Genvinset®
deltaF508

ISO 13485 Quality Management for Medical Devices CERTIFIED

EUROPEAN UNION
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Molecular determination of F508del alleles of the *CFTR* gene

Kit for detecting wild-type and/or F508del alleles in the *CFTR* gene associated with cystic fibrosis disease by Real-Time PCR using TaqMan® probes technology

About Genvinset® deltaF508

Code NLM AA1733/24

Cystic fibrosis (CF; OMIM #219700) is a genetic disorder that affects several different organs. Patients with CF generally suffer from obstructive lung disease with chronic bacterial infection, pancreatic enzyme insufficiency, and high salt content in their sweat. CF is caused by mutations in the *CFTR* gene that encodes the CFTR protein. This anion channel regulates water and ion transportation and maintains epithelial surface hydration.

Cystic fibrosis is typical of an autosomal recessive type of inheritance. Although deletion of F508 (F508del or ΔF508) constitutes ~90% of all CF cases, more than 2100 variants of the *CFTR* gene have been identified. The distribution and frequency of the variants vary in different regions and ethnic groups. In Europe, 82.4% of patients have at least one F508del mutation but the frequency of F508del is higher in Northern Europe than in Southern Europe.

The type II mutation F508del not only decreases the membrane expression but also impairs the channel function, thus causing a severe CF phenotype.

Intended use

Genvinset® deltaF508 is a semi-automated *in vitro* diagnostic kit for the qualitative detection of wt and /or F508del alleles (dbSNP: rs113993960 NM_000492.4:c.1521_1523delCTT) in the Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR*) gene (OMIM: 602421) associated with cystic fibrosis disease, in genomic DNA extracted from whole blood, by Real-Time PCR using specific TaqMan® probes technology.

Patients who can benefit from this determination are those referred by a specialist. 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.

Workflow

Product Information

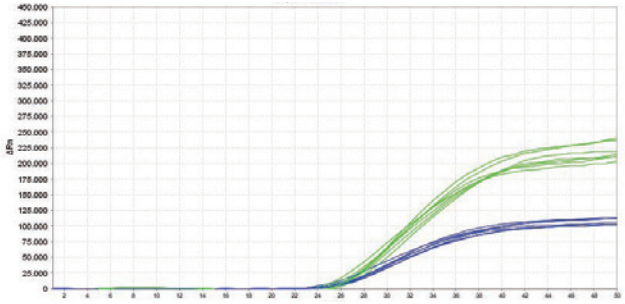
24 tests - Code **NLM AA1733/24**

DESCRIPTION: **Genvinset® deltaF508 24 test**

CE-IVD certified

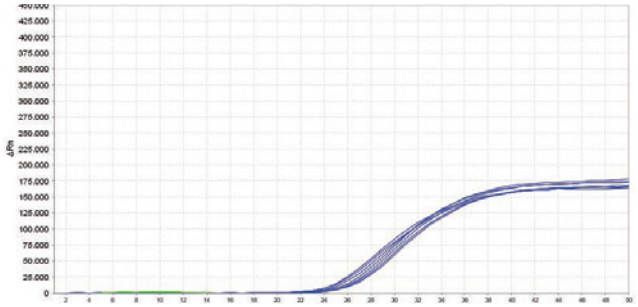
Results

Heterozygous samples for F508del amplification plot



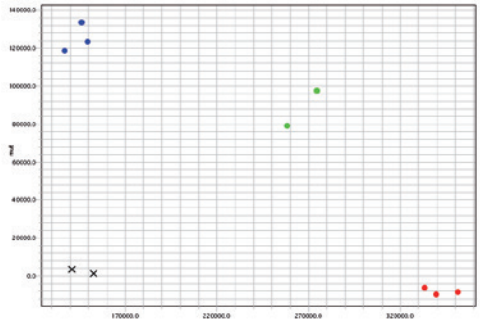
wild-type F508del

Homozygous samples for F508del amplification plot



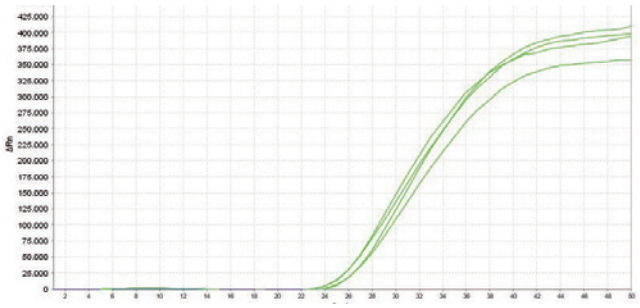
wild-type F508del

deltaF508 scatter plot



wt/wt wt/F508del F508del/F508del Reaction Blank

Homozygous wild-type amplification plot



wild-type F508del

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 cystic fibrosis. Use these results in conjunction with clinical data and results of other tests performed on the patient.

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BDR
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EXCLUSIVE DISTRIBUTOR
Nuclear Laser Medicine srl
Viale delle Industrie, 3 - 20049 Settala (Milan), ITALY
Tel. 02 952451 - Fax 02 95245237-8
Partita Iva 08763060152
segreteria@nlm.it - **www.nlm.it**