Centers for Disease Control and Prevention (CDC)

National Center for Environmental Health (NCEH)

Division of Laboratory Sciences (DLS)

NEWBORN SCREENING AND MOLECULAR BIOLOGY BRANCH (NSMBB)

NEWBORN SCREENING QUALITY Assurance Program (NSQAP) Portal

CFDNAPT USER GUIDE

September 2023

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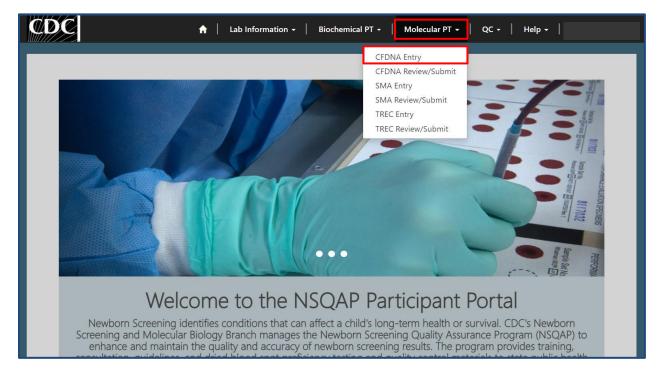
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1. CFDNAPT Program Entry Page

1.1 Navigation

To enter and save CFDNAPT data, navigate to the CFDNAPT program entry page. Access the page from the 'CFDNA Entry' option on the Molecular PT drop-down menu.

1. Click ' Molecular PT' then 'CFDNA Entry' from the drop-down menu.



2. Select 'CFDNA' to navigate to the entry page.

CDC	↑ Lab Information - Biochemical PT - Molecular PT	• QC • Help •
Home > CFDNAPT Entry		
CFDNAPT Entry		
Name 🕇		Created On
CFDNA		2/17/2021 1:20 PM
About NSQAP Self-Service Portal		
This program is cosponsored by the Centers for (CDC) and the Association of Public Health La		

3. You will be directed to the CFDNA entry page to enter method information and data.

1.2 Primary Method Information

Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter method information including primary method, secondary/confirmatory method, and extraction method. Navigation details can be found in section 1.1.

CDC	
Home > Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)	
Cystic Fibrosis DNA Variant Detection Proficienc Testing (CFDNAPT)	У
* Definition of commercial kit – A kit that has been designed by the manufacturer to sequence the CFTR g	ene
Method Information	
Primary Method	
Select a primary method *	
	Q
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * No	~
Was a gene sequencing method used? *	
No	~
Secondary/Confirmatory Method Select a secondary/confirmatory method	٩
Extraction Method	
Select an extraction method *	Q

1. Click on the magnifying glass to see the primary methods list.



2. Search for methods using the search box or page numbers.

		Search	C
~	Name 1		
~	Abbott Molecular CF Genotyping Assay v3		
	Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)		
	All other gene sequencing protocols including Sanger and Next Gen		
	Allele-specific Oligonucleotide PCR		
	Amplification and Polyacrylamide Gel Electrophoresis (PCR-PAGE)		
	Amplification and Restriction Fragment Length Polymorphism Analysis (PCR-RFLP)		
	Astra Biotech CFcheck DE-31		
	Capillary Electrophoresis		
<	1 2 3 4 >		

3. Choose a primary method then click 'Select'.

Abbott Molecular CF Genotyping Assay v3

		Search	Q
		Jouren	
	Name 🕇		
V -	Abbott Molecular CF Genotyping Assay v3		
	Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)		
	All other gene sequencing protocols including Sanger and Next Gen		
1	Allele-specific Oligonucleotide PCR		
et	Amplification and Polyacrylamide Gel Electrophoresis (PCR-PAGE)		
	Amplification and Restriction Fragment Length Polymorphism Analysis (PC	CR-RFLP)	
m	Astra Biotech CFcheck DE-31		
	Capillary Electrophoresis		-
ct		Select Cancel	Remove value
ct		Select	Remove value
		Select	Remove value
Cystic Fibrosis	s DNA Variant Detection Proficiency Testing (CFDNAPT)	Select	Remove value
stic Fik	prosis DNA Variant Detection		
	prosis DNA Variant Detection		
stic Fib	prosis DNA Variant Detectio PT)	on Proficiency	Testing
stic Fib	prosis DNA Variant Detection	on Proficiency	Testing
stic Fik DNAF	prosis DNA Variant Detectio PT)	on Proficiency	Testing

4. If 'Other' is selected, a text box will appear. You are **required** to specify your primary method details.

	o records		
			other
1	Name 🕇		
1	Other		
			Select Cancel Remove valu
1			Select Cancel Remove valu
-1			
stic F	ibrosis DNA Variant	Detection F	
stic F	ibrosis DNA Variant APT)	Detection F	

Definition of commercial kit	- A kit that has been designed by the manufacturer to sequence the CFTR gene	
Method Inform	mation	
rimary Method		
Select a primary metho	d *	
Other		

5. Indicate whether your selected primary method is a custom commercial assay, laboratory developed test, or a commercial assay with restrictions set by your lab by clicking the drop-down arrow.

Cystic Fibrosis DNA Variant Detection Proficiency Testin (CFDNAPT)	g	
* Definition of commercial kit – A kit that has been designed by the manufacturer to sequence the CFTR gene Method Information Primary Method		
Select a primary method * Abbott Molecular CF Genotyping Assay v3	×	Q
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *		~

6. If no restrictions or laboratory specific customizations were made, no additional information is required.

Cystic Fibrosis DNA Variant Detection Proficiency Testir (CFDNAPT)	ŋ		
[#] Definition of commercial kit – A kit that has been designed by the manufacturer to sequence the CFTR gene Method Information			
Primary Method Select a primary method * Abbott Molecular CF Genotyping Assay v3	×	Q	
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * No		,	y
Was a gene sequencing method used? * No		``	~

7. If restrictions were placed or laboratory specific customizations were made, you are required to specify the variants detected. See section 1.5 for additional information on a helpful tool for correctly formatting this information.

<u>NOTE</u>: List the variants detected by your lab, separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may instead reference your website.

Method Information	
Primary Method	
Select a primary method *	
Abbott Molecular CF Genotyping Assay v3	x (
Is your selected method a custom commercial assay OR a lab developed test	OR does your lab place restrictions on a commercial assay? *
Specify variants detected: *	
List the variants detected by your lab-separate each variant by a semicolon $\overset{\rm war}{c}$ if the list your website.	ie variants that your lab detect are on your program's website, you may instead
Was a gene sequencing method used? *	
No	

8. Indicate whether a gene sequencing method was used by clicking the drop-down arrow.

Abbott Molecular CF Genotyping Assay v3	×
s your selected method a custom commercial assay OR a lab developed test OR does your lab place restricti	ns on a commercial assay? *
Yes	
Specify variants detected: *	
M1V p.Met1Val / p.Met1? (c.1A>G),Q2X p.Gl02* (c.4C>T),S4X p.Ser4* (c.11C>A),S13F p.Ser13Phe (c.38C>T),71 p.Leu15Pro (c.44T>C),185+1G->T p.? (c.53+1G-T),W19X p.Trp19* (c.57G>A),C27R p.Gly27Arg (c.79G>A),C27K (c.88C>T),Q39X p.Gln39* (c.115C>T),X46D p.Ala4GAsp (c.137C>A),296+1G->A p.? (c.164+1G>A),296+1G->T (c.164+2T>C),296+3insT p.? (c.165+4dupT),297-3C->T p.? (c.165-3C>T),297-1G->A p.? (c.165+1G>A),E56K p.	p.Gly27* (c.79G>T);Q30X p.Gln30* ? (c.164+1G>T);296+2T->C p.?
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect ar ist your website. Was a gene sequencing method used? *	on your program's website, you may inste
No	

