Genvinset® Thrombo Kits

ISO 13485 Quality Management for Medical Devices CERTIFIED

EUROPEAN UNION
European Regional Development Fund
A WAY TO MAKE EUROPE

Molecular determination of mutations and polymorphisms associated with thrombosis risk

Kits for detecting Factor II / Factor V mutations and MTHFR C677T / A1298C / PAI-1 / rs5985 polymorphisms by Real Time PCR using TaqMan® probes technology

About Genvinset® Thrombo kits

Thrombophilia consists in the predisposition to form blood clots, caused by an underlying hypercoagulation state attributable to inherited or acquired disorders of blood coagulation or fibrinolysis.

Thrombophilia is associated with the risk of deep venous thrombosis and/or venous thromboembolism. Since the discovery of antithrombin deficiency, many other conditions have been described so far, which have then allowed to currently detect an inherited or acquired predisposition in approximately 60-70% of patients with thromboembolic disorders. These prothrombotic risk factors mainly include qualitative or quantitative defects of endogenous coagulation factor inhibitors, increased concentration or function of clotting proteins, defects in the fibrinolytic system, impaired platelet function and hyperhomocysteinemia.

Intented use

Genvinset®
Factor II G20210A

Genvinset® Factor II G20210A is a semi-automated kit for the *in vitro* qualitative detection of the G20210A mutation (NCBI dbSNP rs1799963; NM_000506.5:c.*97G>A) in the prothrombin (*FII*) gene (OMIM: 176930) associated with thrombophilia risk, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (cardiologist), and taking into account the compatibility of the symptoms presented; abnormal clots that may cause long-term or life-threatening health problems, most often in the legs and lungs, and/or his family history (for example, a direct ascendant having had episodes of thrombosis) may be subject to the determination of the mutation in the FII gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Intented use

Genvinset®
Factor V G1691A

Genvinset® Factor V G1691A is a semi-automated kit for the *in vitro* qualitative detection of the G1691A mutation (NCBI dbSNP rs6025; NM_000130.5:c.1601G>A) in the factor V (*FV*) gene (OMIM: 612309) associated with thrombophilia risk, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (cardiologist), and taking into account the compatibility of the symptoms presented; abnormal clots that may cause long-term or life-threatening health problems, most often in the legs and lungs, and/or his family history (for example, a direct ascendant having had episodes of thrombosis) may be subject to the determination of the mutation in the FV gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Intented use

Genvinset®
FIIFV multiplex

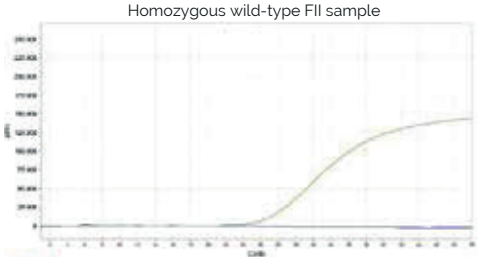
Genvinset® FIIFV multiplex is a semi-automated kit for the *in vitro* qualitative detection of the thrombophilia risk associated G20210A mutation (NCBI dbSNP rs1799963; NM_000506.5:c.*97G>A) in the prothrombin (*FII*) gene (OMIM: 176930) and G1691A mutation (NCBI dbSNP rs6025; NM_000130.5:c.1601G>A) in the factor V (*FV*) gene (OMIM: 612309) associated with thrombophilia risk in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (cardiologist, internist), and taking into account the compatibility of the symptoms presented; abnormal clots that may cause long-term or life-threatening health problems, most often in the legs and lungs, and/or his family history (for example, a direct ascendant having had episodes of thrombosis) may be subject to the determination of the mutation in the *FII* and *FV* genes. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

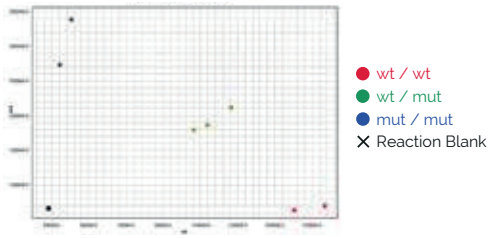
The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Results

