

# CERTIFICATE

Survey of 24 March 2023

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

## Molecular Genetics 28 - CFTR Common Mutations (778):

Validity 24 months:

CFTR Gen (1677 delTA) (R: B5)	CFTR gene (394 del TT) (R: B5)	CFTR gene (Y 1092 X (C>A)) (R: B5)
CFTR Gen (2143 del T) (R: B5)	CFTR gene (621+1 G>T) (R: B5)	CFTR gene (dele2,3 (21kb)) (R: B5)
CFTR Gen (2184 delA) (R: B5)	CFTR gene (G 542 X) (R: B5)	
CFTR Gen (3905insT) (R: B5)	CFTR gene (G 551 D) (R: B5)	
CFTR Gen (A455E) (R: B5)	CFTR gene (G 85 E) (R: B5)	
CFTR Gen (E 60 X) (R: B5)	CFTR gene (I 507 del) (R: B5)	
CFTR Gen (Intron 8) (R: B5)	CFTR gene (M 1101 K) (R: B5)	
CFTR Gen (delta F 508) (R: B5)	CFTR gene (N 1303 K) (R: B5)	
CFTR gene (1078 del T) (R: B5)	CFTR gene (R 1162 X) (R: B5)	
CFTR gene (1717-1 G>A) (R: B5)	CFTR gene (R 117 H) (R: B5)	
CFTR gene (2789+5 G>A) (R: B5)	CFTR gene (R 334 W) (R: B5)	
CFTR gene (3272-26 A>G) (R: B5)	CFTR gene (R 347 P/H) (R: B5)	
CFTR gene (3659 del C) (R: B5)	CFTR gene (R 553 X) (R: B5)	
CFTR gene (3849+10kb C>T) (R: B5)	CFTR gene (W 1282 X) (R: B5)	

(R) analysis is subject to the RiliBÄK

### Participant:

43733  
CG Cytogenomics Medical Laboratory  
CG Cytogenomics  
313 Hrakleiou Av.  
14122 Neo Irakleio



Düsseldorf, 05 May 2023

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

PD Dr. rer. nat. Julia Hentschel  
(Adviser)

# CERTIFICATE OF PARTICIPATION

Survey of 24 March 2023

You have participated in the External Quality Assessment with the following analysis

## Molecular Genetics 28 - CFTR Common Mutations (778):

CFTR Gen (1677 delTA) (R: B5)	CFTR gene (621+1 G>T) (R: B5)
CFTR Gen (2143 del T) (R: B5)	CFTR gene (G 542 X) (R: B5)
CFTR Gen (2184 delA) (R: B5)	CFTR gene (G 551 D) (R: B5)
CFTR Gen (3905insT) (R: B5)	CFTR gene (G 85 E) (R: B5)
CFTR Gen (A455E) (R: B5)	CFTR gene (I 507 del) (R: B5)
CFTR Gen (E 60 X) (R: B5)	CFTR gene (M 1101 K) (R: B5)
CFTR Gen (Intron 8) (R: B5)	CFTR gene (N 1303 K) (R: B5)
CFTR Gen (delta F 508) (R: B5)	CFTR gene (R 1162 X) (R: B5)
CFTR gene (1078 del T) (R: B5)	CFTR gene (R 117 H) (R: B5)
CFTR gene (1717-1 G>A) (R: B5)	CFTR gene (R 334 W) (R: B5)
CFTR gene (2789+5 G>A) (R: B5)	CFTR gene (R 347 P/H) (R: B5)
CFTR gene (3272-26 A>G) (R: B5)	CFTR gene (R 553 X) (R: B5)
CFTR gene (3659 del C) (R: B5)	CFTR gene (W 1282 X) (R: B5)
CFTR gene (3849+10kb C>T) (R: B5)	CFTR gene (Y 1092 X (C>A)) (R: B5)
CFTR gene (394 del TT) (R: B5)	CFTR gene (dele2,3 (21kb)) (R: B5)

(R) analysis is subject to the RiliBÄK

Participant:  
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(Head of Reference Institution)

PD Dr. rer. nat. Julia Hentschel  
(Adviser)

## Listing and evaluation of the results

43733: CG Cytogenomics Medical Laboratory  
CG Cytogenomics

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### Molecular Genetics 28 - CFTR Common Mutations