9. If a gene sequencing method was not used, no additional information is required.

Yes	
pecify variants detected: *	
p.Leu15Pro (c.44T>C);185+1G->T p.? (c. (c.88C>T);Q39X p.Gln39* (c.115C>T);A44	p.Gln2* (c.4C>T);54X p.Ser4* (c.11C>A);513F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);115P 53+1G>T);W19X p.Trp19* (c.57G>A);627R p.Gly27arg (c.79G>A);627X p.Gly27* (c.79G>T);630X p.Gln20* D p.Ala4GAsp (c.137C>A);296+1G>A p.? (c.164+1G>A);296+1G>- T p.? (c.164+1G>T);296+2T->C p.? pT);297-3G->T p.? (c.165-3G <t);297-1g->A p.? (c.165-1G>A);556K p.Glu56iys (c.166G>A);W57G p.Trp57Giy</t);297-1g->
ist the variants detected by your lab-separ st your website.	rate each variant by a semicolon ",". If the variants that your lab detect are on your program's website, you may ins
Vas a gene sequencing method used? *	
No	

10. If a gene sequencing method was used, indicate whether a commercial kit was used.

Note: A commercial kit is defined as a kit that has been designed by the manufacturer to sequence the CFTR gene.

Yes	0
Specify variants detected: *	
M1V p.Met1Val / p.Met1? (c.1A>G)(22X p.Gln2* (c.4C>T);54X p.Ser4* (c.11C>A);513F p.Ser13Phc (c.38C>T);1824dlT p.Phe17Serfs*8 (c.50delT);1 p.Leu15Pro (c.41T>C);185+1G>T p.? (c.533+1G>T);W19X p.Tp19* (c.57G>A);G27R p.G)(27Arg (c.79G>A);G27X p.G)(927* (c.79G>T);230X p.Gln3* (c.88C>T);Q39X p.Gln39* (c.115C>T);A46D p.Ala468p (c.137C>A);296+1G>A p.? (c.164+1G>A);296+1G>A);296+1G>A (c.64A+T>C);296+31nsT p.? (c.164+4GUT);297-3C->T p.? (c.165+3G>A);297+1G>A p.? (c.165+1G>A);256K p.Glu36% (c.166G>A);W57G p.Tp:	30*
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you list your website.	may instead
Was a gene sequencing method used? *	
Yes	
* Was a commercial kit used? *	

11. If a commercial kit was used, no further information is required.

Yes	
Specify variants detected: *	
M1V p.Met1Val / p.Met1? (c.1A>G);Q2X p.Gln2* (c.4C>T);S4X p.Ser4* (c.11C>A);S13F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);L15P p.Leu15Pro (c.44T>C);185+1G->T p.? (c.53+1G>T);W19X p.Trp19* (c.57G>A);G27R p.Gly27Arg (c.79G>A);G27X p.Gly27* (c.79G>T);Q30X p.Gln30* (c.88C>T);Q39X p.Gln39* (c.115C>T);A46D p.Ala46Asp (c.137C>A);296+1G->A p.? (c.164+1G>A);296+1G->T p.? (c.164+1G>T);296+2T->C p.? (c.164+2T>C);296+3insT p.? (c.164+4dupT);297-3C->T p.? (c.165-3C>T);297-1G->A p.? (c.165-1G>A);E56K p.Glu56Lys (c.166G>A);W57G p.Trp57Gly	
ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may ins ist your website.	tead
Nas a gene sequencing method used? *	
Yes	
Was a commercial kit used? *	

12. If a commercial kit was not used, additional gene sequencing regions information is required.

Specify variants detected: *	
M1V p.Met1Val / p.Met1? (c.1A>G);Q2X p.Gln2* (c.4C>T);S4X p.Ser4* (c.11C>A);S13F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);L15P p.Leu15Pro (c.44T>C);185+1G->T p.? (c.53+1G>T);W19X p.Trp19* (c.57G>A);G27R p.Gly27Arg (c.79G>A);G27X p.Gly27* (c.79G>T);Q30X p.Gln30* (c.88C>T);Q39X p.Gln39* (c.115C>T);A46D p.Ala46Asp (c.137C>A);296+1G->A p.? (c.164+1G>A);296+1G->T p.? (c.164+1G>T);296+2T->C p.? (c.164+2T>C);296+3insT p.? (c.164+4dupT);297-3C->T p.? (c.165-3C>T);297-1G->A p.? (c.165-1G>A);E56K p.Glu56Lys (c.166G>A);W57G p.Trp57Gly	•
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may inste list your website.	ead
Was a gene sequencing method used? *	
Yes	~
# Was a commercial kit used? *	
No	~
For non-commercial kits, provide regions of the gene that are sequenced *	
romon-commercial kits, provide regions of the gene that are sequenced	
Specify variants detected: *	_
M1V p.Met1Val / p.Met1? (c.1A>G);Q2X p.Gln2* (c.4C>T);S4X p.Ser4* (c.11C>A);S13F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);115P	^
p.Leu15Pro (c.44T>C);185+1G->T p.? (c.53+1G>T);W19X p.Trp19* (c.57G>A);G27R p.Gly27Arg (c.79G>A);G27X p.Gly27* (c.79G>T);Q30X p.Gln30* (c.88C>T);Q39X p.Gln39* (c.115C>T);A46D p.Ala46Asp (c.137C>A);296+1G->A p.? (c.164+1G>A);296+1G->T p.? (c.164+1G>T);296+2T->C p.? (c.164+2T>C);296+3insT p.? (c.164+4dupT);297-3C->T p.? (c.165-3C>T);297-1G->A p.? (c.165-1G>A);E56K p.Glu56Lys (c.166G>A);W57G p.Trp57Gly	
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may insta list your website.	ead
Was a gene sequencing method used? *	
Yes	~
* Was a commercial kit used? *	
No	~
For non-commercial kits, provide regions of the gene that are sequenced *	
exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	

13. After entering primary method information, continue to the secondary/confirmatory method section (if necessary – section 1.3) or the extraction method section (section 1.4).

1.3 Secondary/Confirmatory Method Information

Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter method information including primary method, secondary/confirmatory method, and extraction method.

Reporting of secondary/confirmatory method information is only required by participants if a secondary/confirmatory method is utilized. If this method is not utilized, proceed to section 1.4 for guidance on reporting extraction method information. If a secondary/confirmatory method is utilized, continue to step 1 for reporting guidance.

