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

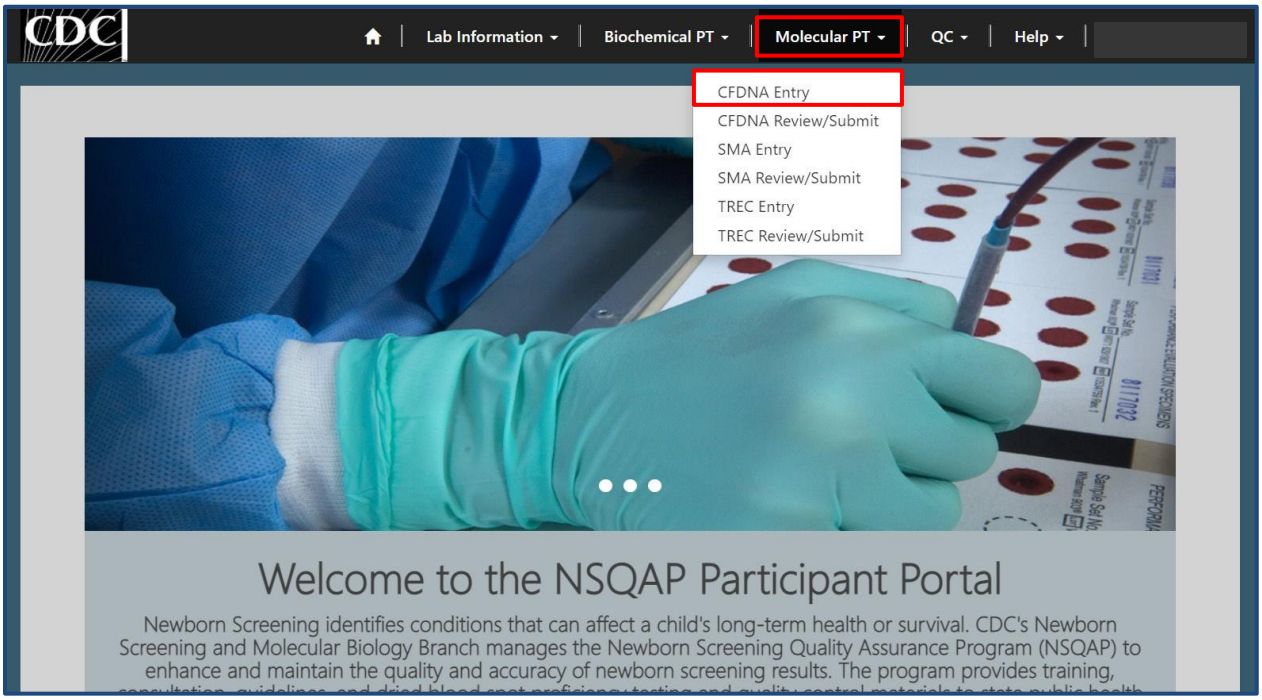
Table of Contents

1. CFDNAPT Program Entry Page

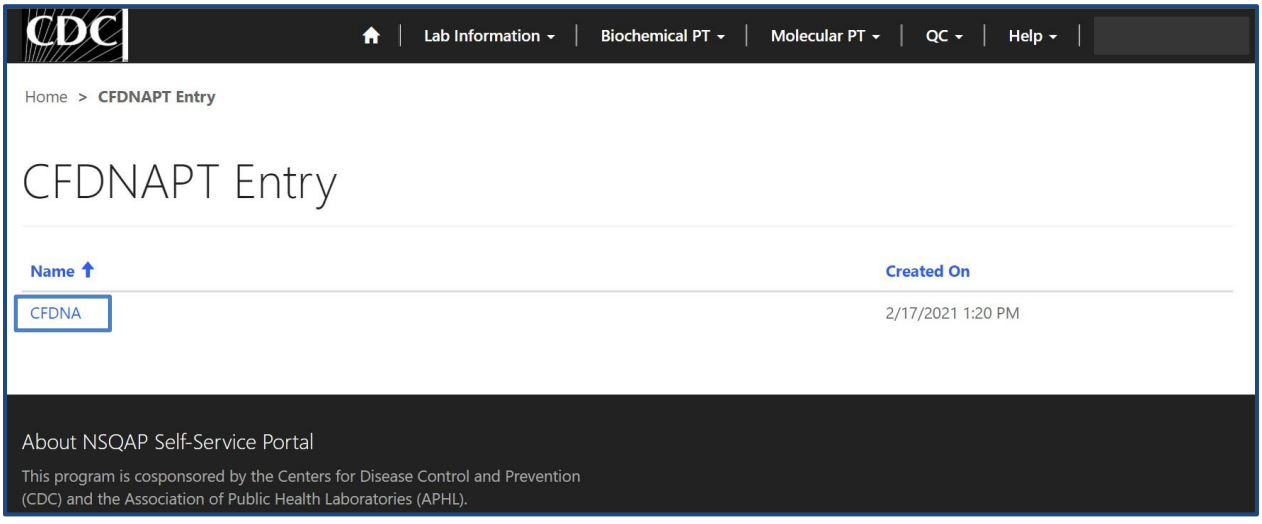