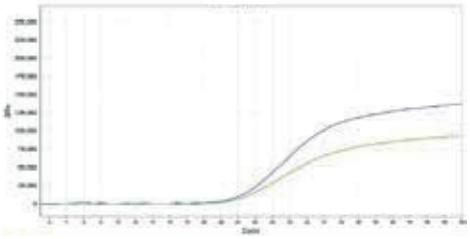
Homozygous wild-type FII sample



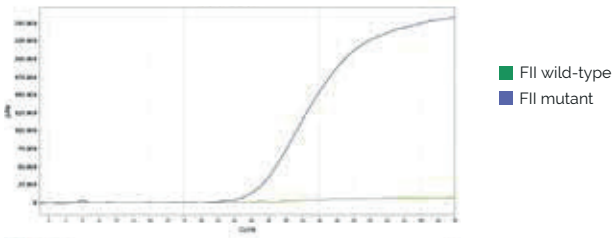
Allelic discrimination plot



Heterozygous FII sample

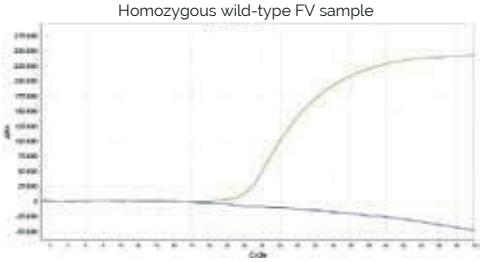


Homozygous mutant FII sample

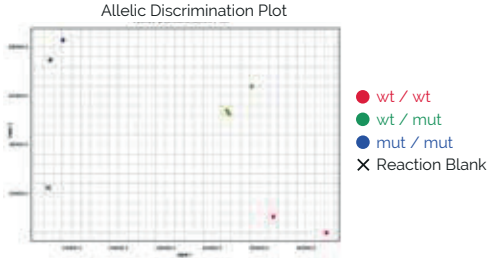


Results

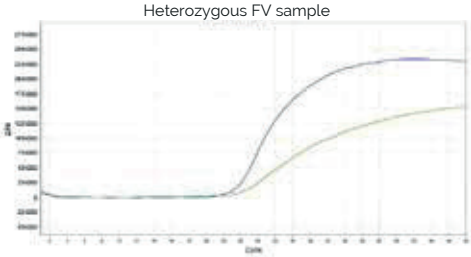
Homozygous wild-type FV sample



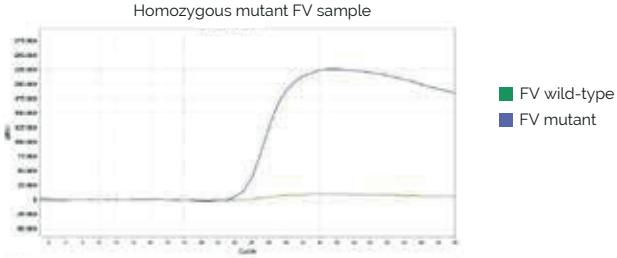
Allelic Discrimination Plot



Heterozygous FV sample

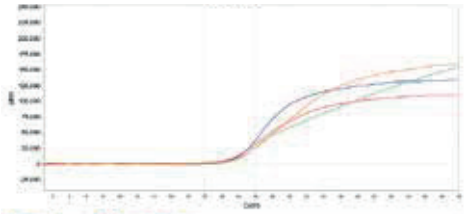


Homozygous mutant FV sample

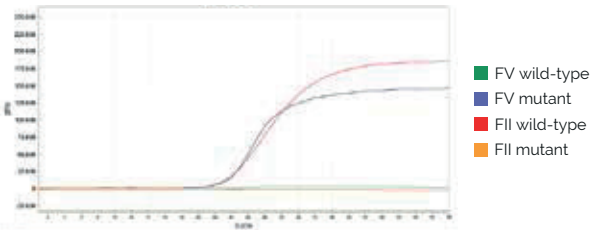


Results

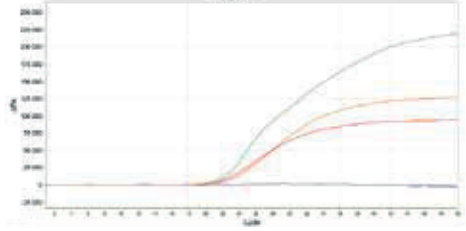
Heterozygous FV and FII sample



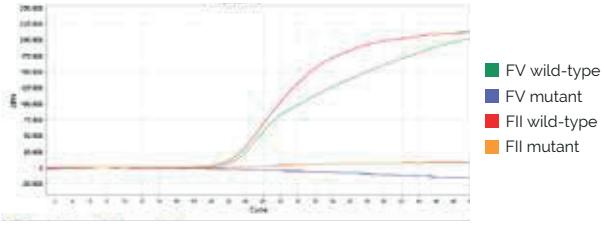
Homozygous mutant FV and homozygous wild-type FII sample



Homozygous wild-type FV and heterozygous FII sample



Homozygous wild-type FV and FII sample



Intented use

Genvinset®
MTHFR A1298C

Genvinset® MTHFR A1298C is a semi-automated kit for the *in vitro* qualitative detection of the A1298C polymorphism (NCBI dbSNP rs1801131; NM_001330358.2:c.1409A>C) in the Methylene tetrahydrofolate reductase (*MTHFR*) gene (OMIM: 607093) in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (reproductive clinician), and taking into account the compatibility of the symptoms presented; women suffering idiopathic recurrent pregnancy loss (RPL) may be subject to the determination of the polymorphism in the *MTHFR* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Intented use