1. Select a secondary/confirmatory method by clicking on the magnifying glass.

Was a gene sequencing method used? *	
Yes	~
* Was a commercial kit used? *	
No	~
For non-commercial kits, provide regions of the gene that are sequenced *	
exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	
Secondary/Confirmatory Method Select a secondary/confirmatory method	
	٩
extraction Method	
Select an extraction method *	
	Q

2. Choose a method then click 'Select'.

гоокир	records		>
		Search	٩
~	Name 🕇		
	Abbott Molecular CF Genotyping Assay v3		
1	Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)		
	All other gene sequencing protocols including Sanger and Next Gen		
	Allele-specific Oligonucleotide PCR		
	Amplification and Polyacrylamide Gel Electrophoresis (PCR-PAGE)		_
	Amplification and Restriction Fragment Length Polymorphism Analysis (PCR-RFLP)		
	Astra Biotech CFcheck DE-31		
	Capillary Electrophoresis		-
<	2 3 4 >		
		Select Cancel Remo	ove value

For non-commercial kits, provide regions of the gene that are sequenced * exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	
econdary/Confirmatory Method	
Select a secondary/confirmatory method	
Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	x C
Select an algorithm for utilization of the secondary/confirmatory method *	

3. Select an algorithm for utilization of the secondary/confirmatory method by clicking on the magnifying glass.

For non-commercial kits, provide regions of the gene that are sequenced * exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	
secondary/Confirmatory Method Select a secondary/confirmatory method	
Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	x Q
Select an algorithm for utilization of the secondary/confirmatory method *	
Select an algorithm for utilization of the secondary/confirmatory method *	٩

4. Choose an algorithm then click **'Select'**.

Look	up records		×
		Search	Q
~	Utilization of Secondary Confirmatory 🕈		
~	Both the primary and secondary methods are used to detect variants		
	Other		
	Secondary method run only when primary method is positive and may find additional variants		
	Secondary method run only when primary method is positive and only for confirmation (NO new	variants identified)	
		Select Cancel Rem	ove value

Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	
	^
Select an algorithm for utilization of the secondary/confirmatory method *	
Both the primary and secondary methods are used to detect variants	×
is your selected method a custom commercial assay OR a lab developed test OR does your lab place restriction	

5. Click the drop-down arrow to indicate whether your selected secondary/confirmatory method is a custom commercial assay, laboratory developed test, or a commercial assay with restrictions set by your lab.

condary/Confirmatory Method Select a secondary/confirmatory method		
Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	×	C
Select an algorithm for utilization of the secondary/confirmatory method *		
Select an algorithm for utilization of the secondary/confirmatory method * Both the primary and secondary methods are used to detect variants	×	C
	×	0
	×	C

6. If no restrictions or laboratory specific customizations were made, no additional information is required.

Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	×	(
select an algorithm for utilization of the secondary/confirmatory method *		
Both the primary and secondary methods are used to detect variants	×	0
s your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *		
s your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *		

7. If restrictions were placed or laboratory specific customizations were made, you are required to specify the variants detected. See section 1.5 for additional information on a helpful tool for correctly formatting this information.

<u>NOTE</u>: List the variants detected by your lab, separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may instead reference your website.

Agena Bioscience iPLEX Pro CFTR Panel (72 mutations) Select an algorithm for utilization of the secondary/confirmatory method * Both the primary and secondary methods are used to detect variants Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes Specify Variants Detected * List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:	lary/confirmatory method		
Both the primary and secondary methods are used to detect variants Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes Specify Variants Detected *	nce iPLEX Pro CFTR Panel (72 mutations)	3	
Both the primary and secondary methods are used to detect variants Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes Specify Variants Detected *			
Both the primary and secondary methods are used to detect variants is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes Specify Variants Detected *			
s your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes Specify Variants Detected *	ithm for utilization of the secondary/confirmatory method *		
Yes Specify Variants Detected *	ary and secondary methods are used to detect variants	3	
Yes			
Yes pecify Variants Detected *			
Yes			
pecify Variants Detected *	I method a custom commercial assav OR a lab developed test OR does vour lab place res	trictions on a commercial assau? *	
	l method a custom commercial assay OR a lab developed test OR does your lab place res	trictions on a commercial assay? *	
	l method a custom commercial assay OR a lab developed test OR does your lab place res	trictions on a commercial assay? *	
	l method a custom commercial assay OR a lab developed test OR does your lab place res	trictions on a commercial assay? *	
ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:		trictions on a commercial assay? *	
ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:		trictions on a commercial assay? *	
ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:		trictions on a commercial assay? *	
ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:		trictions on a commercial assay? *	
.ist the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may in:		trictions on a commercial assay? *	
		trictions on a commercial assay? *	

8. Indicate whether a gene sequencing method was used by clicking the drop-down arrow.

Yes	в
Specify Variants Detected *	
CCT/4_T/r/deTrAga); 306InSAT[p.Arg/Saturdary]; 2000 profile [p.Gluboury] (CT/36G>A); Ebux [p.Glu	
p.Arg75* (c.223C>T);365-366insT p.Trp79Leufs*32(c.233dupT);685E p.Gly85Glu (c.254G>A);394deITT p.Leu88llefs*22;(c.262_263deITT);L88X p.Leu88* (c.263T>A);L88X p.Leu88* (c.263T>A);L88X p.Leu88* (c.263T>A);C1G=263deITT);L88X p.Cly85Glu (c.271G>A);405+1G->A p.? (c.273+1G>A);405+3A->C p.? (c.273+3A>C);406-2A->G p.? (c.273+3A>C);406-2A->G p.? (c.273+3A>C);406-2A->G p.? (c.273+3A>C);405+3A->C p.? (c.27	
(220517A),L00A p.2eu08 (C.20517A),G91K p.Glu991KI (C.271G7A),405+1G7A), 405+1G7A),405+5A72C p.3 (C.273+3A72C),406+2A73C p.3 (C.274G7A),405+3A72C p	.74-
(c.293A>G);P99L p.Pro99Leu (c.296C>T);L102R p.Leu 102Arg (c.305T>G);442delA p.Arg104Glufs*3 (c.310delA)	
list your website.	
Was a gene sequencing method used? *	
Was a gene sequencing method used? *	

9. If a gene sequencing method was not used, no additional information is required.

Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a com Yes	~
Specify Variants Detected * (C.174_177de11AGA);30binsA p.Arg59Lysts*10 (C.175dupA);E60K p.GluboLys (C.178G>A);E60X p.Glub0* (C.178G>1);P67L p.Prd p.Arg75* (c.223C>T);365-366insT p.Trp79Leufs*32(c.233dupT);G85E p.Gly85Glu (c.254G>A);394delTT p.Leu88lefs*22;(c.262_26 (c.263T>A);L88X p.Leu88* (c.263T>G);G91R p.Gly91Arg (c.271G>A);405+1G->A p.? (c.273+1G>A);405+3A->C p.? (c.273+3A 2A>G);406-1G->A p.? (c.274-1G>A);292K p.Glu92Lys (c.274G>A);E92X p.Glu92* (c.274G>T);Q98X p.Gln98* (c.292C>T);Q98R (c.293A>G);299L p.Pro99Leu (c.296C>T);102R p.Leu102Arg (c.305T>G);422del p.Arg104Glufs*3 (c.310delA) List the variants detected by your lab detect are on your proc	53delTT);L88X p.Leu88* A>C);406-2A->G p.? (c.274- t p.Gln98Arg
list the variants detected by your lab-separate each variant by a semicolor , . If the variants that your lab detect are on your pro-	fram's website, you may instead
No	~
xtraction Method	
Select an extraction method *	
	Q

10. If a gene sequencing method was used, indicate whether a commercial kit was used.

Note: A commercial kit is defined as a kit that has been designed by the manufacturer to sequence the CFTR gene.

Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *	
Yes	~
Specify Variants Detected *	
(c.174_177ae11AGA);306insA p.Arg59Lysts*10 (c.175aupA);E60K p.GluboLys (c.178G>A);E60X p.Glub0* (c.178G>1);P67L p.Pro67Le0 (c.200C>1);K75X p.Arg75* (c.223C>T);365-366insT p.Trp79Le0fs*32(c.233dupT);G85E p.Gly85Glu (c.254G>A);394delTT p.Le088ilefs*22;(c.262_263delTT);L88X p.Le088*	*
(c.263T>A);L88X p.Leu88* (c.263T>G);G91R p.Gly91Arg (c.271G>A);405+1G->A p.? (c.273+1G>A);405+3A->C p.? (c.273+3A>C);406-2A->G p.? (c.274-	
2A>G);406-1G->A p.? (c.274-1G>A);E92K p.Glu92Lys (c.274G>A);E92X p.Glu92* (c.274G>T);Q98X p.Gln98* (c.292C>T);Q98R p.Gln98Arg	-
(c.293A>G);P99L p.Pro99Leu (c.296C>T);L102R p.Leu102Arg (c.305T>G);442delA p.Arg104Glufs*3 (c.310delA)	
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may insi	tead
list your website.	
Was a gene sequencing method used? *	
Yes	~
* Was a commercial kit used? *	
	~

11. If a commercial kit was used, no further information is required.

Yes	~
Specify Variants Detected *	
[C174] 17/de1AGAJ;30binsA [p.Arg59Lysts" 10 (C17/adipA);Eb0K] p.Glubouys (C17/8G-3);Eb0K] p.Glubou* (C17/8G-3);Pb7L [p.Prob/Le0 (C20UC>1);X/5) p.Arg75* (c223C>T);365-366insT]p.Trp79Leufs*32(c233dupT);G85E p.Gly85Glu (c254G>A);394deITT p.Leu88llefs*22;(c262_263deITT);L88X p.Leu88 (c263T>A);L88X p.Leu88* (c263T>G);G91R p.Gly91Arq (c271G>A);405+1G>A p.? (c273+1G>A)=A>C p.? (c273+3A>C)	3*
(2A>G);406-1G->A p.? (c.274-1G>A);592 p.Glu92tys (c.274G-A);592 p.Glu92* (c.274G-A);598 p.Glu98* (c.293A>G);2998 p.Glu98* (c.293A>G);2991 p.Pro99Leu (c.296C>T);L102R p.Leu102Arg (c.305T>G);442delA p.Arg104Glufs*3 (c.310delA)	
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you ma list your website.	ay instead
Was a gene sequencing method used? *	
Yes	•
# Was a commercial kit used? *	

12. If a commercial kit was not used, gene sequencing regions information is required.

ls your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *	
Yes	ř
Specify Variants Detected *	
(c.17/4_17/de1/AGA);306inSA p.Arg59Lysts*10 (C.17/5dupA);E60K p.Glu60Uys (C.17/8G>A);E60X p.Glu60* (C.17/8G>1);P67L p.Pro67Leu (C.200C>1 p.Arg75* (c.223C>T);365-366insT p.Trp79Leufs*32(c.233dupT);G85E p.Gly85Glu (c.254G>A);394de1TT p.Leu88llefs*22;(c.262_263de1TT);L88X p.1 (c.263T>A);L88X p.Leu88* (c.263T>G);G91R p.Gly91Arg (c.271G>A);405+1G>A p.2; (C.273+1G>A);405+3A>C p.? (c.273+3A>C);406-2A>G 2A>G);406-1G>A p.? (c.274-1G>A);E92K p.Glu92Lys (c.274G>A);E92X p.Glu92* (c.274G>T);Q98X p.Glu98* (c.292C>T);Q98R p.Gln98Arg (c.293A>G);409L p.Pro99Leu (c.296C>T);L102R p.Leu102Arg (c.301T>G);442de1A p.Arg104Glufs*3 (c.310de1A)	Leu88*
List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, yo list your website.	ou may instead
Was a gene sequencing method used? *	
Yes	~
# Was a commercial kit used? *	
No	~
For non-commercial kits, provide regions of the gene that are sequenced *	
for non-commercial kits, provide regions of the gene that are sequenced	
Specify Variants Detected * [D:174_T/7de1IR4A;]30binsA [D:Arg59Lysts*10 (C:175dupA);Eb0K [D:GlubOUtyS (C:178G>A);Eb0X [D:GlubOU*S (C:178G>A);26UX [D:GlubO* (C:178G>1);P67L [D:Pr067Leu (C:200C> p.Arg75* (C:223C>T);365-366insT [D:Trp79Leufs*32(C:233dupT);G85E [D:Gly85Glu (C:254G>A);394deITT [D:Leu88Ilefs*22;(C:262_263deITT);L88X [D:C253T>A);L88X [D:Leu88* (C:263T>G);G91R [D:Gly91Arg (C:271G>A);405+1G->A] [D; (C:273+1G>A);405+3A->C [D; 0? (C:273+3A>C);406-2A->C [D:C23A>G);406-1G->A [D; ? (C:273+1G>A);292K [D:Glu92Lys (C:274G>A);292X [D:Glu92* (C:274G>T);Q98X [D:Gln98A* (C:292C>T);Q98R [D:Gln98Arg (C:293A>G);P99L [D:Pr099Leu (C:296C>T);L102R [D:Leu102Arg (C:305T>G);422deIA [D:Arg104Glufs*3 (C:310deIA)] List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, list your website.	p.Leu88* G p.? (c.274-
Was a gene sequencing method used? *	
Yes	
* Was a commercial kit used? *	
No	
For non-commercial kits, provide regions of the gene that are sequenced *	
exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	
xtraction Method	
Select an extraction method *	0

1.4 Extraction Method Information

Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter method information including primary method, secondary/confirmatory method, and extraction method.

1. To select an extraction method, click on the magnifying glass and select a method from the search box.

Ex	traction Method	
	Select an extraction method *	
		٩

2. Choose a method then click 'Select'.

		Search Q
~	Name 🕇	
v	In-house alkaline lysis prep	
	In-house boiling prep	
	In-house Chelex method	
	In-house lysis boil prep	
	Other	
	Perkin Elmer/Chemagen Chemagic kit	
	Qiagen Generation DNA Purification & DNA Elution Solutions (also sold as 5 Prime Ea	sy PCR Solutions 1 & 2)
	Qiagen magnetic bead kit (EZ1 or BioSprint 96)	
<	2 >	

3. Continue to section 1.6 for guidance on reporting pathogenic variant data.

For non-commercial kits, provide regions of the gene that are sequenced *		
	exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22	
Ex	raction Method	
	Select an extraction method *	
	In-house alkaline lysis prep 🗶 🔍	
Pa	athogenic Variant Data	
	e variant you wish to enter is not found within the searchable listing, select "other" and then enter the variant in the field that will appear when "other" is selected.	

1.5 Specifying Variants Detected

If restrictions were placed or laboratory specific customizations were made on variants that are detectable using a primary or secondary/confirmatory method, the detected variants must be specified for each method.

The variants detected by your lab should be listed in the indicated text boxes. **Format each variant by separating each with a semicolon ";".** Alternatively, if the variants that your lab detect are on your program's website, reference your website.

