ANEUFAST™ QF-PCR

Rapid Diagnosis of Trisomy 13, 18, 21 and Sex Chromosome Aneuploidies

The **Aneufast™ Kit** is a molecular diagnostic assay designed for rapid detection of chromosomes 13, 18, 21, X and Y aneuploidies by Quantitative Fluorescent PCR (QF-PCR).

The Aneufast™ QF-PCR Kit contains a total of 36 markers in six multiplex reactions that amplify selected Short Tandem Repeats (STRs) and the gender determining sequences Amelogenin-SRY. Two multiplex QF-PCR amplifications of 21 markers (S1 and S2) are designed to be analysed in a single electrophoresis to increase sample throughput. The inclusion of 4 chromosome specific extra markers sets allows definitive results in 100% of cases.

Rapid Prenatal Diagnosis: Aneufast™ QF-PCR Kit Workflow

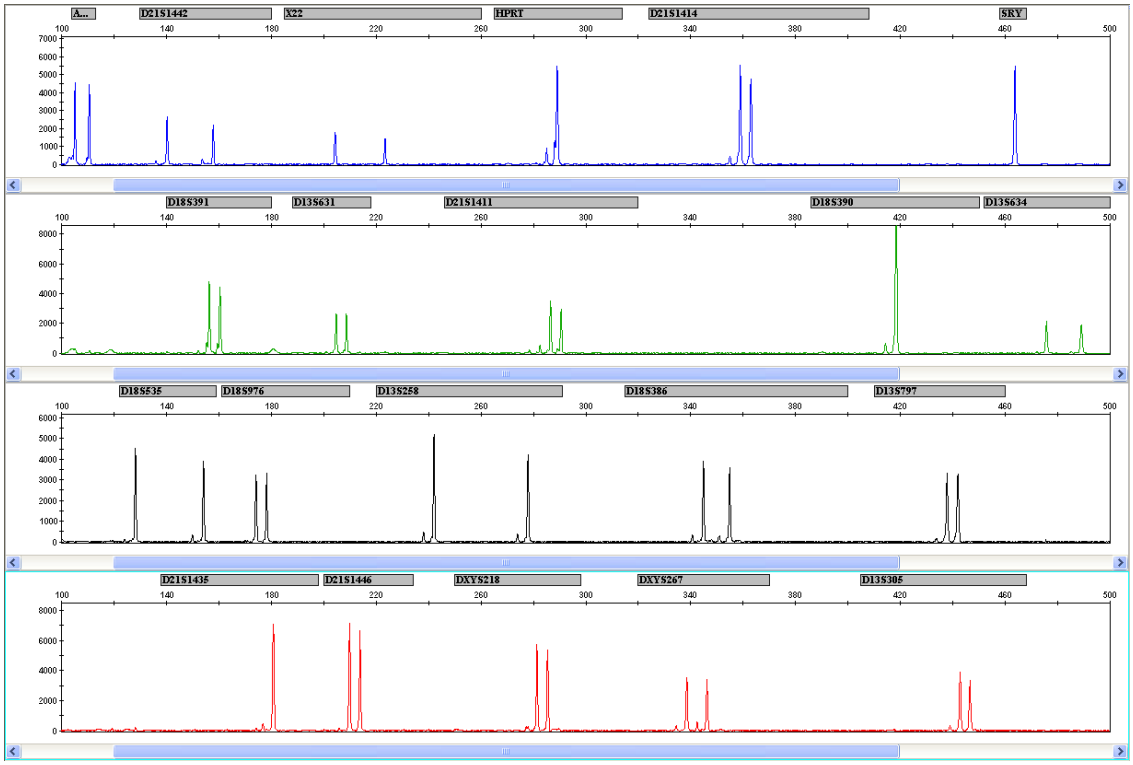
Sample → DNA Extraction 15 min. → QF-PCR Amplification 90 min → Electrophoresis – Analysis 50 min. → **Result**

Aneuploidy screening is performed by amplifying five STRs on chromosomes 13, 18, 21, three pseudoautosomal markers, one X linked, as well as the AMXY and SRY. Markers are distributed in two multiplex QF-PCR assays (S1 and S2) of the **Aneufast™** Kit, in order to reduce the risk of sample mishandling. Following collection of the products and simultaneous electrophoretic analysis, agreement between results from the two multiplexes allows diagnosis to be performed with two independent assays on each sample.

