

ELUCIGENE CF29v2

DNA TESTING IN CYSTIC FIBROSIS

Analysis of mutations in the CFTR gene is now performed as part of disease diagnosis and carrier testing protocols. Further to standard clinical techniques (e.g. sweat test), detection of a mutation in both copies of an individual's CFTR genes provides definitive confirmation of the disease state. While neonatal screening programs focus on IRT (immunoreactive trypsin) analysis, the sequential use of a DNA test (often for F508del) can increase diagnostic accuracy in individuals who screen IRT positive.

The large number of mutations identified to date and the probability of further novel CF mutations being discovered precludes routine testing for all mutations. However, advances in mutation detection now allow screening to identify carriers of the most prevalent mutations in selected populations.

ELUCIGENE CF29v2

Elucigene CF29v2 has been developed to provide laboratories with a simple and accurate means of routinely testing for 29 of the most prevalent mutations in the European Caucasian population and Ashkenazi Jews. Genotype information for F508del makes the test highly valuable in both disease diagnosis and screening applications. Elucigene CF Poly-T (Cat No. PT003B2) is available separately for reflex testing R117H positive patients to investigate CBAVD status.

Elucigene CF29v2 is supplied as four optimised ARMS primer multiplexes for the rapid detection of 29 common CFTR mutations.

MUTATIONS DETECTED BY CF29v2

PRIMER MIX A	PRIMER MIX B	PRIMER MIX C	PRIMER MIX D
D1152H	394delTT	A455E	3120+1G>A
1717-1G>A	621+1G>T	2183AA>G	2789+5G>A
G542X	S1251N	3659delC	1898+1G>A
W1282X	G551D	1078delT	711+1G>T
N1303K	R117H	DI507	G85E
F508del(M)	R1162X	R347P	2184delA
3849+10kbC>T	F508del(N)	R553X	R560T
	R334W	E60X	

ELUCIGENE KITS

All Elucigene kits comply with the EC Directive 98/79/CE, carry the CE mark and are manufactured within quality systems accredited to ISO9001:2008 and ISO13485:2003. Elucigene kits are available for:

Mutation detection

Gel based analysis

- Simple
- Cost efficient
- Easy analysis
- Accurate and reliable ARMS chemistry

Fluorescent based analysis

- Highly multiplexed
- Rapid analysis
- Compatible with ABI 3*** series Genetic Analyzers
- Accurate and reliable ARMS chemistry

Chromosome Aneuploidy

Elucigene QST*R range

- Quantitative Fluorescent PCR (QF-PCR) technique
- Simple set up – just add DNA
- Quick – results in less than 4 hours
- Flexible – range of kits available

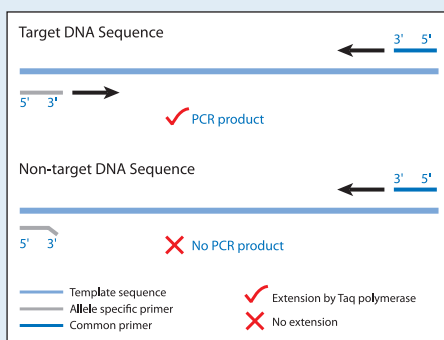
ELUCIGENE CF29v.2

ARMS® CHEMISTRY

The Elucigene range of products for simple and rapid analysis of human genetic disease utilizes the highly accurate Amplification Refractory Mutation System (ARMS) allele specific amplification technology:

- Reliable and accurate mutation detection
- Multiplexed, multiple mutations detected in a single analysis
- Simple protocol
- Fluorescent or gel based ethidium bromide detection
- Gel based assays require minimal equipment
- Extensively validated technology, many publications

ARMS PRINCIPLE



OTHER KITS

The Elucigene range of kits for the *in vitro* diagnosis of human genetic diseases, currently includes mutation detection for cystic fibrosis, thrombophilia, alpha-1-antitrypsin, Ashkenazi Jewish carrier screening and chromosomal aneuploidy

ELUCIGENE CYSTIC FIBROSIS SCREENING BY ARMS

Reliable and accurate mutation detection technology:

- Using ARMS allele specific amplification technology, the Elucigene CF29v2 kit allows 29 mutations to be detected in a single analysis, which saves 'hands-on' time and reagents compared to alternative methods.
- The simple protocol and minimal equipment requirement makes CF DNA analysis available to most laboratories with experience in PCR.
- For convenience and flexibility bulk optimised primer mixes are provided with AmpliTaq Gold®, Dilution Buffer, Gel Loading Dye and a DNA Control. Coloured PCR vials appropriate for 0.5mL or 0.2mL thermocyclers are also included.
- The use of this kit for *in vitro* human diagnostic testing is covered by a PCR licence and CFTR gene licence.

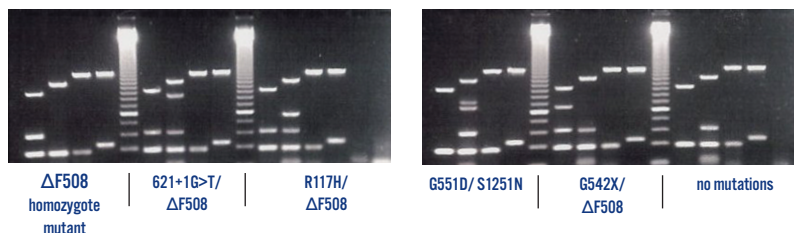
PROTOCOL AND TEST FORMAT

1. Prepare DNA from any source (protocols are provided for extraction of whole liquid blood and dried bloodstains).
2. Add 5µL of AmpliTaq Gold diluted in loading dye and 5µL of test sample or normal DNA control to each primer mix for PCR amplification.
3. Activate AmpliTaq Gold at 94°C (20 mins) and link to PCR (35 cycles)
94°C denaturation (30 secs)
58°C annealing (2 mins)
72°C extension (1 min)
72°C final extension (20 mins) (total time approximately 3.5hrs)

4. Run 3% agarose gel (90 mins)

Laboratories require only a thermal cycler, microfuge and gel electrophoresis equipment.

TYPICAL RESULTS



PRODUCT DETAILS

Cat. No. CF029B1/S or CF029B1/M* Elucigene CF29v2 Kit – 25 tests
Cat. No. CF029B2/S or CF029B2/M* Elucigene CF29v2 Kit – 50 tests
*supplied with 0.2ml(S) or 0.5ml(M) PCR vials

Elucigene kits and reagents are developed and manufactured within quality systems accredited to ISO 9001 and ISO 13485 and comply with the European Community Directive 98/79/EC. ELUCIGENE is a trademark of Gen-Probe Life Sciences Ltd. ARMS® is a registered trademark of AstraZeneca UK Ltd and is used under licence. The ARMS technology is the subject of European Patent No. 0332435, US Patent No. 5599890 and corresponding worldwide patents. AmpliTaq Gold is a registered trademark of Roche Molecular Systems, Inc.

