

# **CERTIFICATE**

Survey of 07 November 2018

You have fulfilled the requirements of the External Quality Assessment with the following analysis

# Set 09 - CFTR Common Mutations (778):

Validity 24 months:

CFTR Gen (1677 delTA) (R: B5) CFTR Gen (2143 del T) (R: B5)

CFTR Gen (2184 delA) (R: B5) CFTR Gen (3905insT) (R: B5)

CFTR Gen (A455E) (R: B5) CFTR Gen (delta F 508) (R: B5)

CFTR gene (1078 del T) (R: B5)

CFTR gene (1717-1 G>A) (R: B5)

CFTR gene (2789+5 G>A) (R: B5) CFTR gene (3272-26 A>G) (R: B5)

CFTR gene (3659 del C) (R: B5)

CFTR gene (394 del TT) (R: B5)

CFTR Gen (E 60 X) (R: B5)

CFTR gene (621+1 G>T) (R: B5)

CFTR gene (dele2,3 (21kb)) (R: B5)

CFTR gene (G 542 X) (R: B5) CFTR gene (G 551 D) (R: B5)

CFTR gene (G 85 E) (R: B5) CFTR gene (I 507 del) (R: B5)

CFTR gene (Intron 8 (Polypyrimidine

Tract)) (R: B5)

CFTR gene (M 1101 K) (R: B5) CFTR gene (N 1303 K) (R: B5) CFTR gene (R 1162 X) (R: B5)

CFTR gene (3849+10kb C>T) (R: B5) CFTR gene (R 117 H) (R: B5) CFTR gene (R 334 W) (R: B5) CFTR gene (R 347 P/H) (R: B5)

CFTR gene (R 553 X) (R: B5)

(R) analysis is subject to the RiliBÄK

Participant: 43733

CG Cytogenomics Medical Laboratory

**CG** Cytogenomics **Byzantioy 149** 14233 Nea Ionia Athens

DAkkS Deutsche Akkreditierungsstelle D-EP-15027-02-00



CFTR gene (W 1282 X) (R: B5)

CFTR gene (Y 1092 X (C>A)) (R: B5)

Düsseldorf, 29 November 2018

Prof. Dr. med. Michael Spannagl (Head of Reference Institution)

Prof. Dr. med. Friedrich Maly (Adviser)



# Set 09 - CFTR Common Mutations

# CFTR Gen (1677 delTA) (N = 29, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt (35)	total
others	29 •	29

Sample 62

Collective	No del / No del = wt (35)	total
others	29 •	29

Sample 63

Collective	No del / No del = wt (35)	total
others	29 •	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR Gen (2143 del T) (N = 33, Rate of success: 100%)

Sample 61

Sample of		
Collective	No del / No del = wt (35)	total
others	33 •	33

Sample 62

Collective	No del / No del = wt (35)	total
others	33 •	33

Sample 63

Collective	No del / No del = wt (35)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR Gen (2184 delA) (N = 31, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt (35)	total
others	31 •	31

Sample 62

Collective	No del / No del = wt (35)	total
others	31 •	31

Sample 65		
Collective	No del / No del = wt (35)	total
others	31 •	31

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# **CFTR Gen (3905insT)** (N = 31, Rate of success: 100%)

Sample 61

Collective	No ins / No ins =wt (22)	total
others	31 •	31

Sample 62

Jampie 02		
Collective	No ins / No ins ⇒wt (22)	total
others	31 •	31

Sample 63		
Collective	No ins / No ins =wt (22)	total
others	31 .	31



# CFTR Gen (A455E) (N = 31, Rate of success: 100%)

Sample 61

Collective	AA 455 = wt (219)	total
others	31 •	31

Sample 63

Collective	AA 455 = wt (219)	total
others	31 •	31

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

#### CFTR Gen (delta F 508) (N = 35, Rate of success: 100%)

Sample 61

Collective	Wild Type (119)	total
others	35 •	35

Sample 63

Sample 05		
Collective	Wild Type (119)	total
others	35 •	35

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

#### CFTR gene (1078 del T) (N = 33, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt (35)	total
others	33 •	33

Sample 63

Collective	No del / No del = wt (35)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (1717-1 G>A) (N = 33, Rate of success: 100%)

Sample 61

Collective	GG = wt (140)	total
others	33 •	33

Sample 63

Collective	GG = wt (140)	total
others	33 •	33

Sample 62

Sample 62 Collective

Sample 62 Collective

Sample 62

Collective

others

others

others

AA 455 = wt

(219)

31 •

Wild Type

(119) 35 •

No del / No del = wt

(35)

33 •

total

31

total

35

total

33

Collective	GG = wt (140)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

#### CFTR gene (2789+5 G>A) (N = 33, Rate of success: 100%)

Sample 61

Sample 61		
Collective	GG = wt (149)	total
others	33 •	33

Sample 62

Jample 02		
Collective	GG = wt (149)	total
others	33 •	33



Sample 63

Collective	GG = wt (149)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (3272-26 A>G) (N = 33, Rate of success: 100%)

Sample 61

Collective	AA = wt (157)	total
others	33 •	33

Sample	62
--------	----

Collective	AA = wt (157)	total
others	33 •	33

Sample 63

Collective	AA = wt (157)	total	
others	33 •	33	

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (3659 del C) (N = 33, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt (35)	total
others	33 •	33

Sam	n	P	6
Juili	Ρ.		-

Sample 62		
Collective	No del / No del = wt (35)	total
others	33 •	33

Sample 63

Collective	No del / No del = wt (35)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (3849+10kb C>T) (N = 32, Rate of success: 90,6%)

