

CERTIFICATE

Survey of 30 May 2025

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 gene (Allel 1) - ACMG classification

CFTR-gene nomenclature (R:B5)

CFTR gene (Allel 2) - ACMG classification

CFTR-gene - IVS8-Poly-T (R:B5)

(R) analysis is subject to the RiliBÄK

Customer:

10043733

CG Cytogenomics Medical Laboratory

CG Cytogenomics

313 Hrakleiou Av.

14122 Neo Irakleio



Düsseldorf, 26 June 2025



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



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

CERTIFICATE OF PARTICIPATION

Survey of 30 May 2025

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

Molecular Genetics 28 - CFTR Common Mutations (778):

CFTR gene (Allel 1) - ACMG classification

CFTR-gene nomenclature (R:B5)

CFTR gene (Allel 2) - ACMG classification

CFTR-gene - IVS8-Poly-T (R:B5)

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

Listing and evaluation of the results

10043733: CG Cytogenomics Medical Laboratory
CG Cytogenomics

Survey of 30 May 2025

Adviser:

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778

Molecular Genetics 28 - CFTR Common Mutations

Analyte	Sam- ple	Method	Manu- facturer	Device	Your specification(s)	Correct specification(s)	TV- Type	Meets criteria
CFTR gene (Allel 1) - c. -nomenclature	(2) 31	9999	ZY		c.1624G>T	c.1624G>T	M	+
	(2) 32				c.254G>A	c.254G>A	M	+
CFTR gene (Allel 1) - p. -nomenclature	(2) 31	9999	ZY		p.(Gly542*)	p.(Gly542*)	M	+
	(2) 32				p.(Gly85Glu)	p.(Gly85Glu)	M	+
CFTR gene (Allel 1) - ACMG classification	31	9999	ZY		pathogenic	pathogenic	M	+
	32				pathogenic	pathogenic	M	+
CFTR gene (Allel 1) - IVS8-Poly-T	(1) 31	9999	ZY		9T	9T, 9T + 10 TG	M	+
	(1) 32				7T	7T, 7T + 11 TG	M	+
CFTR gene (Allel 2) - c. -nomenclature	(2) 31	9999	ZY		c.1624G>T	c.1624G>T	M	+
	(2,4) 32					Wild Type	M	+
CFTR gene (Allel 2) - p. -nomenclature	(2) 31	9999	ZY		p.(Gly542*)	p.(Gly542*)	M	+
	(2,4) 32					Wild Type	M	+
CFTR gene (Allel 2) - ACMG classification	31	9999	ZY		pathogenic	pathogenic	M	+
	(3) 32				pathogenic	not applicable	M	+
CFTR gene (Allel 2) - IVS8-Poly-T	(1) 31	9999	ZY		9T	9T, 9T + 10 TG	M	+
	(1) 32				7T	7T, 7T + 11 TG	M	+

** BRAVO **

- (1) To obtain the certificate "CFTR-gene - IVS8-Poly-T (R:B5)", at least 2 of the possible 2 examinations must be correctly determined, your number is 2.
 (2) To obtain the certificate "CFTR-gene nomenclature (R:B5)", at least 4 of the possible 4 examinations must be correctly determined, your number is 4.
 (3) The result for this sample was considered as correct due to additional information in the comment field.
 (4) The result was evaluated as correct.

Individual summary of results

10043733: CG Cytogenomics Medical Laboratory
CG Cytogenomics

Survey of 30 May 2025

778

Molecular Genetics 28 - CFTR Common Mutations

CFTR gene (Allel 1) - ACMG classification (N = 58, Rate of success: 94,8%)

Sample 31

Collective	pathogenic	not applicable	total
allele combination 1	1 ●	0	1
allele combination 2	55 ●	2	57

Sample 32

Collective	pathogenic	not applicable	total
allele combination 1	0	1 ●	1
allele combination 2	54 ●	3	57

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

CFTR gene (Allel 2) - ACMG classification (N = 58, Rate of success: 81%)

Sample 31

Collective	pathogenic	not applicable	total
allele combination 1	1 ●	0	1
allele combination 2	54 ●	3	57

Sample 32

Collective	pathogenic	likely pathogenic	significance uncertain (VUS)	not applicable	total
allele combination 1	1 ●	0	0	0	1
allele combination 2	7	1	1	38 ●	47

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

CFTR gene (Allel 1) - c. -nomenclature (N = 61, Rate of success: 91,8%)

Sample 31

Collective	c.1624G>T	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	56 ●	4	60

Sample 32

Collective	c.254G>A	Wild Type	total
allele combination 1	0	1 ●	1
allele combination 2	55 ●	5	60

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

CFTR gene (Allel 1) - p. -nomenclature (N = 61, Rate of success: 91,8%)

Sample 31

Collective	p.(Gly542*)	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	56 ●	4	60

Sample 32

Collective	p.(Gly85Glu)	Wild Type	total
allele combination 1	0	1 ●	1
allele combination 2	55 ●	5	60

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

CFTR gene (Allel 2) - c. -nomenclature (N = 61, Rate of success: 91,8%)

Sample 31

Collective	c.1624G>T	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	55 ●	5	60

Sample 32

Collective	c.254G>A	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	0	48 ●	48

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

CFTR gene (Allel 2) - p. -nomenclature (N = 61, Rate of success: 90,2%)

Sample 31

Collective	p.(Gly542*)	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	55 ●	5	60

Sample 32

Collective	p.(Gly85Glu)	Wild Type	total
allele combination 1	1 ●	0	1
allele combination 2	0	47 ●	47

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

CFTR gene (Allel 1) - IVS8-Poly-T (N = 53, Rate of success: 98,1%)

Sample 31

Collective	7T	9T	9T, 10 TG	total
allele combination 1	1	36 ●	16 ●	53

Sample 32

Collective	7T	7T, 10 TG	7T, 11 TG	total
allele combination 1	37 ●	1	15 ●	53

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

CFTR gene (Allel 2) - IVS8-Poly-T (N = 53, Rate of success: 98,1%)

Sample 31

Collective	9T	7T, 10 TG	9T, 10 TG	total
allele combination 1	37 ●	1	15 ●	53

Sample 32

Collective	7T	7T, 10 TG	7T, 11 TG	total
allele combination 1	37 ●	1	15 ●	53

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