Newborn Screening Quality Assurance Program
Cystic Fibrosis DNA Variant Detection
Proficiency Testing Program (CFDNAPT)

In co-sponsorship with Association of Public Health Laboratories (APHL)
Provided by the Newborn Screening and Molecular Biology Branch
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REPORT AUTHORIZATION

This report has been reviewed and authorized by Dr. Christopher Greene, Acting Laboratory Chief, Molecular Quality Improvement Program, and Dr. Stanimila Nikolova, Laboratory Chief of the Proficiency Testing and Reference Materials Lab.

CONFIDENTIALITY STATEMENT

NSQAP participant information and evaluations are strictly confidential and only available by accessing the NSQAP Participant Portal.

Introduction

This report summarizes all results submitted within the data-reporting period for the Quarter 1, 2026 proficiency testing program for cystic fibrosis (CF) variant detection (CFDNAPT) from the Newborn Screening Quality Assurance Program (NSQAP). The contents provide certification profiles for the distributed specimens, the primary and secondary screening methods and the DNA extraction methods used by participants, the summary of reported genotypes, and the overall summary of reported clinical assessments. An evaluation of submitted individual laboratory results is attached.

Certification of PT Specimens

The Quarter 1 panel consisted of five dried blood spot (DBS) specimens (20261011001, 20261011002, 20261011003, 20261011004, 20261011005) prepared from CF patients, carriers, or unaffected individuals. All variants are characterized at CDC using Sanger sequencing and then confirmed in DBS specimens using next generation sequencing technologies and/or a multiplex genotyping assay. DNA was extracted from DBS specimens with Qiagen Generation DNA Purification and DNA Elution Solutions and was assayed using Luminex Molecular Diagnostics xTAG CF 60 v2 to verify performance. Table 1 provides specimen certification information.

Specimen Consensus

If more than 10 U.S. laboratories report results, we require at least 80% of those laboratory results to agree with the CDC result to evaluate a specimen. If fewer than 80% of the reported results agree with the CDC result, the specimen will be marked as “Not Evaluated”.

If fewer than 10 U.S. laboratories report results, we do not apply the 80% consensus threshold. In these cases, all submitted results are evaluated, and the specimen will be evaluated regardless of the level of agreement with the CDC resultNSQAP occasionally challenges cutoff levels for certain analytes by enriching samples near those levels. The NSQAP PT Committee closely reviews all specimen data. Specimens that are not evaluated are considered educational.

All specimens included in the 2026 Q1 CFDNA PT event met the 80% consensus.

Table 1. Specimen Certification

Specimen	Allele 1	Allele 2	Genotype	Clinical Assessment
20261011001	G542X (p.Gly542X)	I336K (p.Ile336Lys)	G542X (p.Gly542X)/ I336K (p.Ile336Lys)	Screen Positive- 1 or 2 variants
20261011002	F508del (p.Phe508del)	3849+10kbC>T (c.3718-2477C>T)	F508del (p.Phe508del)/ 3849+10kbC>T (c.3718-2477C>T)	Screen Positive- 1 or 2 variants
20261011003	W1282X (p.Trp1282X)	N1303K (p.Asn1303Lys)	W1282X (p.Trp1282X)/ N1303K (p.Asn1303Lys)	Screen Positive- 1 or 2 variants
20261011004	F508del (p.Phe508del)	+	F508del (p.Phe508del)/ +	Screen Positive- 1 or 2 variants
20261011005	F508del (p.Phe508del)	Q493X (p.Gln493X)	F508del (p.Phe508del)/ Q493X (p.Gln493X)	Screen Positive- 1 or 2 variants

The + in the genotype indicates that no pathogenic variants were detected during specimen certification for one or both chromosomes.

Distribution of PT Specimens

On January 28, 2026, NSQAP distributed a panel of five unknown DBS specimens to 33 laboratories in the United States and 48 laboratories in other countries to detect pathogenic variants in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene.

Participant Results

Participants reported their data through the NSQAP Participant Portal. Data was received from 72 of the 81 participants by the data reporting deadline. Participants tested specimens by the analytical schemes they routinely use for newborn screening. Reported data included *CFTR* analysis method(s), DNA extraction method, variant panel(s), screening algorithms, alleles found for each specimen, and clinical assessments. If a *CFTR* analysis method was not commercially available, the participant provided the variant panel or regions sequenced for evaluation.

Reported Method Data

Tables 2 – 4 provide the primary and secondary *CFTR* analysis methods and the DNA extraction methods reported by participants.