Sample 61

Collective	CC = wt (143)	total
others	32 •	32

Sample 02			
Collective	CC = wt (143)	TT (145)	total
others	31 •	1	32

Sample 63

Sample 03				
Collective	CC = wt (143)	CT (144)	TT (145)	total
others	1	2	29 •	32

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (394 del TT) (N = 24, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt	total
	(35)	
others	24 •	24

Sample 62		
Collective	No del / No del = wt (35)	total
others	24 •	24

Sample 63

Collective	No del / No del = wt (35)	total
others	24 •	24



# CFTR Gen (E 60 X) (N = 30, Rate of success: 100%)

Sample 61

Collective	EE 60 = wt (216)	total
others	30 •	30

Sample 63

Jumpie 05		
Collective	EE 60 = wt (216)	total
others	30 •	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

#### CFTR gene (621+1 G>T) (N = 33, Rate of success: 100%)

Sample 61

Collective	GG = wt (180)	total
others -	33 •	33

Sample 63

Sample 63		
Collective	GG = wt (180)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (dele2,3 (21kb)) (N = 31, Rate of success: 100%)

Sample 61

Collective	No Del = wt (36)	total
others	31 •	31

Sample 63

Collective	No Del = wt (36)	total
others	31 •	31

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (G 542 X) (N = 33, Rate of success: 100%)

Sample 61

Collective	GG 542 = wt (125)	total
others	33 •	33

Sample 63

Collective	GG 542 = wt (125)	total
others	33 •	33

Sample 62

Sample 62 Collective

Sample 62 Collective

others

Sample 62 Collective

others

others

EE 60 = wt

(216)

30 •

GG = wt

(180)

No Del = wt

(36)

31 •

33 •

total

30

total

33

total

31

Collective	GG 542 = wt (125)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (G 551 D) (N = 33, Rate of success: 100%)

Sample 61

Collective	GG 551 = wt (130)	total
others	33 •	33

Sample 62

Sample 62		
Collective	GG 551 = wt (130)	total
others	33 •	33



Sample 63

Collective	GG 551 = wt (130)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (G 85 E) (N = 33, Rate of success: 100%)

Sample 61

Collective	GG 85 = wt (182)	total
others	33 •	33

Sample 62

Collective	GG 85 = wt (182)	total
others	33 •	33

Sample 63

Collective	GG 85 = wt (182)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (I 507 del) (N = 32, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt (35)	total
others	32 •	32

Sample 62

Collective	No del / No del = wt (35)	total
others	32 •	32

Sample 63

Collective	No del / No del = wt (35)	total
others	32 •	32

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (Intron 8 (Polypyrimidine Tract)) (N = 26, Rate of success: 100%)

Sample 61

Collective	7T / 7T (193)	total
others	26 •	26

Sample 6

Sample 02		
Collective	7T / 7T (193)	total
others	26 •	26

Sample 63

Collective	7T / 7T (193)	total
others	26 •	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (M 1101 K) (N = 28, Rate of success: 100%)

Sample 61

Collective	MM 1101 = wt (170)	total
others	28 •	28

Sample 62

Sample 62		
Collective	MM 1101 = wt (170)	total
others	28 •	28

Sample 63

Collective	MM 1101 = wt (170)	total
others	28 •	28



# CFTR gene (N 1303 K) (N = 33, Rate of success: 100%)

Sample 61

Collective	NN 1303 =wt (122)	total
others	33 •	33

Sample 62

Collective	NN 1303 =wt (122)	total
others	33 •	33

Sample 63

Collective	NN 1303 =wt (122)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (R 1162 X) (N = 33, Rate of success: 100%)

Sample 61

Collective	RR 1162 = wt (173)	total
others	33 •	33

Sample 62

Collective	RR 1162 = wt (173)	total
others	33 •	33

Sample 63

Collective	RR 1162 = wt (173)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (R 117 H) (N = 26, Rate of success: 100%)

Sample 61

Collective	RR 117 = wt (146)	total
others	26 •	26

Sample 62

Collective	RR 117 = wt (146)	total
others	26 •	26

Sample 63

Sample 03		
Collective	RR 117 = wt (146)	total
others	26 •	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (R 334 W) (N = 33, Rate of success: 100%)

Sample 61

Collective	RR 334 = wt (161)	total
others	33 •	33

Sample 62

21		
Collective	RR 334 = wt (161)	total
others	33 •	33

Sample 63

Collective	RR 334 = wt (161)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (R 347 P/H) (N = 33, Rate of success: 100%)

Sample 61

Sample of		
Collective	RR 347 = wt (135)	total
others	33 •	33

Sample 62

Sample 62		
Collective	RR 347 = wt (135)	total
others	33 •	33



Sample 63

Collective	RR 347 = wt (135)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (R 553 X) (N = 33, Rate of success: 100%)

Sample 61

Collective	RR 553 = wt (133)	total
others	33 •	33

Sample 62

Collective	RR 553 = wt (133)	total
others	33 •	33

Sample 63

Collective	RR 553 = wt (133)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

## CFTR gene (W 1282 X) (N = 33, Rate of success: 100%)

Sample 61

Collective	WW 1282 = wt (158)	total
others	33 •	33

Sample 62

Collective	WW 1282 = wt (158)	total
others	33 •	33

Sample 63

Collective	WW 1282 = wt (158)	total
others	33 •	33

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

# CFTR gene (Y 1092 X (C>A)) (N = 29, Rate of success: 100%)

Sample 61

Collective	YY 1092 = wt (167)	total
others	29 •	29

Sample 6

Sample 02		
Collective	YY 1092 = wt (167)	total
others	29 •	29

Sample 63

Collective	YY 1092 = wt (167)	total
others	29 •	29