Samples with fewer than two informative markers on each chromosome can be tested using chromosome-specific multiplex PCR assays to include up to eight STRs on chromosomes 21 and 18, seven on chromosome 13, eight markers on the X and four on both XY. These sets of additional markers (M13, M18, M21 and MXY), which are included in the **Aneufast™** QF-PCR kit, can also be useful for additional confirmation of sample identity in all aneuploidy cases, by testing a second aliquot obtained from the original sample.

Diagnosis of Chromosome X Monosomy

The **Aneufast™** QF-PCR MXY assay includes a total of 11 selected highly polymorphic markers on the sex chromosomes and an additional non-polymorphic sequence to confirm Chromosome X Monosomy (Turner Syndrome) independently from allele frequency calculations.



Main Features:

- Sample to result in less than 3 hours
- Extra Markers on Chromosomes 13, 18, 21, X and Y included in the box.
- Optimised to work on all Applied Biosystems sequencers
- Five Dye-Labeling allows simultaneous analysis of several loci.
- Only product on the market with 4 pseudoautosomal markers for XY aneuploidy and mosaic cases.

Ordering Information	
Cat No	**Description**
mlg.anf.100	100 tests
mlg.anf.50	50 tests
mlg.anf.25	25 tests

Manufactured by:
molGENTIX SL
Verge de Guadalupe 18
E-08950 Esplugues de Llobregat
Barcelona, Spain

Distributed by:
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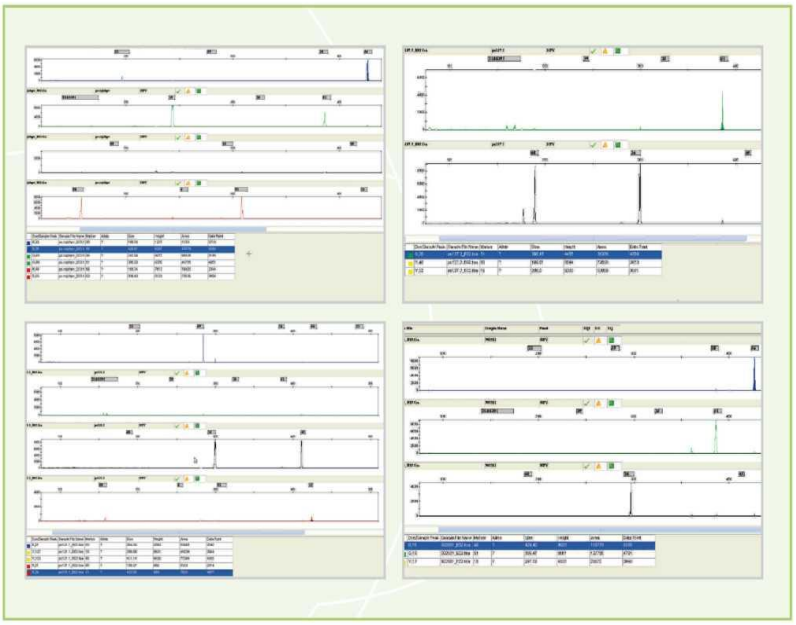
f HPV typing™

MULTIPLEX-FLUORESCENT PCR

Multiplex Fluorescent-PCR Kit for Human Papilloma Virus (HPV) Genotyping

Human Papillomavirus (HPV) infection is the main cause of high-grade cervical intraepithelial neoplasia and cancer. The named low risk types, including HPV-6, and -11, mostly cause the development of genital condylomata. The high-risk types, HPV-16, -18, -31, -33, -35, -39, -45, -51, -52, -56, -58, -59, -66 and -68, induce cervical squamous intraepithelial lesions (SIL), which in turn are classified as low (LSIL) and high grade (HSIL) in severity, and which may progress to cervical cancer.



Although several HPV types have been characterised as high-risk, recently published results show that knowing the **specific HPV type** has the **greatest clinical value**, because not all high risk HPV types have the same carcinogenic potential. **HPV16, HPV18, HPV31, and HPV33** infection persistence has been associated with **high absolute risks for progression** to high-grade cervical lesions, compared to the other high risk HPV type-specific infections that have been associated with low absolute risks. Identifying the specific HPV types could benefit women at **higher risk of progression to CIN3 or worse***.

The **f HPV typing™** kit is a molecular diagnostic assay designed for simultaneous **detection and genotyping** of 16 HPV-specific types by multiplex fluorescent PCR.