Genvinset®
MTHFR C677T

Genvinset® MTHFR C677T is a semi-automated kit for the *in vitro* qualitative detection of the C677T polymorphism (NCBI dbSNP rs1801133; NM_001330358.2:c.788C>T) in the Methylene tetrahydrofolate reductase (*MTHFR*) gene (OMIM: 607093) in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (reproductive clinician), and taking into account the compatibility of the symptoms presented; women suffering idiopathic recurrent pregnancy loss (RPL) may be subject to the determination of the polymorphism in the *MTHFR* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Intented use

Genvinset®
MTHFR multiplex

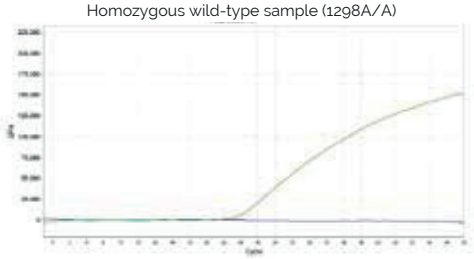
Genvinset® MTHFR multiplex is a semi-automated kit for the *in vitro* qualitative detection of the A1298C polymorphism (NCBI dbSNP rs1801131; NM_001330358.2:c.1409A>C) and the C677T polymorphism (NCBI dbSNP rs1801133; NM_001330358.2:c.788C>T) in the Methylene tetrahydrofolate reductase (*MTHFR*) gene (OMIM: 607093) in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (reproductive clinician), and taking into account the compatibility of the symptoms presented; women suffering idiopathic recurrent pregnancy loss (RPL) may be subject to the determination of the polymorphism in the *MTHFR* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

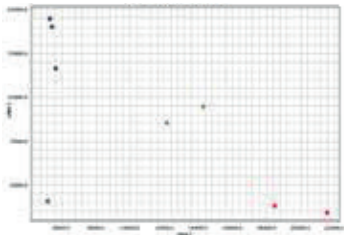
Results

Homozygous wild-type sample (1298A/A)



