDROME PANEL	<ul style="list-style-type: none"> • BCS1L • COQ2 • COX10 • COX15 • DLD • PDHA1 • SCO2 • SURF1 • TACO1 	9 genes	2600
LONG QT SYNDROME (LQT1-12)	<ul style="list-style-type: none"> • KCNQ1 encoding KQT-like voltage-gated potassium channel 1 for LQT1 • KCNH2 encoding Potassium channel, voltage-gated, H2 for LQT • SCN5A encoding Alpha polypeptide of voltage-gated sodium channel type V for LQT3 • ANK2 encoding Ankyrin-B for LQT4 • KCNE1 encoding Voltage-gated potassium channel, Isk related subfamily, member 1 for LQT5 • KCNE2 encoding Voltage-gated potassium channel, Isk related subfamily, member 2 for LQT6 • KCNJ2 encoding Inwardly rectifying potassium channel for LQT7 • CACNA1C encoding Calcium channel, L type, alpha 1 polypeptide isoform 	12 genes (LQT1-LQT12)	2600

	<ul style="list-style-type: none"> for LQT8 • CAV3 encoding Caveolin 3 for LQT9 • SCN4B encoding Sodium channel, voltage-gated, type IV beta subunit for LQT10 • AKAP9 encoding A-kinase anchor protein-9 for LQT11 • SNTA1 encoding alpha-1 syntrophin gene for LQT12 		
MITOCHONDRIAL DISEASE	<ul style="list-style-type: none"> • MTRNR1 • MTRNR2 • MTND1 • MTND2 • MTND3 • MTND4L • MTND4 • MTND5 • MTND6 • MTCO1 • MTCO2 • MTCO3 • MTATP8 • MTATP6 • MTCYB • MTF • MTTV • MTTL1 • MTTI • MTTQ • MTTM • MTTW • MTTA • MTTN • MTTT • MTTY • MTTT1 • MTTD • MTTK • MTTG • MTTT • MTTT • MTTT • MTTT 	Entire mitochondrial genome (16569 bp) including all the 37 genes (2 rRNA genes, 22 tRNA genes, and 13 protein-coding genes) and non-coding regions	1400
MITOCHONDRIAL DISEASES DUE TO MUTATIONS IN NUCLEAR GENES	<ul style="list-style-type: none"> • BCS1L • COQ2 • COX6B1 • COX10 • COX15 • DGUOK • DLD • MPV17 • OPA1 • PDHA1 • PEO1 (C10ORF2, Twinkle) • POLG (POLG1) • POLG2 • RRM2B • SCO1 • SCO2 • SLC25A4 (ANT1) • SUCLA2 • SUCLG1 • SURF1 • TACO1 • TK2 • TIMM8A (DDP1) • TYMP (ECGF1, TP) 	24 genes	3000
mtDNA DEPLETION SYNDROME	<ul style="list-style-type: none"> • DGUOK • MPV17 • OPA1 • PEO1 (C10ORF2, Twinkle) • POLG (POLG1) • POLG2 • RRM2B • SLC25A4 (ANT1) • SUCLA2 • SUCLG1 • TK2 • TYMP (ECGF1, TP) 	12 genes	2800
NOONAN, LEOPARD, COSTELLO and CARDIOFACIOCUTANEOUS SYNDROME	<ul style="list-style-type: none"> • PTPN11 • SOS1 • RAF1 • BRAF • MAP2K1 • MAP2K2 • HRAS • KRAS • SHOC2 	9 genes	1300

PARKINSON DISEASE	<ul style="list-style-type: none"> • DJ1 • LRRK2 • PARK2 • PINK1 • SNCA 	5 genes	3000
PEO PANEL	<ul style="list-style-type: none"> • OPA1 • PEO1 (C10ORF2, Twinkle) • POLG (POLG1) • POLG2 • RRM2B • SLC25A4 (ANT1) 	6 genes	2800
PERIODIC FEVER SYNDROMES	<ul style="list-style-type: none"> • ELANE (ELA2) • LPIN2 • MEFV • MVK • NLRP3 (CIAS1) • PSTPIP1 • TNFRSF1A 	7 genes	1800
PRIMARY CILIARY DYSKINESIA	<ul style="list-style-type: none"> • DNAAF1/LRRC50 • DNAAF2/c14orf104 • DNAH5 • DNAH11 • DNAI1 • DNAI2 • RSPH4A • RSPH9 • TXNDC3 • OFD1 • RPGR • CFTR 	12 genes	3400
SHORT QT SYNDROME	<ul style="list-style-type: none"> • CACNA1B • CACNA1C • KCNH2 • KCNJ2 • KCNQ1 	5 genes	2275
STARGARDT DISEASE 1, CONE-ROD DYSTROPHY, RETINITIS PIGMENTOSA, FUNDUS FLAVIMACULATUS, AGE-RELATED MACULAR DYSTROPHY	<ul style="list-style-type: none"> • ABCA4 (ABCR) 	1 gene	2100
SUDDEN DEATH	<ul style="list-style-type: none"> • ABCC9 • ACTC1 • ACTN2 • AKAP9 • ANK2 • CACNA1B • CACNA1C • CACNB2 • CALR3 • CASQ2 • CAV3 • CSRP3 • DES • DSC2 • DSG2 • DSP • DTNA • EYA4 • FBN1 • FBN2 • FKTN • GJA5 • GPD1L • JPH2 • JUP • KCNA5 • KCNE1 • KCNE2 • KCNE3 • KCNH2 • KCNJ2 • KCNQ1 • LAMP2 • LDB3 • LMNA • LRP6 • MEFA2 • MYBPC3 • MYH6 • MYH7 • MYL2 • MYL3 • MYLK2 • MYOZ2 • NEXN • NPPA • PKP2 • PLN • PRKAG2 • PSEN1 • PSEN2 • RBM20 • RYR2 • SCN1B • SCN3B • SCN4B • SCN5A • SGCD • SLC25A4 	72 genes	3400

