

CFTR31

SPOT ON INFINITI™

The Automated Multiplexing MDx Solution



Product Design

- ▶ The INFINITI™ CFTR31 Assay is designed to identify patients with CFTR genetic variants.
- ▶ The INFINITI CFTR31 Assay utilizes the CFTR31 Intellipac™, CFTR31 Amp Mix and CFTR31 BioFilmChip™ Microarray.
- ▶ The INFINITI CFTR31 Assay is automated by the 510(k) cleared INFINITI Analyzer.
- ▶ Clinical studies to support a 510(k) application are currently in progress.

Benefits



VERSATILITY	◆	Multiplexed determination of 25 genetic mutations and 6 reflex tests on one BioFilmChip Microarray
EFFICIENCY	◆	Automatic reporting of appropriate reflex test results without need for retesting of patient sample
AGILITY	◆	<i>Load N Go</i> automation with the INFINITI Analyzer
INTEGRITY	◆	Replicate determinations on single BioFilmChip Microarray ensure quality results

Genetic Variants

ACOG/ACMG recommended panel for population screening programs (April 2001)

G85E	R117H	I148T	621+1G>T	711+1G>T	1078delT
R334W	R347P	A455E	Delta I507	Delta F508	1717-1G>A
G542X	G551D	R553X	R560T	W1282X	N1303K
1898+1G>A	2184delA	2789+5G>A	3120+1G>A	R1162X	3659delC
3849+10kbC>T					

Reflex Tests

R117H heterozygous or homozygous variants will display reflex test Intron8 5T-7T-9T Delta I507 or Delta F508. Homozygous variants will display Exon10 I506V-I507V-F508C.

Sample Type and Volume

0.2 - 2.0 ml of peripheral whole blood in EDTA (purple-top) tube
50 ng DNA / reaction

Product Information

Product No.	Product Name	Description	Pack Size
01 106	INFINITI CFTR31 BioFilmChip	12 BioFilmChips/magazine	4 Magazines / pack
01 206	INFINITI CFTR31 Intellipac	24 tests/IntelliPac	2 Intellipac / pack
01 306	INFINITI CFTR31 Amp Mix	250 ul/vial	4 vials / pack

Please contact AutoGenomics to obtain product information and for product status updates.



Clinical Relevance

- ▶ The Cystic Fibrosis Transmembrane Regulator (CFTR) is a protein coded by the CF gene to regulate chloride channels within cells.
- ▶ Over 1300 variants have been identified, however most are extremely rare at less than 0.1%.¹
- ▶ Severity of CF disease varies widely with different genetic variants.
- ▶ CFTR Estimated Carrier Risk:²
 - One in 29 European Caucasians
 - One in 46 Hispanic Americans
 - One in 65 African Americans
 - One in 90 Asian Americans

Clinical Utility

- ▶ The ACOG/ACMG Working Group recommends use of a mutation panel that includes all CF Causing mutations with a frequency of $\geq 0.1\%$ in the General US Population.¹
- ▶ The F508del mutation accounts for 66% of all CFTR mutations in a pan-ethnic population.¹
- ▶ CFTR mutation testing is recommended for all pregnant couples and those contemplating pregnancy.²
- ▶ “The Working Group recommends no additions to the general population screening panel be made at this time.”¹
- ▶ Variants chosen should be associated with classical CF rather than with milder phenotypes because the decision making process largely impacts reproductive decisions.
- ▶ Clinical Utility for mutations additional to ACOG recommendations have not been established.

References

1. Wayne Grody et al., “Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel”, *Genetics In Medicine*, Sep./Oct. 2004, Vol.6, No.5, p.387-390.
2. Robert Desnick et al., “Laboratory Standards and Guidelines for Population-based Cystic Fibrosis Carrier Screening”, *Genetics in Medicine*, Mar./Apr. 2001, Vol.3, No.2, p.149-154.