CDC	
Home > Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)	
Cystic Fibrosis DNA Variant Detection Proficien Testing (CFDNAPT)	су
* Definition of commercial kit – A kit that has been designed by the manufacturer to sequence the CFTR	l gene
Method Information Primary Method	
Select a primary method *	
	Q
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *	
Yes	~
Specify variants detected: * List the variants detected by your lab-separate each variant by a semicolon ";". If the variants that your lab detect are on your program's website, you may instead list your website.	
Was a gene sequencing method used? *	
No	~
Secondary/Confirmatory Method Select a secondary/confirmatory method Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)	٩
Select an algorithm for utilization of the secondary/confirmatory method *	
	Q
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * Yes	~
Specify Variants Detected *	
List the variants detected by your lab-separate each variant by a semicolon "?". If the variants that your lab detect are on your program's website, you may instead list your website.	ß

To assist with formatting the variant list, the <u>delim.co</u> website can be used as a tool to generate the formatted list. The tool can be found <u>here</u>.

Delím.co - Free Comma Delimiter	Converter	About	Creators
Free Comma Separating Tool Do you often need to take a spreadsheet of data and convert to a comma-delimited list? Be it for taking a list of zip codes or names to make an SQL query, or to take data from a CSV and be able to paste into an array. At delim.co we make that just a little easier. Enter your non-delimited data on the left, hit the button, and boom, separated data on the right. Special configs are below if the defaults arent what you need!			
Column Data Here Delimite	ed Data H	ere	
1 1 2 3 4 5 6 7 7 6 7 8 9 4 9 10 11 12 12 13 14 15 15 16 16 17 18 19			
Converter Settings			
			O
About Delim.co			0
delimiter : de·lim·it·er A delimiter is a sequence of one or more characters used to specify the boundary between separate, independ data streams. An example of a delimiter is the comma character, which acts as a field delimiter in a sequence			

1. Paste the unformatted variant list in the box labeled 'Column Data Here'.

	t the button, and boom, separated data on the the defaults aren't what you need!
Column Data Here	Delimited Data Here
10 P7X p.Arg75* (c.224C1) 13 S5-36in.pl.rtp79(uefs2)(c.234dpT) 13 G555 p.Cly85CU (c.254GA) 13 G46d1T p.Lu881(c.253TA) 15 L88X p.Lu688* (c.253TA) 15 L88X p.Lu688* (c.253TA) 15 L88X p.Lu688* (c.253TA) 16 L88X p.Lu688* (c.253TA) 16 L88X p.Lu688* (c.253TA) 16 G1R p.Clu28* (c.253TA) 16 G1R p.Clu28* (c.253TA) 16 G1R p.Clu28* (c.253TA) 16 G1R p.Lu68* (c.253TA) 17 G1R p.Lu68* (c.253TA) 17 G1R p.Lu68* (c.253TA) 17 G1R p.Lu68* (c.274GA) 17 G1R p.Lu68* (c.274GA) 18 G2X p.Clu22* (c.274GA) 19 G2X p.Clu22* (c.274GA) 19 G2X p.Clu22* (c.274GA) 10 G2X p.Lu68* (c.293C1) 20 G28 p.Clu78* (c.293C1) 21 G28 p.Lu688* (c.293C1) 21 G28 p.Lu688* (c.293C1) 21 G28 p.Lu688* (c.2365T6) 21 G28 p.Lu688* (c.2365T	

2. Select the semicolon option for the drop-down menu.

Delim.co - Free Comma Delimiter		Creators
Enter your non-delimited data on t	he left, hit the button, and boom, separated data on the e below if the defaults aren't what you need!	
Column Data Here	Delimited Data Here	- 1
31 365-366jnst[p.rtp?9le.uf*si2(c.233dupT) 32 685E p.cly85du (c.25405A) 33 394delTT p.teu881lefs*22 34 (c.262_263delTT) j.teu88* (c.26375A) 35 L88X p.teu88* (c.26375A) 36 L88X (c.1eu88* (c.26375A) 36 L88X (c.26375A) 37 G914 p.cly91ar(c.27105A) 38 405-165A (c.273130xC)	2 3 4 5 6 7 8 9 10	1
40 406-2A-56 p.? (c.274-2A56) 41 406-16-5A p.? (c.274-165A) 42 E92K p.Glu92Lys (c.27465A) 43 E92K p.Glu92Lys (c.27465A) 44 Q98K p.Glu98Lys (c.27465A) 45 E92K p.Glu98Lys (c.27465A) 46 Q98K p.Glu98Lys (c.27465A) 47 D91L p.Glu98Lys (c.27465A) 48 P.P.091Les (c.29267T) C.29365(T) 46 P99L p.Pro99Les (c.29365(T) 47 L107R p.Leu02Arg (c.30575) 48 424de1A p.Arg104Glufs*3 (c.310de1A) v	1 11 12 33 5paces 14 15 16 New Line 17 19	000
	Converter Settings	8 ⁺

3. Select the blue right pointing arrow.

Delim.co - Free Comma Delimiter	Converter About	Creators
Enter your non-delimited data on the left, hit the bu right. Special configs are below if the def	tton, and boom, separated data on the	
Column Data Here	Delimited Data Here	
30 R75X p.Arg75* (c.223C1) 31 355-366inT[p.Tp.PPJeuf5*32(c.233dupT) 32 394delTT p.leu8811cf*22 33 394delTT p.leu8811cf*22 35 (s.252_253delTT) 35 (s.262_263delTT) 35 (s.262_263delTT) 36 (s.262_263delTT) 37 (c.484) 39 (s.267) 39 (s.267) 39 (s.276) 30 (s.276) 30 (s.276) 30 (s.276) 31 (s.274) 31 (s.274) 31 (s.274) 32 (s.274) 34 (s.274) 35 (s.28) 36 (s.274) 37 (s.376) 38 (s.277) 3940 (s.376) <td>1 2 3 4 5 6 7 7 8 9 9 10 11 11 12 12 12 13 14 15 16 15 16 10 11 11 12 12 13 14 15 16 16 17 17 18 19 19 19 19 19 19 19 19 19 19</td> <td></td>	1 2 3 4 5 6 7 7 8 9 9 10 11 11 12 12 12 13 14 15 16 15 16 10 11 11 12 12 13 14 15 16 16 17 17 18 19 19 19 19 19 19 19 19 19 19	
Converter Sett	ings	e G

4. Copy formatted data and paste into the NSQAP CFDNAPT Portal page.



1.6 Specimen Results - Pathogenic Variant Data Entry

Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter CFDNAPT pathogenic variant results including allele 1, allele 2, and clinical assessment for each specimen. Navigation details can be found in section 1.1.