	<ul style="list-style-type: none"> • SNTA1 • TAZ • TCAP • TGFB3 • TGFBR2 • TMEM43 • TMPO • TNNC1 • TNNT3 • TNNT2 • TPM1 • TTN • VCL 		
USHER SYNDROME and NON-SYNDROMIC DEAFNESS	<p>USHER TYPE 1</p> <ul style="list-style-type: none"> • MYO7A encoding Myosin 7A for Usher syndrome type 1A/B (USH1A/B) • USH1C encoding Harmonin for Usher syndrome type 1C (USH1C) • CDH23 encoding Cadherin23 for Usher syndrome type 1D (USH1D) • PCDH15 encoding Protocadherin15 for Usher syndrome type 1F (USH1F) • USH1G encoding Usher syndrome type-1G protein for Usher syndrome type 1G (USH1G) <p>USHER TYPE 2</p> <ul style="list-style-type: none"> • USH2A encoding Usherin for Usher syndrome type 2A (USH2A) • GPR98 (exons 8, 20, 31-41 & 89) encoding the G-protein-coupled 7-transmembrane receptor for Usher syndrome type 2C (USH2C) • WHRN encoding Whirlin for Usher syndrome type 2D (USH2D) <p>USHER TYPE 3</p> <ul style="list-style-type: none"> • CLRN1 encoding Clarin 1 for Usher syndrome type 3A (USH3A) <p>NON-SYNDROMIC DEAFNESS</p> <ul style="list-style-type: none"> • GJB6 • MYO6 • OTOF • SLC26A4 (PDS) • TMC1 • TMIE • TMPRSS3 • TMC1 • MTT5 (tRNA^{ser}(UCN)) • MTRNR1 (6 mutations in 12S rRNA) 	19 genes	3400
USHER SYNDROME AND RETINITIS PIGMENTOSA	<ul style="list-style-type: none"> • USH2A encoding Usherin 	USH2A next generation sequencing with del/dup testing (Targeted aCGH)	2000

X-LINKED MENTAL RETARDATION	<ul style="list-style-type: none"> • ABCD1 • ACSL4 • AFF2 • AGTR2 • AP1S2 • ARHGEF6 • ARHGEF9 • ARX • ATP6AP2 • ATP7A • ATRX • BCOR • BRWD3 • CASK • CDKL5 • CUL4B • DCX • DKC1 • DLG3 • DMD • FANCB • FGD1 • FLNA • FMR1 • FTSJ1 • GDI1 • GK • GPC3 • GRIA3 • HCCS • HPRT • HSD17B10 • HUWE1 • IDS • IGBP1 • IL1RAPL1 • JARID1C • KIAA2022 • KLF8 • L1CAM • LAMP2 • MAGT1 • MAOA • MBTPS2 • MECP2 • MED12 • MID1 • MTM1 • NDP • NDUFA1 • NHS • NLGN3 • NLGN4 • NSDHL • NXF5 • OCRL • OFD1 • OPHN1 • OTC • PAK3 • PCDH19 • PDHA1 • PGK1 • PHF6 • PHF8 • PLP1 • PORCN • POBP1 • PRPS1 • RAB39B • RPL10 • RPS6KA3 • SHROOM4 • SLC16A2 • SLC9A6 • SMC1A • SMS • SOX3 • SRPX2 • SYN1 • SYP • TIMM8A • TM4SF2 • TSPAN7 • UBE2A • UPF3B • ZCCHC12/SIZN2 • ZDHHC15 • ZDHHC9 • ZNF41 • ZNF674 • ZNF711 • ZNF81 	93 genes	4200
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