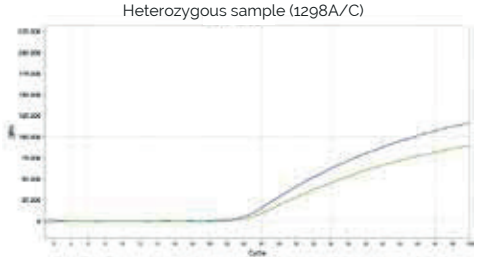
- Wild-type 1298A/A
- Mutant 1298C/C

Allelic Discrimination Plot



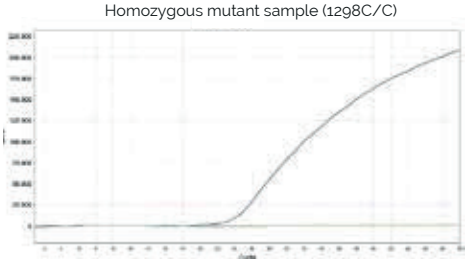
- wt/wt
- wt/mut
- mut/mut
- Reaction Blank

Heterozygous sample (1298A/C)



- Wild-type 1298A/A
- Mutant 1298C/C

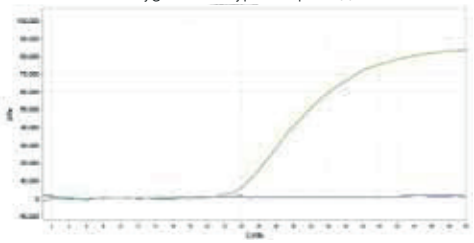
Homozygous mutant sample (1298C/C)



- Wild-type 1298A/A
- Mutant 1298C/C

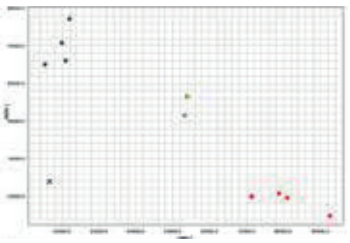
Results

Homozygous wild-type sample (677C/C)



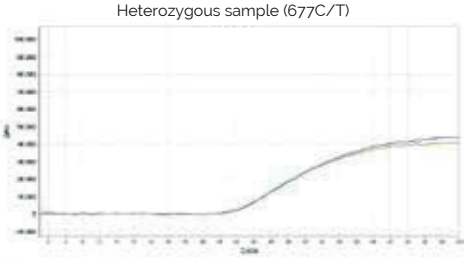
- Wild-type 677C/C
- Mutant 677T/T

Allelic Discrimination Plot



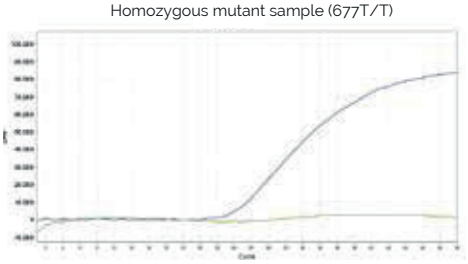
- wt/wt
- wt/mut
- mut/mut
- Reaction Blank

Heterozygous sample (677C/T)



- Wild-type 677C/C
- Mutant 677T/T

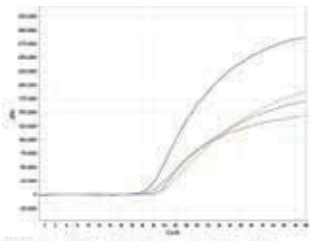
Homozygous mutant sample (677T/T)



- Wild-type 677C/C
- Mutant 677T/T

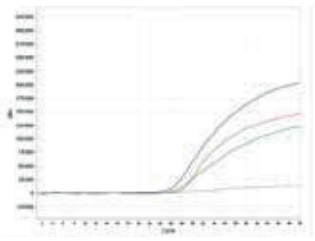
Results

Heterozygous 1298A/C and 677C/T sample



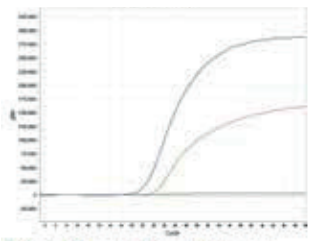
- Wild-type 1298A/A
- Mutant 1298C/C
- Wild-type 677C/C
- Mutant 677T/T