1.1 Navigation

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.



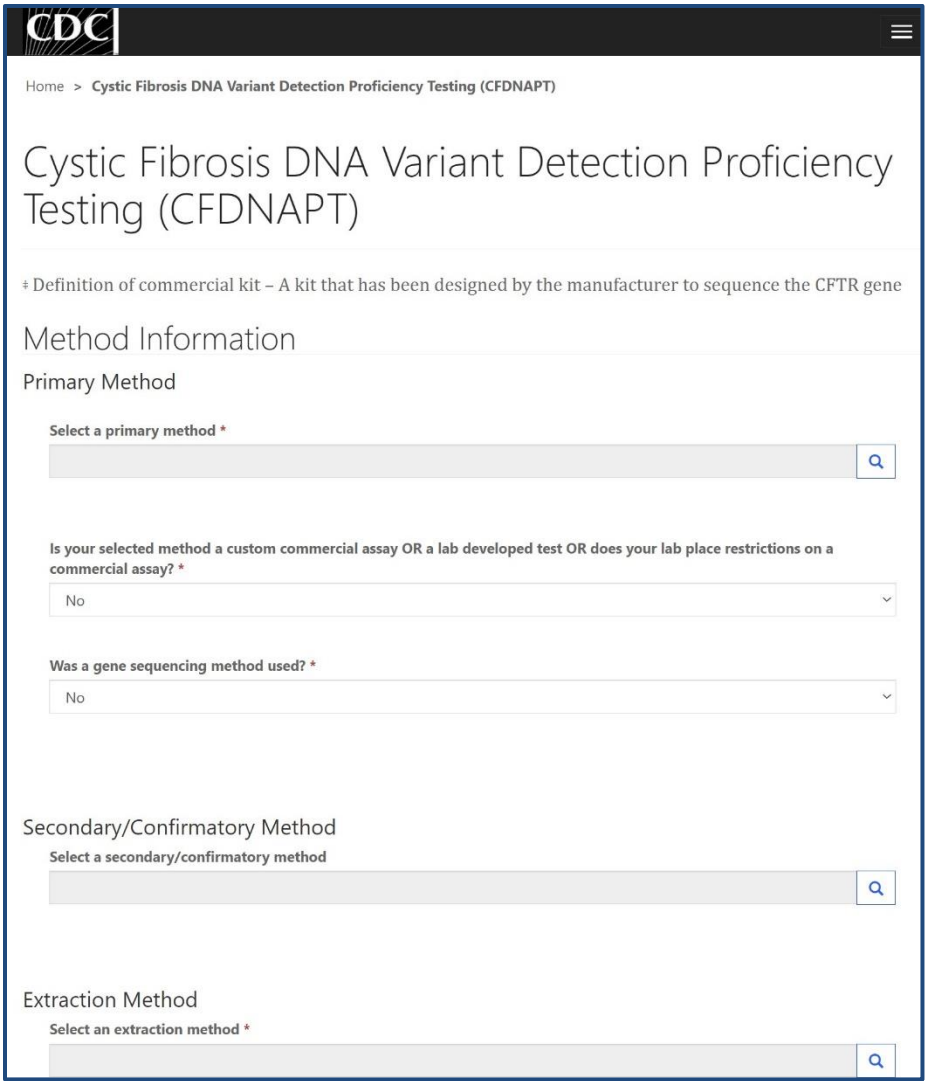
2. Select **'CFDNA'** to navigate to the entry page.



