



Diagnostic kits and reagents

CF Screening of Jewish Populations

Cystic Fibrosis Screening of Jewish Populations

It is estimated that there are approximately 13 million Jews in the world today, the vast majority of whom live in either the United States of America or Israel. The Jewish population is identified through its history and ethnic origin as either Ashkenazi or Sephardic. The Ashkenazi Jews originate primarily from central and eastern Europe, the Sephardic Jews originate from the mediterranean areas of Europe. The Ashkenazi's are by far the largest population of Jews, 90% of Jews in the United States are of Ashkenazi origin. Although these two populations have distinct and different origins, their culture and values have resulted in limited variance in their respective gene pools. Unfortunately the lack of genetic variance has led to the high incidence of inherited diseases in the Ashkenazim and Sephardim.

Cystic Fibrosis is the most common autosomal recessive disease in the Caucasian population. There are in excess of 1000 mutations identified in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, although the single most common mutation, ΔF508, accounts for approximately 70% of all CF chromosomes in Caucasians. In Jewish populations there is significantly less variation in the number of CFTR mutations observed, 98% of Ashkenazi CF chromosomes are identified by only 6 mutations. Similarly only 6 CFTR mutations account for 100% of Tunisian Sephardic Jewish CF chromosomes. Therefore cystic fibrosis genetic screening can be extremely informative and effective in Jewish populations.

ELUCIGENE™ CF7 and ELUCIGENE™ CF-MEP

ELUCIGENE™ CF7 and ELUCIGENE™ CF-MEP have been developed specifically for effective cystic fibrosis screening in Jewish populations and each gives excellent detection coverage of CF chromosomes. ELUCIGENE™ CF7 is particularly effective for CF screening in Ashkenazi's, while ELUCIGENE™ CF-MEP detects the five most prevalent CFTR mutations in Sephardic Jews. When used together a total of 11 different mutations can be identified giving an excellent coverage in Jews from various ethnic backgrounds.

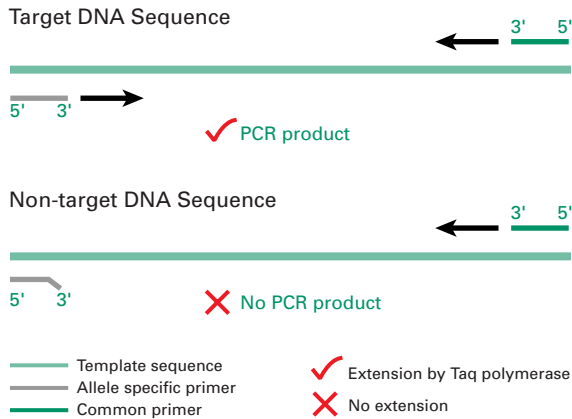
CFTR Mutation Distribution Among Jews of Different Ethnic Groups¹ and Mutation Coverage Utilising ELUCIGENE™ CF7 and ELUCIGENE™ CF-MEP

	ASHKENAZI	SEPHARDI			
ELUCIGENE™ CF-MEP					
Mutation		Tunisia	Libya	Turkey	Morocco
*D1152H					6%
W1089X				10%	
G85E				30%	
405+1G>A		48%			
S549R(T>G)			18%		6%
ELUCIGENE™ CF7	98%	52%	73%	50%	78%
Total Coverage	98%	100%	91%	90%	84%

¹Kerem et al. Highly variable incidence of cystic fibrosis and mutation distribution among different Jewish ethnic groups in Israel. Human Genetics 96: 193-197 (1995).

*D1152H is also included in ELUCIGENE™ CF7

ELUCIGENE™ Cystic Fibrosis Screening in Ashkenazi and Sephardic Jews



Reliable and accurate mutation detection technology:-
Using ARMS™ allele specific amplification technology, the ELUCIGENE™ CF7 and ELUCIGENE™ CF-MEP kits allow effective and simple CFTR mutation analysis in Jewish populations.

The simple protocol and minimal equipment requirement makes DNA analysis of the the CFTR gene available to most laboratories with experience in PCR.

Each kit contains an optimised primer solution, a DNA control, AmpliTaq Gold®, Dilution Buffer and Loading Dye. Laboratories require only a thermal cycler, microfuge and gel electrophoresis equipment.

The use of each of these kits for *in vitro* diagnostic testing is covered by a PCR licence and CFTR gene licence.

elucigene CF7

ELUCIGENE™ CF7 provides laboratories with a simple and accurate means of routinely testing for the most prevalent mutations found in the Ashkenazi Jewish population.

W1282X
ΔF508
G542X
3849+10kb C>T
N1303K
1717-1G>A
D1152H

elucigene CF-MEP

ELUCIGENE™ CF-MEP detects the 5 most prevalent CFTR mutations in the Sephardic Jewish population.

W1089X
G85E
D1152H
S549R(T>G)
405+1G>A

Product and Contact Details

Cat. No. CF007B4 ELUCIGENE™ CF7 – 100 tests
Cat. No. YF005B4 ELUCIGENE™ CF-MEP – 100 tests

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ELUCIGENE™ CF7/CF-MEP

% CF Chromosome coverage in the Jewish Population



* Estivill, X. et al (1997) Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations, Human Mutation, 10: 135-154.

** Based on Abelovich, D. et al (1992) Screening for five mutations detects 97% of Cystic Fibrosis (CF) chromosomes and predicts a carrier frequency of 1:29 in the Jewish Ashkenazi population, Am. J. Hum. Genet. 51:951-956.