CDC		↑	Lab Information	-	Biochemical PT 🗸	Molecular PT	- qc -	Help 🗕 📔	
Home > Cystic Fibrosis DNA	/ariant Dete	ection Prof	ficiency Testing (CF	DNAP	T)				
Cystic Fibro (CFDNAPT)	sis D	NA	Variant	D	etection	Profic	ciency	Testing	g
Pathogenic Varia If the variant you wish to enter is a Specimen Number 20211011001			chable listing, select "c	other" ar	nd then enter the variant ir	n the field that will	appear when "oth	er" is selected.	
Allele 1 *		Allele 2 *		_	Clinical Assessment *		Comments		
Gardinan Marakan	Q		٩			~			
Specimen Number 20211011002									
Allele 1 *		Allele 2 *			Clinical Assessment *		Comments		
	Q		Q			~			
Specimen Number 20211011003									
Allele 1 *		Allele 2 *		-	Clinical Assessment *		Comments		
Specimen Number 20211011004	٩		٩			~			
Allele 1 *		Allele 2 *		_	Clinical Assessment *		Comments		
	۹		٩			~			
Specimen Number 20211011005									
Allele 1 *		Allele 2 *		1	Clinical Assessment *		Comments		
Participating laboratories must generat laboratory normally sends specimens to convene to discuss response actions fo Use of trade names is for identification Laboratories.	o referral labora r the participant	tories for rout t which may ir	tine or confirmatory testir nclude termination from t	ig. If part he progra	icipants are found to have fals am.	ified or shared result	s or specimens, the N	SQAP committee will	
Save									
About NSQAP Self-Service This program is cosponsored by t (CDC) and the Association of Publ									

1. Click the magnifying glass to select a variant value for 'Allele 1' and 'Allele 2' for each specimen.

Pa	athogenic Va	riant Da	ata				
lf tł	ne variant you wish to ente	r is not found wi	thin the searchable list	ing, select "other" ar	nd then enter the variant in the	field that will	appear when "other" is selected.
	Specimen Number						
	20211011001						
	Allele 1 *	Q	Allele 2 *	Q	Clinical Assessment *	~	Comments

2. Search for variants using the search box. Click 'Select'.

				Search Q
-	Display Name	Variant cDNA name 🕹	Variant protein name	Variant legacy name
~	No variants detected	No variants detected	No variants detected	No variants detected
G330X (p.Gly330X) c.5		c.988G>T	p.Gly330X	G330X
1119delA (p.Gly330GlufsX39) c.98		c.987deIA	p.Gly330GlufsX39	1119delA
L320V (p.Leu320Val) c.958T>G		c.958T>G	p.Leu320Val	L320V
	1078delT (p.Phe316LeufsX12)	c.948delT	p.Phe316LeufsX12	1078delT
	G314E (p.Gly314Glu)	c.941G>A	p.Gly314Glu	G314E
	F311L (p.Phe311Leu)	c.933C>G	p.Phe311Leu	F311L
<	F311del (p.Phe312del)	5 6 7	8 43 >	
riant y cimer	1 2 3 4 genic Variant Dat you wish to enter is not found with n Number	ta	[Select Cancel Remove value
riant y	1 2 3 4 genic Variant Dat gou wish to enter is not found with Number 1001	ta	[in the field that will appear when "other" is se

3. Variants can be quickly located by typing the variant name in the provided search box.

				1898 C
~	Display Name	Variant cDNA name 🕇	Variant protein name	Variant legacy name
	1898+5G>T (c.1766+5G>T)	c.1766+5G>T	None	1898+5G>T
	1898+3A>G (c.1766+3A>G)	c.1766+3A>G	None	1898+3A>G
	1898+1G>T (c.1766+1G>T)	c.1766+1G>T	None	1898+1G>T
	1898+1G>C (c.1766+1G>C)	c.1766+1G>C	None	1898+1G>C
	1898+1G>A (c.1766+1G>A)	c.1766+1G>A	None	1898+1G>A

4. Choose a clinical assessment code for each specimen by clicking the drop-down arrow.

	3	"other" and then enter the variant in the field that	E 1
Specimen Number			
20211011001			
Allele 1 *	Allele 2 *	Clinical Assessment *	Comments
No variants detect 🗶 Q	No variants detect 🗶 🤇	۲ v	
		Screen Negative-Normal	
Specimen Number		Screen Positive-1 or 2 variants	
20211011002		Assay Failure	
Allele 1 *	Allele 2 *	Clinical Assessment *	Comments
Q			

5. If 'Assay Failure' is chosen as the clinical assessment, choose it for both Allele 1 and Allele 2.

e Lo	okup rec	ords							×
1u 1							assay	٥	
-	Display Na	ime	Varia	nt cDNA name	÷	Variant protein name	Variant le	gacy name	
~	Assay Failu	re	Assay	Failure		Assay Failure	Assay Faile	ıre	
2									
-									
lu							Select Cance	el Remove value	
3					_				
		-							
geni	c Varia	ant D	Data						
nt you wish	to enter is r	not found	within the searcha	ble listing, sel	ect "oth	er" and then enter the varian	t in the field that v	vill appear when "ot	her" is s
nen Numb	ər								
11001									
1 *			Allele 2 *			Clinical Assessment	·	Comments	

6. If necessary, enter any comments into the appropriate comment box.

e variant you wish to	enter is	not found	d within the searchable li	sting, sel	ect "other	" and then enter the variant in th	e field that	will appear when "other" is select
Specimen Number								
20211011001								
Allele 1 *			Allele 2 *			Clinical Assessment *		Comments
Assay Failure	×	Q	Assay Failure	×	Q	Assay Failure	~	
Specimen Number 20211011002								
Allele 1 *			Allele 2 *			Clinical Assessment *		Comments
Allele 1								

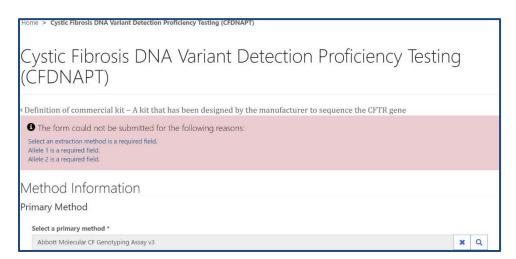
1.7 Save

1. Save all information and data by clicking the **'Save'** button located at the bottom of the page.

NOTE: All information and results must be saved at the same time. Data cannot be partially saved.

Allele 1 *		Allele 2 *		Clinical Assessment *	Comments
CFTRdup6b-10 (c.(Q	4259del5 (p.Leu13	x Q	Screen Positive-1 or 2 varia	
pecimen Number					
20211011005					
Allele 1 *		Allele 2 *		Clinical Assessment *	Comments
R31L (p.Arg31Leu)	Q	Q30X (p.Gln30X)	×Q	Screen Negative-Normal ~	
			not share NSQAP P	T test results or specimens with any other labora	
tory normally sends specimens ne to discuss response actions	s to referral la for the partic	aboratories for routine or confir ipant which may include termir	matory testing. If p nation from the pro	articipants are found to have falsified or shared gram.	

2. If you attempt to save the form without entering **all required fields** you will receive an error message. Complete the missing fields and click 'Save' again.



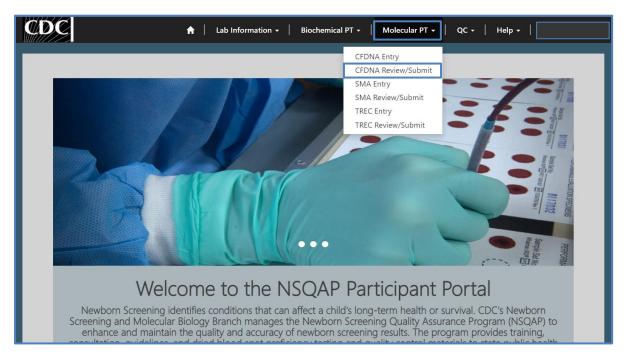
3. After you have successfully saved your results, you will be redirected to the 'CFDNA Review/Submit Page'. See section 2 for more guidance on this page.