The **f HPV typing™** kit uses 16 primers amplifying within the **E6** and **E7 regions** of the HPV genome, the most likely to be retained after viral integration. Extracted DNA is amplified using a multiplex PCR with a set of 16 fluorescently labelled primers recognising HPV types **6, 11, 16, 18, 31, 33, 35, 39, 45, 51, 52, 56, 58, 59, 66** and **68** plus a human STR used as an internal control. This sequence is added to check DNA integrity and PR inhibitors; it is also helpful in detecting DNA mixtures due to sample mishandling. Different labelling of primers allows generation of similar sized amplicons in the same PCR reaction.

f HPV typing™ PCR Kit Workflow

Sample | DNA Extraction → | F-PCR Amplification → | Electrophoresis – Analysis | **Result**

Main Features:

- Suitable for different samples, such as fresh or paraffin-embedded tissues (biopsies), liquid-based samples, e.g. thin prep, and ano-genital and oral swab samples.
- Differentiates between persistent and new infections
- Sample to result in less than 4 hours
- Optimised to work on all Applied Biosystems sequencers
- The fluorescent PCR allows multiplex amplification in the same tube
- All cervical cancer-associated genotypes identified in one run
- Simple, automated computer analysis

Ordering Information	
Cat No	**Description**
mlg.hpv.100	100 tests
mlg.hpv.50	50 tests
mlg.hpv.25	25 tests

*1.-Skinner SR, Wheeler CM, Romanowski B, et al. Progression of HPV infection to detectable cervical lesions or clearance in adult women: Analysis of the control arm of the VIVIANE study. Int J Cancer. 2016 May 15;138(10):2428-38. doi: 10.1002/ijc.29971.
2.-Elfström KM, Smelov V, Johansson AL, et al. Long-term HPV type-specific risks for ASCUS and LSIL: a 14-year follow-up of a randomized primary HPV screening trial. Int J Cancer. 2015 Jan 15;136(2):350-9.
3.-Thomsen LT, Frederiksen K, Munk C, et al. Long-term risk of cervical intraepithelial neoplasia grade 3 or worse according to high-risk human papillomavirus genotype and semi-quantitative viral load among 33,288 women with normal cervical cytology. Int J Cancer. 2015 Jul 1;137(1):193-203.
4.-Kjær SK, Frederiksen K, Munk C, Iftner T, et al. Long-term absolute risk of cervical intraepithelial neoplasia grade 3 or worse following human papillomavirus infection: role of persistence. J Natl Cancer Inst. 2010 Oct 6;102(19):1478-88.
5.-Muñoz N, Castellsagué X, de González AB, Gissmann L. Chapter 1: HPV in the etiology of human cancer. Vaccine. 2006;24(suppl 3):S1–S10.

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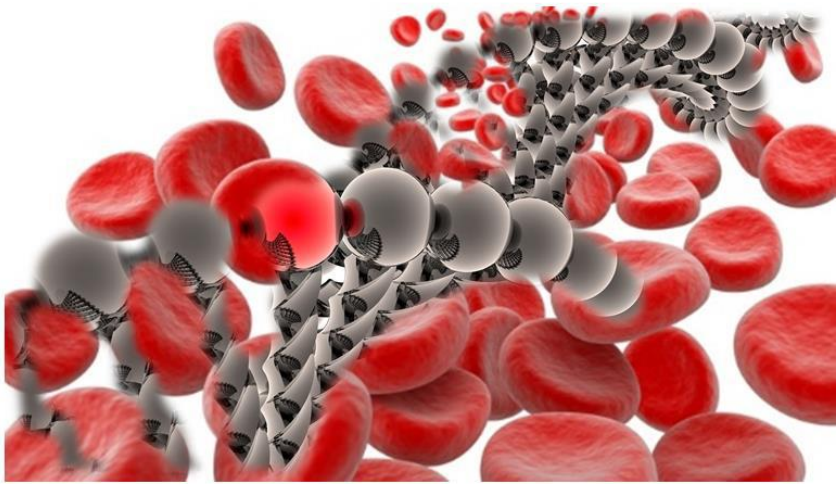
SNP Detective CVD-1 Thrombophilia Panel



Cardiovascular Diseases (CVDs) are common, but in many cases they can be avoided. Atherosclerosis and venous thrombosis are the two major manifestations of CVD. Both are caused by complex interactions of environmental and genetic parameters.