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CFTR Gen (1677 delTA)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (2143 del T)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (2184 delA)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (3905insT)	11	9999	ZY		No ins / No ins =wt	No ins / No ins =wt	M	+
	12				No ins / No ins =wt	No ins / No ins =wt	M	+
CFTR Gen (A455E)	11	9999	ZY		AA 455 = wt	AA 455 = wt	M	+
	12				AA 455 = wt	AA 455 = wt	M	+
CFTR Gen (E 60 X)	11	9999	ZY		E 60 X	E 60 X	M	+
	12				EE 60 = wt	EE 60 = wt	M	+
CFTR Gen (Intron 8)	11	9999	ZY		9T / 7T	9T / 7T	M	+
	12				7T / 7T	7T / 7T	M	+
CFTR Gen (delta F 508)	11	271	ZY		delta F508 Het	delta F508 Het	M	+
	12				Wild Type	Wild Type	M	+
CFTR gene (1078 del T)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (1717-1 G>A)	11	9999	ZY		GG = wt	GG = wt	M	+
	12				GG = wt	GG = wt	M	+
CFTR gene (2789+5 G>A)	11	9999	ZY		GG = wt	GG = wt	M	+
	12				GG = wt	GG = wt	M	+
CFTR gene (3272-26 A>G)	11	9999	ZY		AA = wt	AA = wt	M	+
	12				AA = wt	AA = wt	M	+
CFTR gene (3659 del C)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (3849+10kb C>T)	11	9999	ZY		CC = wt	CC = wt	M	+
	12				CC = wt	CC = wt	M	+
CFTR gene (394 del TT)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (621+1 G>T)	11	271	ZY		GG = wt	GG = wt	M	+
	12				GG = wt	GG = wt	M	+
CFTR gene (G 542 X)	11	271	ZY		GG 542 = wt	GG 542 = wt	M	+
	12				GG 542 = wt	GG 542 = wt	M	+
CFTR gene (G 551 D)	11	9999	ZY		GG 551 = wt	GG 551 = wt	M	+
	12				GG 551 = wt	GG 551 = wt	M	+
CFTR gene (G 85 E)	11	9999	ZY		GG 85 = wt	GG 85 = wt	M	+
	12				GG 85 = wt	GG 85 = wt	M	+
CFTR gene (I 507 del)	11	9999	ZY		No del / No del = wt	No del / No del = wt	M	+
	12				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (M 1101 K)	11	9999	ZY		MM 1101 = wt	MM 1101 = wt	M	+
	12				MM 1101 = wt	MM 1101 = wt	M	+
CFTR gene (N 1303 K)	11	267	ZY		NN 1303 =wt	NN 1303 =wt	M	+
	12				NN 1303 =wt	NN 1303 =wt	M	+
CFTR gene (R 1162 X)	11	9999	ZY		RR 1162 = wt	RR 1162 = wt	M	+
	12				RR 1162 = wt	RR 1162 = wt	M	+
CFTR gene (R 117 H)	11	9999	ZY		RR 117 = wt	RR 117 = wt	M	+
	12				RR 117 = wt	RR 117 = wt	M	+

## Listing and evaluation of the results

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CG Cytogenomics

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Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CFTR gene (R 334 W)	11	9999	ZY		RR 334 = wt	RR 334 = wt	M	+
	12				RR 334 = wt	RR 334 = wt	M	+
CFTR gene (R 347 P/H)	11	9999	ZY		RR 347 = wt	RR 347 = wt	M	+
	12				RR 347 = wt	RR 347 = wt	M	+
CFTR gene (R 553 X)	11	9999	ZY		RR 553 = wt	RR 553 = wt	M	+
	12				RR 553 = wt	RR 553 = wt	M	+
CFTR gene (W 1282 X)	11	9999	ZY		WW 1282 = wt	WW 1282 = wt	M	+
	12				WW 1282 = wt	WW 1282 = wt	M	+
CFTR gene (Y 1092 X (C>A))	11	9999	ZY		YY 1092 = wt	YY 1092 = wt	M	+
	12				YY 1092 = wt	YY 1092 = wt	M	+
CFTR gene (dele2,3 (21kb))	11	9999	ZY		No Del = wt	No Del = wt	M	+
	12				No Del = wt	No Del = wt	M	+

\*\* BRAVO \*\*

## Individual summary of results

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### Molecular Genetics 28 - CFTR Common Mutations

**CFTR Gen (1677 delTA)** (N = 26, Rate of success: 100%)

Sample 11

Collective	No del / No del = wt	total
all methods	26 ●	26

Sample 12

Collective	No del / No del = wt	total
all methods	26 ●	26

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 = 26, Rate of success: 100%)

Sample 11

Collective	No del / No del = wt	total
all methods	26 ●	26

Sample 12

Collective	No del / No del = wt	total
all methods	26 ●	26

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 = 25, Rate of success: 100%)

Sample 11

Collective	No del / No del = wt	total
all methods	25 ●	25

Sample 12

Collective	No del / No del = wt	total
all methods	25 ●	25

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 = 23, Rate of success: 100%)

Sample 11

Collective	No ins / No ins = wt	total
all methods	23 ●	23

Sample 12

Collective	No ins / No ins = wt	total
all methods	23 ●	23

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

**CFTR Gen (A455E)** (N = 24, Rate of success: 100%)

Sample 11

Collective	AA 455 = wt	total
all methods	24 ●	24

Sample 12

Collective	AA 455 = wt	total
all methods	24 ●	24

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

**CFTR Gen (E 60 X)** (N = 24, Rate of success: 95,8%)

Sample 11

Collective	EE 60 = wt	E 60 X	total
all methods	1	23 ●	24

Sample 12

Collective	EE 60 = wt	total
all methods	24 ●	24

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



## CFTR Gen (Intron 8) (N = 20, Rate of success: 100%)

Sample 11

Collective	9T / 7T	total
all methods	20 ●	20

Sample 12

Collective	7T / 7T	7T / 5T	total
all methods	19 ●	1	20

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 = 31, Rate of success: 100%)