Table 2. Reported Primary Methods

Primary Method	# of Labs
Luminex Molecular Diagnostics CFTR IVD 39 v2	9
Luminex Molecular Diagnostics xTAG CF 60 v2	9
Next Gen Sequencing - Illumina MiSeqDx CF 139 Variant Assay	7
All other gene sequencing protocols including Sanger and Next Generation	6
Elucigene Diagnostics CF-EU2v1	5
ViennaLab Diagnostics GmbH CF StripAssay, GER	5
Devyser CFTR NGS	4
Real-time PCR Allelic Discrimination Assay (ie TaqMan)	3
Agena Bioscience iPLEX Pro CFTR panel (72 mutations)	2
Sequenom assays other than HerediT CF (MALDI-TOF Mass Spectrometry)	2
Allele-specific Oligonucleotide PCR	1
Elucigene Diagnostics CF4v2	1
In-house Amplification Refractory Mutation System	1
In-house Hydrolysis Probe Assay	1
Luminex Molecular Diagnostics xTAG CF 71 v2	1
Next Gen Sequencing – Multiplicom Molecular Diagnostics CFTR MASTR v2	1
ViennaLab Diagnostics GmbH CF StripAssay (4-410)	1
Other	13

Table 3. Reported Secondary Methods

Secondary Method	# of Labs
All other gene sequencing protocols including Sanger and Next Generation	7
Luminex Molecular Diagnostics CFTR IVD 39 v2	3
PCR/Heteroduplex Analysis/Gel Electrophoresis	3
Agena Bioscience iPLEX Pro CFTR panel (72 mutations)	1
Luminex Molecular Diagnostics xTAG CF 60 v2	1
Next Gen Sequencing - Illumina MiSeqDx CF 139 Variant Assay	1
Next Gen Sequencing - Illumina MiSeqDx CF Clinical Sequencing Assay	1
Other	7

Table 4. Reported DNA Extraction Methods

Extraction Method	# of Labs
Qiagen Generation DNA Purification & DNA Elution Solutions (also sold as 5 Prime Easy PCR Solutions 1 & 2)	16
Qiagen QIAamp spin columns (manual or robotic)	7
In-house alkaline lysis prep	5
Qiagen magnetic bead kit (EZ1 or BioSprint 96)	5
ViennaLab GenXtract	4
Sigma Aldrich Extract-N-Amp	3
In-house boiling prep	2
In-house lysis boiling prep	2
Revvity/Chemagen Chemagic kit	1
Other	27

Allele Assessment Data

Tables 5a – 5e show the genotypes identified by the participants and the genotype errors for each specimen.

Table 5a. **Specimen 20261011001**

Genotype Identified	Number of Labs	Number of Genotype Errors
G542X (p.Gly542X)/ I336K (p.Ile336Lys)	30	0
G542X (p.Gly542X)/ +	36	0
I336K (p.Ile336Lys)/ +	1	1
+ / +	4	1
G542X (p.Gly542X)/ Other [I336K(c.1007T>A)]	1	0

Table 5b. **Specimen 20261011002**

Genotype Identified	Number of Labs	Number of Genotype Errors
F508del (p.Phe508del)/ 3849+10kbC>T (c.3718-2477C>T)	63	0
F508del (p.Phe508del)/ +	6	1
+ / +	1	1
F508del (p.Phe508del)/ Other [3849+10kb C>T (c3717 + 12191C>T*)]	1	0
F508del (p.Phe508del)/ Other [3717+12191C>T* and R668C]	1	0

* Variant 3849+10kbC>T can be referred to as c.3718-2477C>T or c.3717+12191C>T

Table 5c. **Specimen 20261011003**

Genotype Identified	Number of Labs	Number of Genotype Errors
W1282X (p.Trp1282X)/ N1303K (p.Asn1303Lys)	64	0
+ / +	8	2

Table 5d. **Specimen 20261011004**

Genotype Identified	Number of Labs	Number of Genotype Errors
F508del (p.Phe508del)/ +	70	0
+ / +	1	1
F508del (p.Phe508del)/ Other [+]	1	0

Table 5e. **Specimen 20261011005**

Genotype Identified	Number of Labs	Number of Genotype Errors
F508del (p.Phe508del)/ Q493X (p.Gln493X)	39	0
F508del (p.Phe508del)/ +	32	0
+ / +	1	1

Clinical Assessment Data

Table 6 provides a summary of participants' clinical assessments for each specimen.

Table 6. Clinical Assessments Reported for each Specimen

Clinical Assessment	20261011001	20261011002	20261011003	20261011004	20261011005
Screen Negative- Normal	4	1	8	1	1
Screen Positive – 1 or 2 variants	68	71	64	71	71
Incorrect Clinical Assessment	1	1	2	1	1

Evaluations

Evaluations are based on the correct allele identification and clinical assessment for each specimen. Participants are evaluated according to their screening method(s), variant panel, and screening algorithm. A specimen is classified as "Unacceptable" if any of the alleles and/or clinical assessment reported is incorrect according to the laboratory's panel and algorithm. Submissions are not evaluated when a participant is using a custom assay, and the variant panel or regions sequenced are not provided. In Quarter 1 2026, there were eight unacceptable allele assessments, six unacceptable clinical assessments.

Future Shipments

The Newborn Screening Quality Assurance Program will ship Quarter 3 PT specimens for CFDNAPT on July 22, 2026.

Direct Inquiries

If you have any comments or questions about CFDNAPT technical analysis, contact Miyono Hendrix by e-mail at zuj7@cdc.gov. For data reporting questions, contact NSQAP at nsqapdmt@cdc.gov.

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