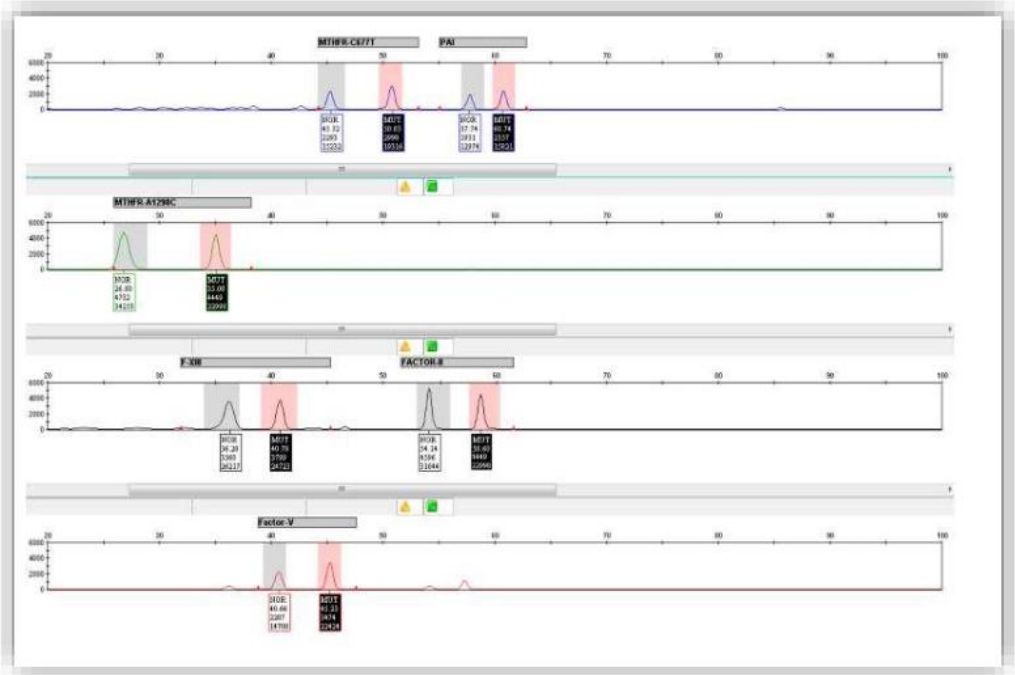
An unhealthy lifestyle in combination with certain genetic variants can contribute to atherosclerosis. Relevant genes include those involved in endothelial dysfunction, hyperlipidemia, hypertension, and inflammation. A combination of adverse influences (female hormone intake, immobilization, surgery or cancer) and variations in genes responsible for the coagulation system can also lead to thrombosis. Testing for genetic variations and adequate prophylaxis contribute to a lower risk of developing CVD.

The **SNP Detective CVD-1 Thrombophilia Panel** is a fragment analysis based kit for the detection of risk factor genes which raise susceptibility for thrombophilia. The system contains 6 primer pairs for amplification of 6 different target sequences on the genes in the panel. Twelve single-base-specific DNA probes are also used for detection of SNPs. The kit uses a five-dye fluorescent system which allows multiplex amplification/ligation in the same tube for automated SNP (Single Nucleotide Polymorphism) analysis.

The kit incorporates an external control with a heterozygous pattern for all related mutations, to ensure the performance of the ligation reagents and to identify problems with genomic DNA samples and DNA probes.

SNP Detective CVD-1 Panel Workflow

DNA extraction 60 min. → DNA quantification 15 min. → Multiplex PCR 120 min. → Probe ligation 120 min. → Electrophoresis 30 min. → Analysis 15 min.



Regions to be examined	MTHFR C677T SNP (Ala222Val) MTHFR A1298C SNP (Glu429Ala) FII Prothrombin G20210A SNP FV LEIDEN R506Q FXIII V34L SNP (Val34Leu) PAI-1 (Plasminogen activator inhibitor-1)

Main Features:

- Sample to result in less than 7 hours
- Includes an external control with a heterozygous pattern for all related mutations, ensuring performance and reliability
- Using the same tube reduces handling time and circumvents possible cross-contamination
- Works on capillary electrophoresis platforms with five-dye technology
- Precise and quick genotyping for all mutations in the same plot screen
- Multiple samples can be analyzed in seconds, using GeneMapper® or GeneMarker® Software
- Wide range of DNA concentration (2 to 100ng/ul) and low DNA quality requirements (A260/A280 = 0.9 to 2.2) enable analysis of all specimen types without DNA calculation and dilution

Ordering Information

Cat No	Description
208100	100 tests
208050	50 tests
208025	25 tests

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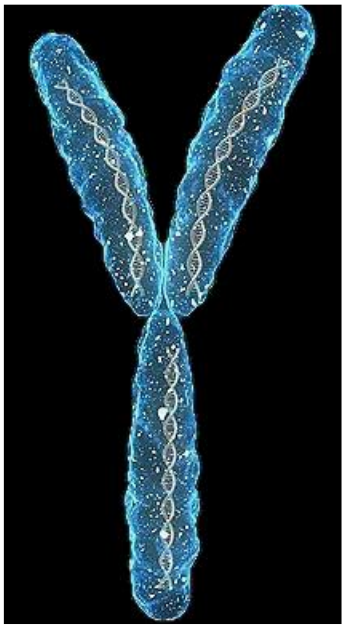
Y Chromosome Deletion Extended Kit