Sample 11

Collective	delta F508 Het	total
all methods	31 ●	31

Sample 12

Collective	Wild Type	total
all methods	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 (1078 del T) (N = 26, Rate of success: 100%)

Sample 11

Collective	No del / No del = wt	total
all methods	26 ●	26

Sample 12

Collective	No del / No del = wt	total
all methods	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 (1717-1 G>A) (N = 27, Rate of success: 100%)

Sample 11

Collective	GG = wt	total
all methods	27 ●	27

Sample 12

Collective	GG = wt	total
all methods	27 ●	27

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 = 27, Rate of success: 100%)

Sample 11

Collective	GG = wt	total
all methods	27 ●	27

Sample 12

Collective	GG = wt	total
all methods	27 ●	27

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 = 26, Rate of success: 100%)

Sample 11

Collective	AA = wt	total
all methods	26 ●	26

Sample 12

Collective	AA = wt	total
all methods	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 (3659 del C) (N = 26, Rate of success: 100%)

Sample 11

Collective	No del / No del = wt	total
all methods	26 ●	26

Sample 12

Collective	No del / No del = wt	total
all methods	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 (3849+10kb C>T) (N = 27, Rate of success: 100%)**
**Sample 11**

Collective	CC = wt	total
all methods	27 ●	27

**Sample 12**

Collective	CC = wt	total
all methods	27 ●	27

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 = 18, Rate of success: 100%)**
**Sample 11**

Collective	No del / No del = wt	total
all methods	18 ●	18

**Sample 12**

Collective	No del / No del = wt	total
all methods	18 ●	18

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 = 28, Rate of success: 100%)**
**Sample 11**

Collective	GG = wt	total
all methods	28 ●	28

**Sample 12**

Collective	GG = wt	total
all methods	28 ●	28

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 = 28, Rate of success: 100%)**
**Sample 11**

Collective	GG 542 = wt	total
all methods	28 ●	28

**Sample 12**

Collective	GG 542 = wt	total
all methods	28 ●	28

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 = 28, Rate of success: 100%)**
**Sample 11**

Collective	GG 551 = wt	total
all methods	28 ●	28

**Sample 12**

Collective	GG 551 = wt	total
all methods	28 ●	28

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 = 27, Rate of success: 100%)**
**Sample 11**

Collective	GG 85 = wt	total
all methods	27 ●	27

**Sample 12**

Collective	GG 85 = wt	total
all methods	27 ●	27

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 = 28, Rate of success: 100%)**
**Sample 11**

Collective	No del / No del = wt	total
all methods	28 ●	28

**Sample 12**

Collective	No del / No del = wt	total
all methods	28 ●	28

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 = 23, Rate of success: 100%)**
**Sample 11**

Collective	MM 1101 = wt	total
all methods	23 ●	23

**Sample 12**

Collective	MM 1101 = wt	total
all methods	23 ●	23

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

**CFTR gene (N 1303 K) (N = 28, Rate of success: 100%)**
**Sample 11**

Collective	NN 1303 =wt	total
all methods	28 ●	28

**Sample 12**

Collective	NN 1303 =wt	total
all methods	28 ●	28

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 = 27, Rate of success: 100%)**
**Sample 11**

Collective	RR 1162 = wt	total
all methods	27 ●	27

**Sample 12**

Collective	RR 1162 = wt	total
all methods	27 ●	27

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 = 21, Rate of success: 100%)**
**Sample 11**

Collective	RR 117 = wt	total
all methods	21 ●	21

**Sample 12**

Collective	RR 117 = wt	total
all methods	21 ●	21

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 = 26, Rate of success: 100%)**
**Sample 11**

Collective	RR 334 = wt	total
all methods	26 ●	26

**Sample 12**

Collective	RR 334 = wt	total
all methods	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 347 P/H) (N = 27, Rate of success: 100%)**
**Sample 11**

Collective	RR 347 = wt	total
all methods	27 ●	27

**Sample 12**

Collective	RR 347 = wt	total
all methods	27 ●	27

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 = 28, Rate of success: 100%)**
**Sample 11**

Collective	RR 553 = wt	total
all methods	28 ●	28

**Sample 12**

Collective	RR 553 = wt	total
all methods	28 ●	28

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 = 27, Rate of success: 100%)

Sample 11

Collective	WW 1282 = wt	total
all methods	27 ●	27

Sample 12

Collective	WW 1282 = wt	total
all methods	27 ●	27

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 = 26, Rate of success: 100%)

Sample 11

Collective	YY 1092 = wt	total
all methods	26 ●	26

Sample 12

Collective	YY 1092 = wt	total
all methods	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 (dele2,3 (21kb)) (N = 27, Rate of success: 100%)

Sample 11

Collective	No Del = wt	total
all methods	27 ●	27

Sample 12

Collective	No Del = wt	total
all methods	27 ●	27

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