مختبر فرايبورج الطبي الشرق الأوسط (ذ.م.م)
FREIBURG MEDICAL LABORATORY MIDDLE EAST (L.L.C.)

Recommended approach for genetic testing for Cystic Fibrosis:

1. **Sequencing of the complete gene (CFTR)**
   Overall detection rate: ~95%
   (The remaining 5% can be covered by MLPA)
   TAT: up to 8 weeks
   Material: 2 x EDTA blood

2. **Initial screening for the 50 most common mutations and 3 polymorphisms**
   Detection rate:
   85-90% in European patients
   Approx. 40% in Arabic patients
   (Offered Middle East panels have a detection rate of approx. 50%.)
   TAT: up to 3 weeks
   Material: 1 x EDTA blood

CFTRdele2,3	R334W	R553X	R1066C
E60X	R347P	R560T	Y1092X(C>A)
P67L	R347H	1811+1.6kbA>G	M1101K
G85E	A455E	1898+1G>A	D1152H
394delTT	I507del	2143delT	R1158X
444delA	F508del	2184delA	R1162X
R117C	1677delTA	2347delG	3659delC
R117H	V520F	W846X	3849+10KbC>T
Y122X	1717-1G>A	2789+5G>A	S1251N
621+1G>T	G542X	Q890X	3905insT
711+1G>T	S549N	3120+1G>A	W1282X
L206W	S549R (T>G)	3272-26A>G	N1303K
1078delT	G551D	Intron 8 polyT (5T/7T/9T) and TGn	

In case of any questions please contact FREIBURG MEDICAL LABORATORY / www.fml-dubai.com
Tel: +971-4-396 2227, Fax: +971-4-396 2228, E-mail: info@fml-dubai.com