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

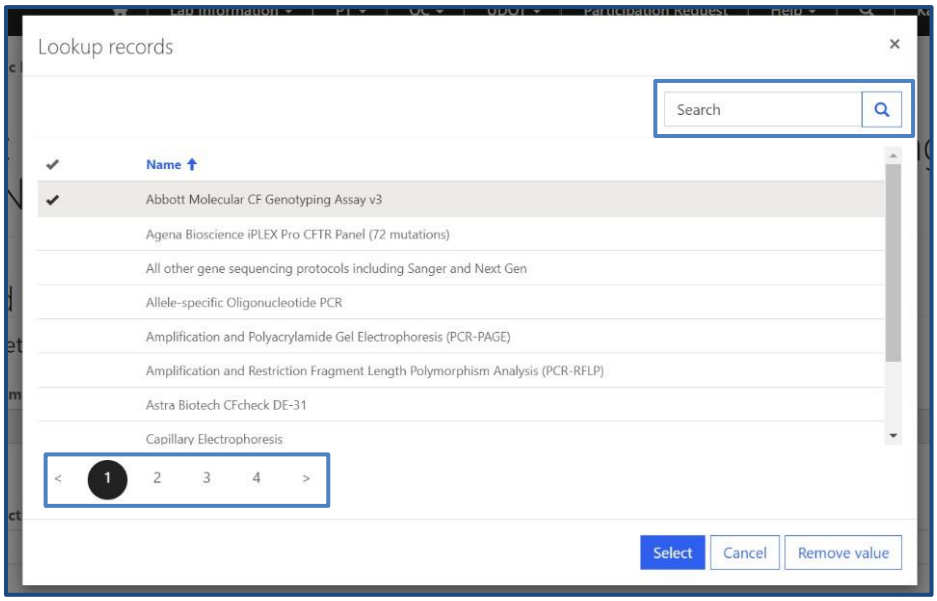
1.2 Primary Method Information

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.

