



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
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	2800
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	2800
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	2800

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	2800
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) • 6 Mitochondrial transfer RNAs for leucine/glutamine/histidine/lysine/serine1/serine2 (MTTL1, MTTO, MTTH, MTTK, MTTS1 and MTTS2) • 3 Mitochondrial complex 1 subunits ND1, ND5 and ND6 (MTND1, MTND5, and MTND6) 	23 DCM genes	2800
LONG QT SYNDROME (LQT1-10)	<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 for LQT8 • CAV3 encoding Caveolin 3 for LQT9 • SCN4B encoding Sodium channel, voltage-gated, type IV beta subunit for LQT10 	10 genes (LQT1-LQT10)	2800

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 • 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	2100
NOONAN, LEOPARD, COSTELLO and CARDIOFACIO CUTANEOUS SYNDROME	<ul style="list-style-type: none"> • PTPN11 • SOS1 • RAF1 • BRAF • MAP2K1 • MAP2K2 • HRAS • KRAS 	8 genes	1500
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
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) 	19 genes	3400

	<p>NON-SYNDROMIC DEAFNESS</p> <ul style="list-style-type: none"> • GJB6 • MYO6 • OTOF • SLC26A4 (PDS) • TMC1 • TMIE • TMPRSS3 • TMC1 • MTT5 (tRNAser(UCN)) • MTRNR1 (6 mutations in 12S rRNA) 		
<p>USHER SYNDROME AND RETINITIS PIGMENTOSA</p>	<ul style="list-style-type: none"> • USH2A encoding Usherin 	<p>USH2A next generation sequencing with del/dup testing (Targeted aCGH)</p>	<p>2000</p>

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