

## Identification of 50 common cystic fibrosis European mutations

Yourgene® Cystic Fibrosis Base is commercially available pan-European cystic fibrosis testing kit designed specifically to address the most common mutations found across populations of European origin. The assay identifies 50 mutations in total and also analyses the intron 8 polyT tract with accurate measurement of the adjacent TG repeat.

### Key Benefits

- High Quality CE-IVD marked product
- Simple to use
  - Minimal hands on time and ease of use
  - Fits comfortably into existing patient care pathways, with low DNA input volume
- Rapid turnaround time
- Clinically relevant with 80-90% coverage of the most prevalent mutations (region dependent)
  - Population-specific bolt on panels available
- Simple Data Interpretation
  - Consistency across results – ease of interpretation
  - Data review using GeneMapper™ and GeneMarker® software (no data transfer required)



### Performance you can rely on

# 100%

## Accuracy, Repeatability & Reproducibility

	Positive Percent Agreement (PPA)	Negative Percent Agreement (NPA)	Overall Agreement (OA)
Accuracy	100%	100%	100%
Repeatability	100%	100%	100%
Reproducibility	100%	100%	100%

Accuracy was compared to Sanger sequencing. Accuracy of the Yourgene® Cystic Fibrosis Base assay (previously known as the Elucigene® CF-EU2v1 assay) was assessed by evaluating 159 samples representing a wide variety of CFTR gene variants from 4 separate sources.

## Mutations detected by Yourgene® Cystic Fibrosis Base

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)	TGn

## Ordering Information

Assay	Catalogue Number	Product Description	Kit Size
Yourgene® Cystic Fibrosis Base	CFEU2v1	Yourgene® Cystic Fibrosis Base for qualitative detection of 50 Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutations, common in European populations	50 Tests
Yourgene® Cystic Fibrosis Lite	CF4HTB1	Yourgene® Cystic Fibrosis Base Lite for qualitative detection of 4 Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutations, common in European populations	25 Tests
Yourgene® Cystic Fibrosis Base France	CF30v2	Yourgene® Cystic Fibrosis Base France for qualitative detection of 29 Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutations, common in French populations	25 Tests
Yourgene® Cystic Fibrosis Base Germany	CF1DEB2	Yourgene® Cystic Fibrosis Base Germany for qualitative detection of 31 Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutations, outlined by the G-BA for newborn screening in Germany	50 Tests
Yourgene® Cystic Fibrosis Plus UK	CF1UKB1	Yourgene® Cystic Fibrosis Plus UK can be used with Yourgene Cystic Fibrosis Base to increase mutation coverage for an additional 11 mutations common in British populations	25 Tests
Yourgene® Cystic Fibrosis Plus Iberian	CFIBNB1	Yourgene® Cystic Fibrosis Plus Iberian can be used with Yourgene Cystic Fibrosis Base to increase mutation coverage for an additional 12 mutations common in Iberian populations	25 Tests
Yourgene® Cystic Fibrosis Plus Italian	CFITAB1	Yourgene® Cystic Fibrosis Plus Italian can be used with Yourgene Cystic Fibrosis Base to increase mutation coverage for an additional 14 mutations common in Italian populations	25 Tests

Yourgene® Cystic Fibrosis range previously known as Elucigene® Cystic Fibrosis range

## About YOURGENE HEALTH

Yourgene Health is an international molecular diagnostics group which develops integrated genomic technologies and services enabling genomic medicine.

### For further information

**Tel:** +44 (0) 161 669 8122

**Email:** [info@yourgenehealth.com](mailto:info@yourgenehealth.com)



**Yourgene Health**  
Skelton House  
Lloyd Street North  
Manchester Science Park  
Manchester  
M15 6SH  
United Kingdom

[yourgenehealth.com](http://yourgenehealth.com)