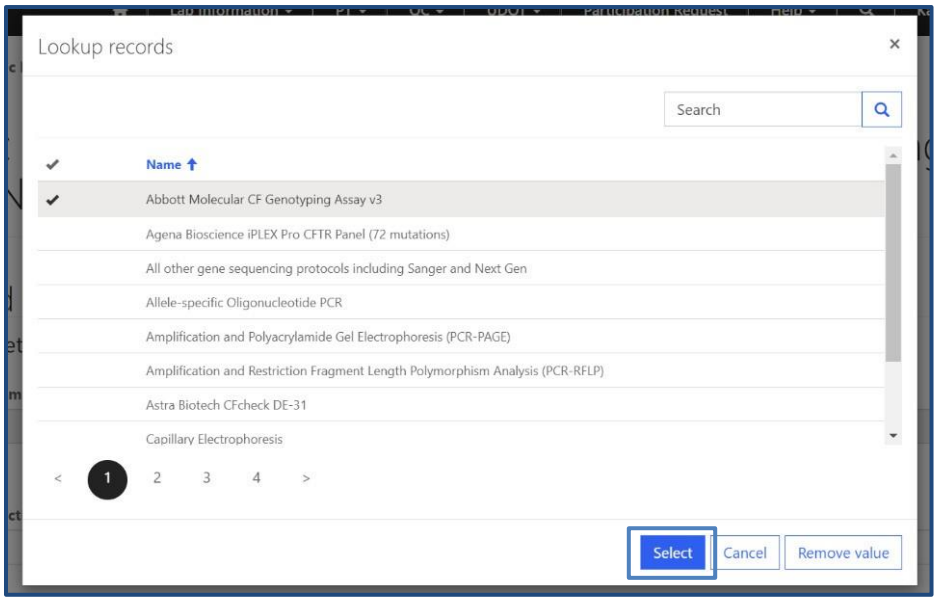


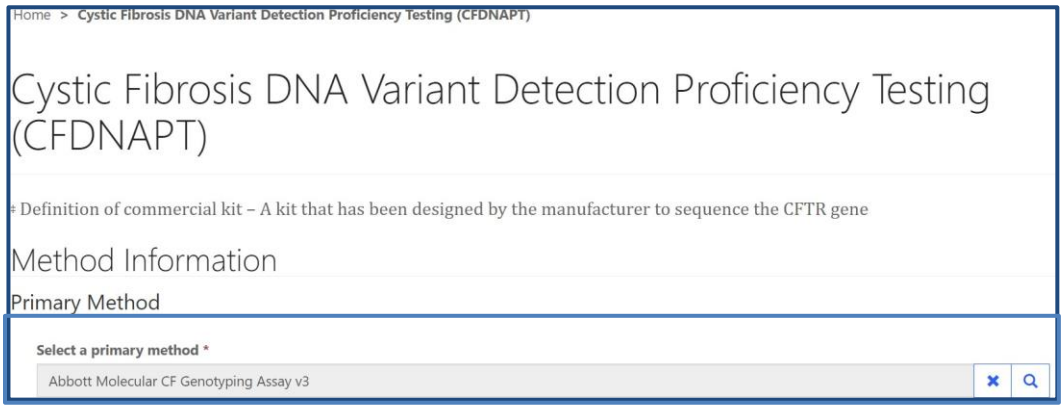
1. Click on the magnifying glass to see the primary methods list.

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

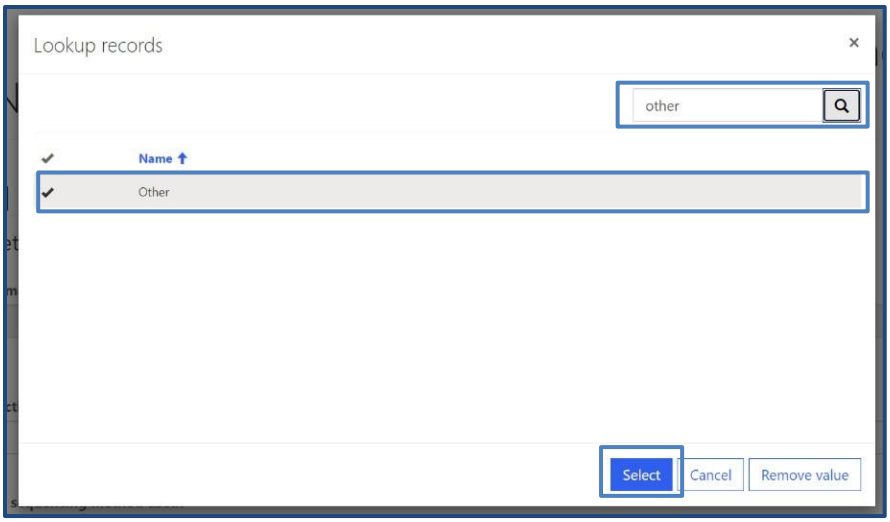


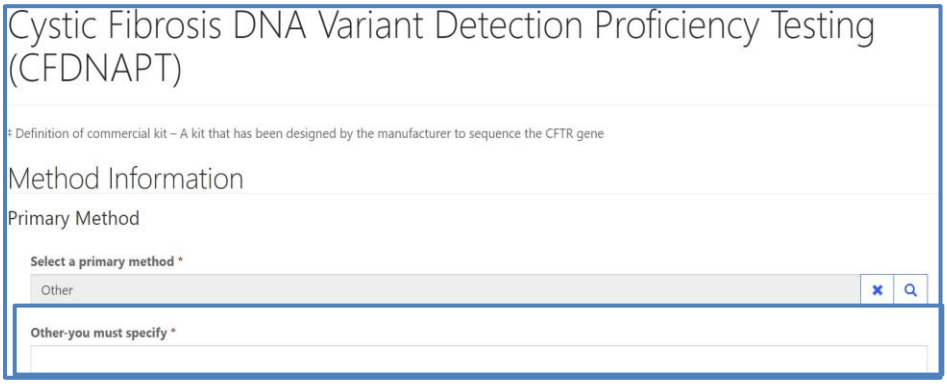
3. Choose a primary method then click ‘Select’.