2. CFDNAPT Review & Submit Page

2.1 Navigation

CFDNAPT program participants should review and submit data in the NSQAP Portal after program information and results have been entered and saved. (see section 1).

1. Select 'Molecular PT' then 'CFDNA Review/Submit' from the drop-down menu.

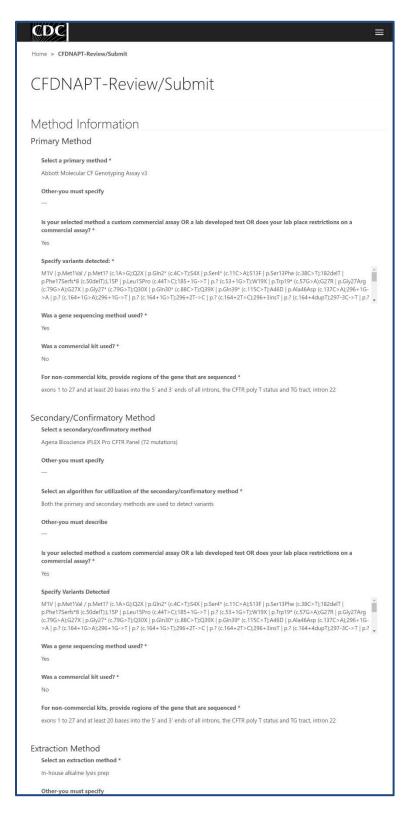


2. The CFDNA Review/Submit landing page will appear. Select **'CFDNA'** to navigate to the review and submit page.

CDC	A Lab Information - Biochemical PT -	Molecular PT 🗸 QC 🖌 🍐 Help 🗸 🍐			
Home > CFDNAPT- Review/Submit					
CFDNAPT- Revie	ew/Submit				
Name 🕇	Submitted By	Modified On			
CFDNA		2/25/2021 1:16 PM			
About NSQAP Self-Service Portal					
This program is cosponsored by the Centers f (CDC) and the Association of Public Health La					

2.2 Review

Navigate to the 'CFDNA Review/Submit Page' to review CFDNAPT program method information and specimen results in a read-only format. Navigation details can be found in section 2.1.

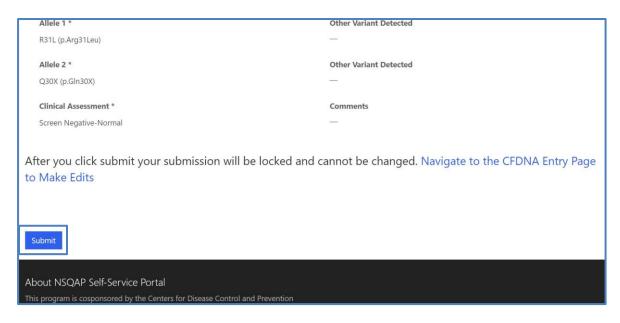


Pathogenic Variant Data	
Specimen Number	
20211011001	
Allele 1 * Q1412X (p.Gln1412X)	Other Variant Detected
Allele 2 *	Other Variant Detected
C276X (p.Cys276X)	_
Clinical Assessment *	Comments
Screen Negative-Normal	-
Considered Manufact	
Specimen Number 20211011002	
Allele 1 *	Other Variant Detected
849delG (p.Leu240X)	
Allele 2 *	Other Variant Detected
D1152H (p.Asp1152His)	
Clinical Assessment *	Comments
Screen Negative-Normal	-
Considered Manufact	
Specimen Number 20211011003	
Allele 1 * 4374+1G>T (c.4242+1G>T)	Other Variant Detected
Allele 2 *	Other Variant Detected
L1254X (p.Leu1254X)	_
Clinical Assessment *	Comments
Screen Positive-1 or 2 variants	-
Specimen Number	
20211011004	
Allele 1 * CFTRdup6b-10 (c.(743+1_744-1)_(1584+1_1585-1)dup)	Other Variant Detected
Allele 2 * 4259del5 (p.Leu1376SerfsX8)	Other Variant Detected
Clinical Assessment *	Comments
Screen Positive-1 or 2 variants	_
Specimen Number	
20211011005	
Allele 1 *	Other Variant Detected
R31L (p.Arg31Leu)	-
Allele 2 *	Other Variant Detected
Q30X (p.Gln30X)	
Clinical Assessment *	Community
Screen Negative-Normal	Comments
After you click submit your submission will be CFDNA Entry Page to Make Edits	locked and cannot be changed. Navigate to the
_	
Submit	
About NECAR Call Convice Parts	
About NSQAP Self-Service Portal	

1. If edits are necessary, navigate back to the CFDNA entry page and make or click the link located at the bottom of the review and submit page labeled **'Navigate to the CFDNAPT Entry Page to Make Edits'**.

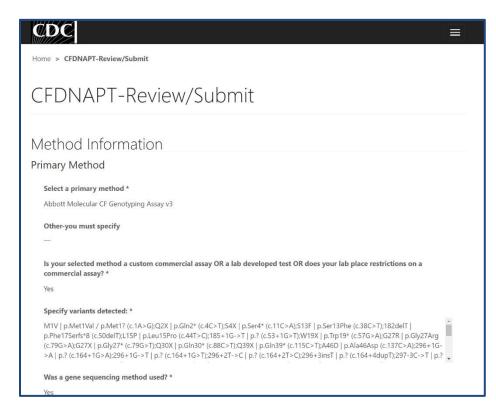
Specimen Number	
20211011005	
Allele 1 *	Other Variant Detected
R31L (p.Arg31Leu)	_
Allele 2 *	Other Variant Detected
Q30X (p.Gln30X)	—
Clinical Assessment *	Comments
Screen Negative-Normal	—
	nd cannot be changed. Navigate to the CFDNA Entry Page
to Make Edits	
Submit	

2. If no further edits are needed, results can be submitted by clicking the 'Submit' button. See section 2.3 for additional details.

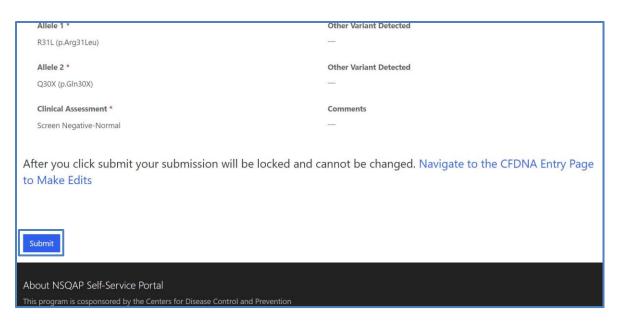


2.3 Submit

1. Navigate to the 'CFDNA Review/Submit Page' to submit CFDNAPT method information and data.

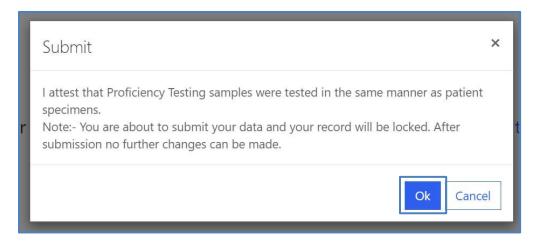


2. After reviewing the CFDNA review and submit page, click on the '**Submit**' button located at the bottom of the page.

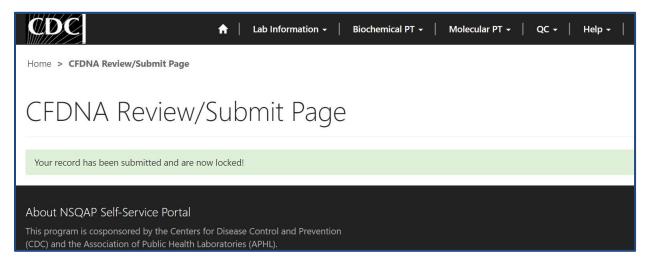


3. You will be prompted to confirm that you are ready to submit. Click **'Ok'** to confirm and submit your CFDNAPT results.