Heterozygous 1298A/C and homozygous wild-type 677C/C sample



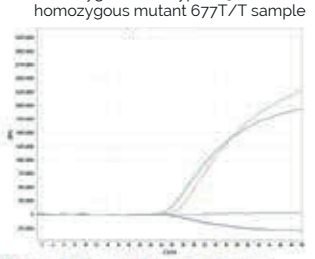
- Wild-type 1298A/A
- Mutant 1298C/C
- Wild-type 677C/C
- Mutant 677T/T

Homozygous mutant 1298C/C and homozygous wild-type 677C/C sample



- Wild-type 1298A/A
- Mutant 1298C/C
- Wild-type 677C/C
- Mutant 677T/T

Homozygous wild-type 1298A/A and homozygous mutant 677T/T sample



- Wild-type 1298A/A
- Mutant 1298C/C
- Wild-type 677C/C
- Mutant 677T/T

Genvinset®
PAI-1 4G/5G

Intended use

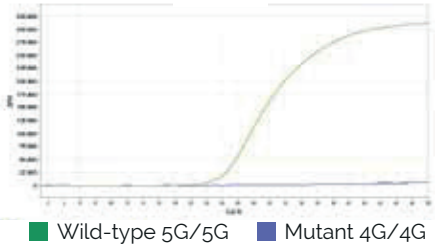
Genvinset® PAI-1 4G/5G is a semi-automated kit for the *in vitro* qualitative detection of the 4G/5G deletion (NCBI dbSNP rs1799762; NM_000602.5:c.-820G[(4_5)]) in the promoter region of the Plasminogen activator inhibitor 1 (*SERPINE1*) gene (OMIM: 173360) associated with thrombophilia risk, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (cardiologist), and taking into account the compatibility of the symptoms presented; abnormal clots that may cause long-term or life-threatening health problems, most often in the legs and lungs, and/or his family history (for example, a direct ascendant having had episodes of thrombosis) may be subject to the determination of the mutation in the *SERPINE1* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

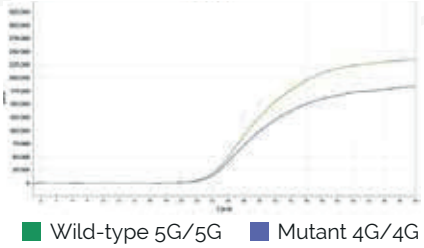
The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Results

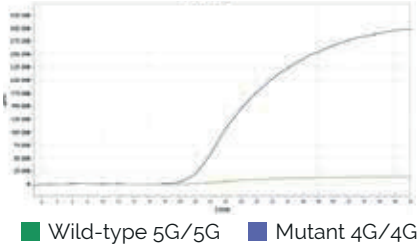
Homozygous wild-type sample (5G/5G)



Heterozygous sample (4G/5G)



Homozygous mutant sample (4G/4G)



Genvinset®
PAI-1/FXIII

Intended use

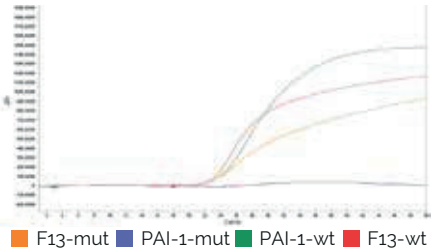
Genvinset® PAI-1/FXIII multiplex is a semi-automated kit for the *in vitro* qualitative detection of both the 4G/5G deletion (NCBI dbSNP rs1799762; NM_000602.5:c.-820G[(4_5)]) at the promoter region of the Plasminogen activator inhibitor 1 (*SERPINE1*) gene (OMIM: 173360) and the rs5985 (NCBI dbSNP rs5985; NM_000129.4:c.103G>T) at the coagulation factor XIII A chain (F13A1) gene (OMIM: 134570) associated with thrombophilia risk, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific hydrolysis probes.

The patient referred by the corresponding health specialist (cardiologist) and taking into account the compatibility of the symptoms presented; abnormal clots that may cause long-term or life-threatening health problems, most often in the legs and lungs, and/or his family history (for example, a direct ascendant having had episodes of thrombosis) may be subject to the determination of the mutation in the *SERPINE1* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

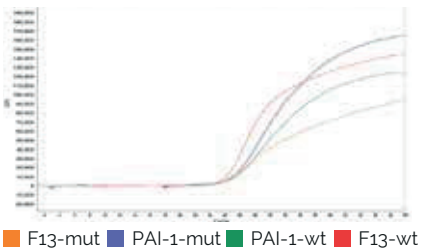
The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the instruction for use.