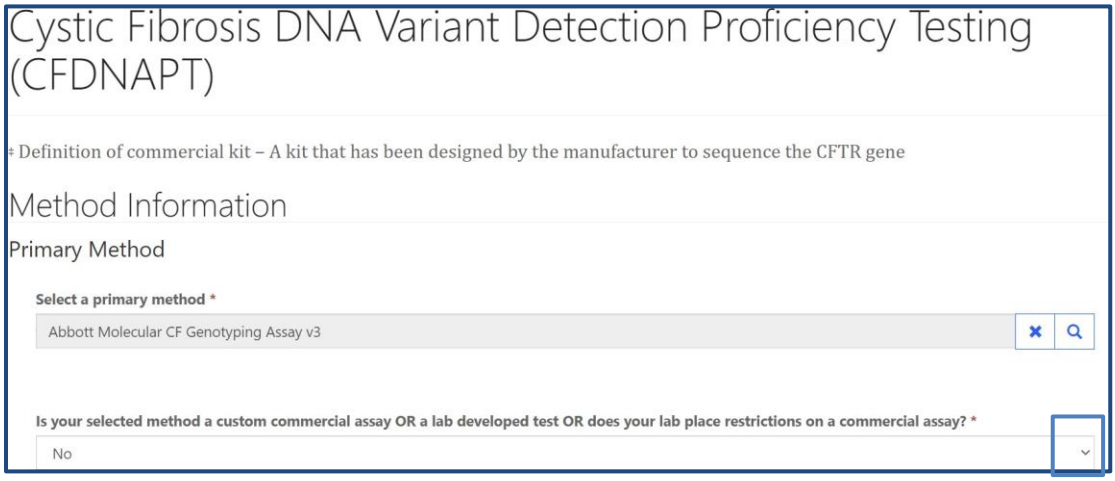


4. If 'Other' is selected, a text box will appear. You are **required** to specify your primary method details.

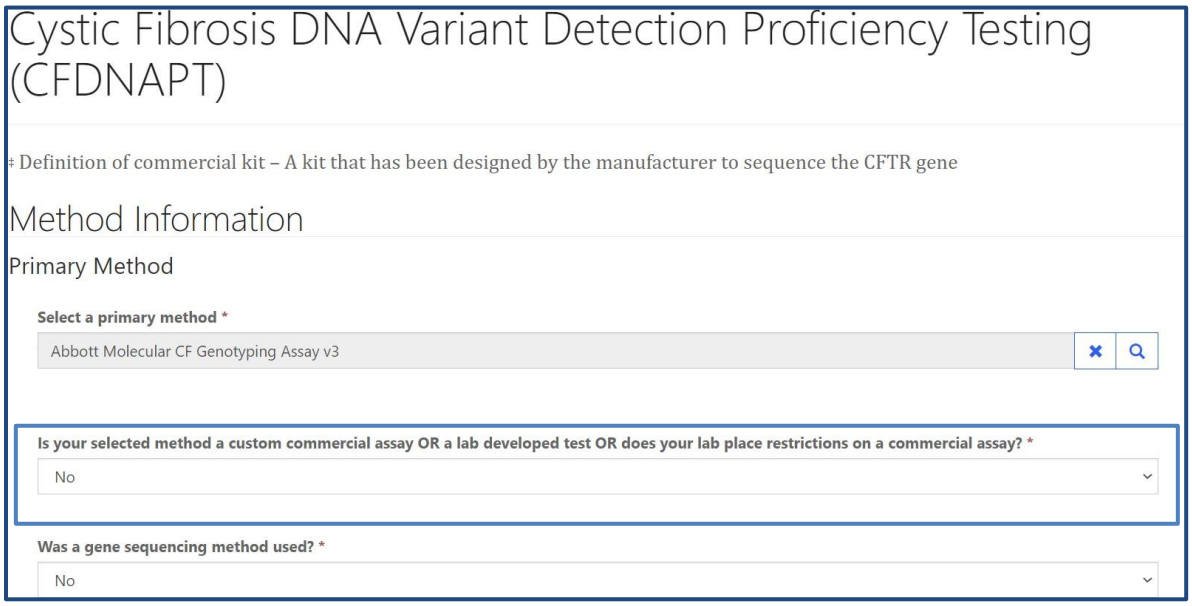




5. Indicate whether your selected primary method is a custom commercial assay, laboratory developed test, or a commercial assay with restrictions set by your lab by clicking the drop-down arrow.