The **GML Y Chromosome Deletion Extended Kit** is a fragment analysis based kit for the diagnosis of Y-chromosome deletions routinely performed in the detection of male infertility in men with azoospermia or severe oligozoospermia

The GML Y Chromosome Deletion Extended kit comprises 20 primer pairs that are homologous to previously identified and mapped sequence-tagged sites (STS). Y chromosome deletions in the regions that are amplified by these primer sets have been associated with male infertility. The system uses a five-dye fluorescent system for automated DNA fragment analysis, which allows multiplex amplification and electrophoresis of over 20 STS, including the AMXY marker (Chr.X 105 bp, Chr.Y 110 bp; Xp22.1, Yp11.2), simultaneously.

Y Chromosome Deletion Extended Kit Workflow

DNA extraction 60 min. → DNA quantification 15 min. → PCR 120 min. → Electrophoresis 45 min. → Analysis 15 min.

Diagnostic testing uses the PCR amplification of sequence-tagged sites (STS) of the AZFa, AZFb and AZFc regions of the Y-chromosome. Successful amplification of an STS marker indicates its presence, whereas the absence of PCR amplification is indicative of a deletion. Primers for a total of 20 markers are combined into a single multiplex PCR reaction. All of the loci recommended by the European Academy of Andrology (EAA) and the European Quality Monitoring Network Group (EMQN) are included in the kit.

The ZFX/ZFY genes are used as an internal control for the PCR amplification, as these primers amplify unique fragments both on the Y-chromosome (ZFY) and on the X-chromosome (ZFX). The SRY gene is included in the analysis as control for the testis determining factor on the short arm of the Y chromosome and for the presence of Y-specific sequences in cases where the ZFY gene is absent.

The Y-Del Extended Primer Mix includes a primer pair to amplify chromose- specific sequences of the paralogous gene Amelogenin, which has a high degree of sequence identity between chromosomes Chr.X and Chr.Y. However, nucleotide differences occur within each locus and can be used to generate chromose-specific PCR product. The primer pair included in the Y-Del Extended Primer Mix exploits a 5bp deletion (Chr.X 105bp, Chr.Y 110bp; Xp22.1, Yp11.2) to generate one Chr.X-specific product that is 5bp shorter than the corresponding product on the Y chromosome.

STS included in the Y Chromosome Deletion Extended Kit

Marker	Region	Location	Bin Range (bp)	Dye
AmelXY	Amelogenin	Xp22.1, Yp11.2	105-110	FAM
sY88	AZFa	Yq11.221	118-128	FAM
sY153	AZFb	Yq11.223- Yq11.23	134-144	FAM
sY121	AZFb	Yq11.222	180-200	FAM
sY1065	AZFa	Yq11.1	235-245	FAM
sY127	AZFb	Yq11.223	267-277	FAM
sY105	AZFb	Yq11.222	291-305	FAM
sY143	AZFb	Yq11.223	305.5-315	FAM
sY1191	AZFc gr/gr	Yq11.223	375-395	FAM
ZFX/Y	ZFY/ZFX	Xp22.11/Yp11.2	485-495	FAM
sY1291	AZFc gr/gr	Yq11.223	517-537	FAM
sY255	AZFc	Yq11.23 (223)	118-128	JOE
sY160	Terminal	Yq12	231-241	JOE
sY82	AZFa	Yq11.21	260-268	JOE
sY83	AZFa	Yq11.21	270-280	JOE
sY134	AZFb	Yq11.223	297-308	JOE
sY86	AZFa	Yq11.21 (221)	312-322	JOE
sY84	AZFa	Yq11.1 (221)	323-333	JOE
sY254	AZFc	Yq11.23 (223)	375-386	JOE
SRY(sY14)	SRY	Yp11.31	455-470	JOE

Main Features:

- Sample to result in less than 4.5 hours
- Relative quantification between Chr.X and Chr.Y (AMXY marker) allows the pre-assessment of possible Klinefelter syndrome
- Precise and quick detection for all STSs in the same plot screen
- Contains all 20 markers determined by the European Molecular Genetics Quality Network guidelines
- SRY is a control marker for the testis determining factor on the short arm of the chromosome and enables detection of XX males arising from Y to X translocations

Ordering Information	
Cat No	**Description**
217100	100 tests
217050	50 tests
217025	25 tests

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