Results

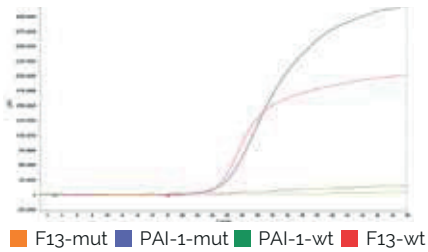
Homozygous wild-type sample (PAI/F13)



Heterozygous sample (PAI/F13)



Homozygous mutant sample (PAI/F13)



Workflow

qPCR Results

Product Information

FACTOR II

48 tests - Code NLM AA1608/48
CE-IVD certified

DESCRIPTION: Genvinset® Fattore II G20210A Real Time

FACTOR V

48 tests - Code NLM AA1609/48
CE-IVD certified

DESCRIPTION: Genvinset® Fattore V G1691A Real Time

MTHFR C677T

48 tests - Code NLM AA1610/48
CE-IVD certified

DESCRIPTION: Genvinset® MTHFR C677T Real Time

MTHFR A1298C

48 tests - Code NLM AA1616/48
CE-IVD certified

DESCRIPTION: Genvinset® MTHFR A1298C Real Time

FIIFV multiplex

24 tests - Code NLM AA1740/24
48 tests - Code NLM AA1740/48
CE-IVD certified

DESCRIPTION: Genvinset® FIIFV multiplex, 24T
Genvinset® FIIFV multiplex, 48T

MTHFR multiplex

24 tests - Code NLM AA1742/24
48 tests - Code NLM AA1742/48
CE-IVD certified

DESCRIPTION: Genvinset® MTHFR multiplex, 24T
Genvinset® MTHFR multiplex, 48T

PAI1 4G/5G

24 tests - Code NLM AA1743/24
48 tests - Code NLM AA1743/48
CE-IVD certified

DESCRIPTION: Genvinset® PAI-1 4G/5G, 24T
Genvinset® PAI-1 4G/5G, 48T

PAI-1/FXIII multiplex

24 tests - Code NLM AA1766/24
48 tests - Code NLM AA1766/48

DESCRIPTION: Genvinset® PAI-1/FXIII multiplex, 24T
Genvinset® PAI-1/FXIII multiplex, 48T

About BDR

Blackhills Diagnostic Resources S.L.U. (BDR) was founded in Zaragoza in 2012.

As a biotechnology company, BDR is focused on researching, developing, producing and selling in vitro diagnostic products for human diagnostics (CE-IVD).

BDR is specialized in Real Time PCR (qPCR), fragment analysis (QF-PCR) and NGS technologies, apart from having its own bioinformatics department in charge of the development of specific software for the analysis of results.

Our quality system complies with the most demanding international standards. We have all the required certifications for European manufacturers (EN ISO 13485 and CE certification in compliance with the Regulation (EU) 2017/746 as well as the Directive 98/79/EC). In addition, our continuous improvement program includes the yearly participation in External Quality Assessments (EQAs) through different international entities, always obtaining a high score.

EXCLUSIVE DISTRIBUTOR
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Partita Iva 08763060152
segreteria@nlm.it - **www.nlm.it**

BLACKHILLS DIAGNOSTIC RESOURCES S.L.U.
SRN: ES-MF-000001091 (Spain) · FIC GVS-THP REV.05

ISO 13485 Quality Management for Medical Devices CERTIFIED

Limitations

- Mutations or polymorphisms at annealing primer/probe sites are possible and may result in the lack of allele definition. Other technologies could be necessary to resolve the genotyping.
- Data and result interpretation should be revised by qualified personnel.
- This product is an auxiliary tool for the diagnosis of patients with suspected thrombophilia. Use these results in conjunction with clinical data and results of other tests performed on the patient.