<u>NOTE</u>: You are only allowed to submit your results **ONCE**. You must review and confirm your entered information is accurate **BEFORE** submitting.



4. After submitting you will be directed to a confirmation page.



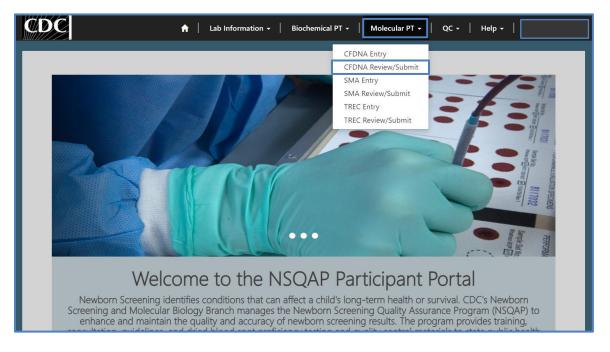
5. Once your CFDNAPT results are submitted you will no longer be able to access the 'CFDNA Entry' page. You can view your submitted data in a read-only format by accessing the review and submit page (see sections 2.1 and 2.2).

2.4 Save Data – Pdf Format

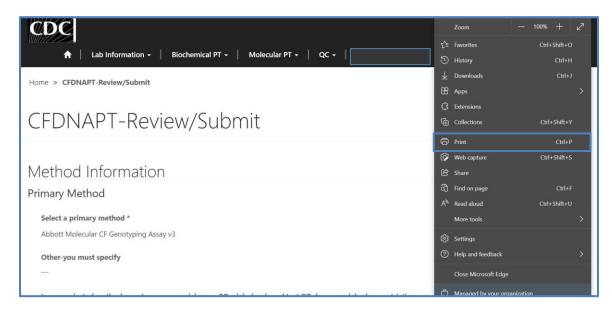
Submitted data can be saved in a pdf format by using the 'Save a PDF' function included in your web browser.

Note: The location and appearance of this functionality will vary depending on the web browser being used.

1. Navigate to the review and submit page as described in section 2.1.



2. Locate the "Print' function on your web browser.



3. Select 'Save as PDF'.

Home > CFDNAPT-Review/Submit
CFDNAPT-Review/Submit
Method Information
Primary Method Select a primary method * Abbott Molecular CF Genotyping Assay v3
Other-you must specify —
Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? * assay? * Yes
Specify variants detected: * M1V p.Met1Val / p.Met1? (c:1A>G);Q2X p.Gin2* (c:4C>T);S4X p.Ser4* (c:11C>A);S13F p.Ser13Phe (c:38C>T);182delT p.Phe17Serfs*8 (c:50delT);L15P p.Leu15Pro (c:44T>C);185+1G>T p.? (c:53+1G>T);W19X p.Trp19* (c:57G>A);G27R p.Giy27Arg (c:79G>A);G27X

4. Select 'Landscape' as the layout choice.

Print ? Total: 8 pages	Home > CFDNAPT-Review/Submit
Printer Save as PDF V	CFDNAPT-Review/Submit
Layout Ortrait	Method Information
O Landscape	Primary Method Select a primary method * Abbott Molecular CF Genotyping Assay v3
Pages O All	Other-you must specify
e.g. 1-5, 8, 11-13	Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *
More settings ~ Troubleshoot printer issues	Yes Specify variants detected: *
Save Cancel	M1V p.Met1Val / p.Met1? (c.1A>G);Q2X p.Gin2* (c.4C>T);S4X p.Ser4* (c.11C>A);S13F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);L15P p.Leu15Pro (c.44T>C);185+1G->T p.? (c.53+1G>T);W19X p.Trp19* (c.57G>A);G27R p.Gly27Arg (c.79G>A);G27X

5. Select 'More Settings'.

Pages	Abbott Molecular CF Genotyping Assay v3
	Other-you must specify
e.g. 1-5, 8, 11-13	
More settings \sim	assay? * Yes
Troubleshoot printer issues	Specify variants detected: *
Save Cancel	M1V p.Met1Val / p.Met1? (c.1A>G);Q2X p.Gin2* (c.4C>T);S4X p.Ser4* (c.11C>A);S13F p.Ser13Phe (c.38C>T);182delT p.Phe17Serfs*8 (c.50delT);L15P p.Leu15Pro (c.44T>C);185+1G->T p.? (c.53+1G>T);W19X p.Trp19* (c.57G>A);G27R p.Gly27Arg (c.79G>A);G27X

6. Adjust the scale percentage to 60%.

Total: 4 pages	Home > CFDNAPT-Review/Submit	
· · · · · · · · · · · · · · · · · · ·	CFDNAPT-Review/Submit	
Fewer settings A		
	Method Information	
Paper size	Primary Method	
(. w/	Select a primary method *	
Letter 🗸	Abbott Molecular CF Genotyping Assay v3	
	Other-you must specify	
Scale (%)		
	Is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *	
60	Yes	
	Specify variants detected: *	
Pages per sheet	Mini (See 1997) 2004; Str. 1997; Str. 1997; St. 2015; St. 1997; St. 2017; St	
1 ~	Was a gene sequencing method used? *	
	Yes	
Margins	Was a commercial kit used? *	
	No	
Default 🗸	For non-commercial kits, provide regions of the gene that are sequenced *	
	exons 1 to 27 and at least 20 bases into the 5 and 3 ends of all introns, the CFIR poly T status and TG tract, intron 22	
Save Cancel	Secondary/Confirmatory Method	
Current	Select a secondary/confirmatory method	
Other-you must specify	Agena Bioscience IPLDX Pro CFTR Panel (72 mutations)	

7. Select 'Save' to save the pdf file to your local drive's folder of choice.

Print ?	
al: 4 pages	Home > CFDNAFT-Review/Submit
×	CFDNAPT-Review/Submit
settings 🔨	
	Method Information
ze	Primary Method
	Select a primary method *
er 🗸 🗸	Abbott Molecular CF Genotyping Assay v3
	Other-you must specify
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	is your selected method a custom commercial assay OR a lab developed test OR does your lab place restrictions on a commercial assay? *
	163
	Specify variants detected: *
s per sheet	MVI () pAMATRIA ()
\sim	Was a gene sequencing method used? *
	Yes
	Was a commercial kit used? *
	No
t ~	For non-commercial kits, provide regions of the gene that are sequenced *
	exons 1 to 27 and at least 20 bases into the 5' and 3' ends of all introns, the CFTR poly T status and TG tract, intron 22
Save Cancel	Secondary/Confirmatory Method
Califer	Select a secondary/confirmatory method
	Agena Bioscience iPLEX Pro CFTR Panel (72 mutations)