

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 <i>Base</i>	CFEU2v1	Identifies 50 of the most frequently observed CFTR gene mutations within populations of European origin	50 Tests
Yourgene® Cystic Fibrosis <i>Lite</i>	CF4HTB1	Bolt on to detect the 4 most common mutations in the European population	25 Tests
Yourgene® Cystic Fibrosis <i>Base France</i>	CF30v2	Bolt on to detect the 29 most common mutations in the French population	25 Tests
Yourgene® Cystic Fibrosis <i>Base Germany</i>	CF1DEB2	Bolt on to detect the 31 mutations outlined by the G-BA for newborn screening in Germany	50 Tests
Yourgene® Cystic Fibrosis <i>Base Germany</i>	CF1DEBX	Bolt on to detect the 31 mutations outlined by the G-BA for newborn screening in Germany	10 Tests
Yourgene® Cystic Fibrosis <i>Plus UK</i>	CF1UKB1	Bolt on to Yourgene® Cystic Fibrosis <i>Base</i> to increase the mutation coverage for the British population	25 Tests
Yourgene® Cystic Fibrosis <i>Plus Iberian</i>	CFIBNB1	Bolt on to Yourgene® Cystic Fibrosis <i>Base</i> to increase the mutation coverage for the Iberian population	25 Tests
Yourgene® Cystic Fibrosis <i>Plus Italian</i>	CFITAB1	Bolt on to Yourgene® Cystic Fibrosis <i>Base</i> to increase the mutation coverage for the Italian population	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.

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