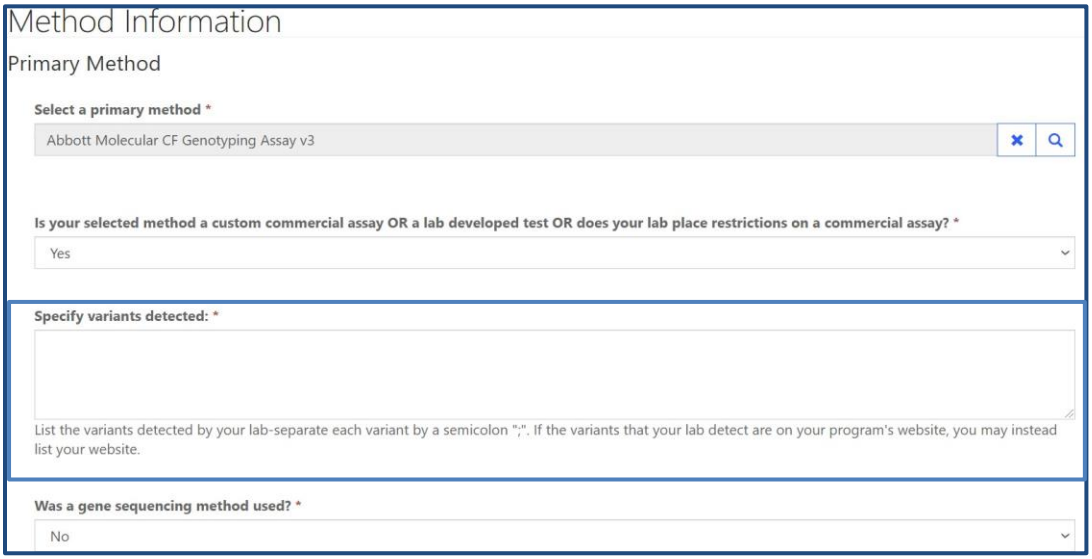


6. If no restrictions or laboratory specific customizations were made, no additional information is required.

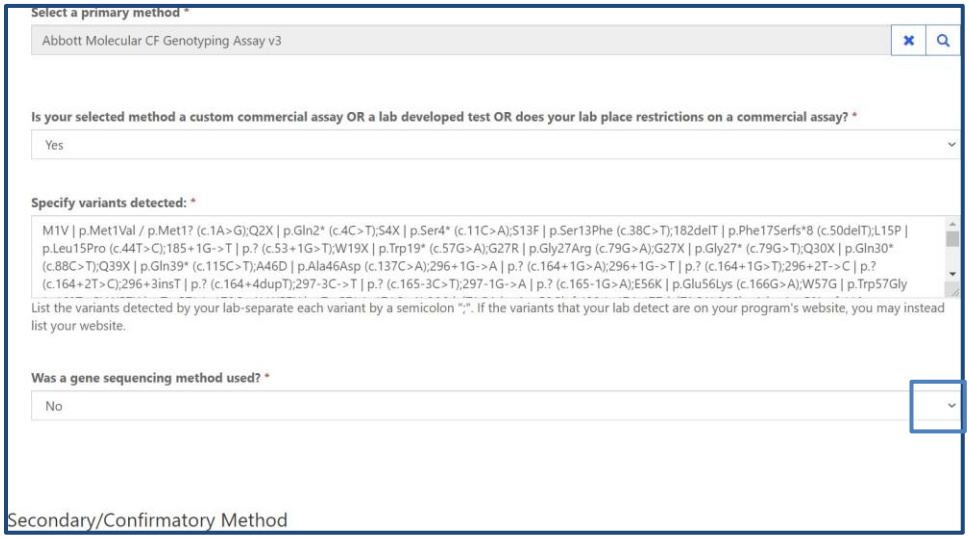


7. If restrictions were placed or laboratory specific customizations were made, you are required to specify the variants detected. See section 1.5 for additional information on a helpful tool for correctly formatting this information.

   <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.



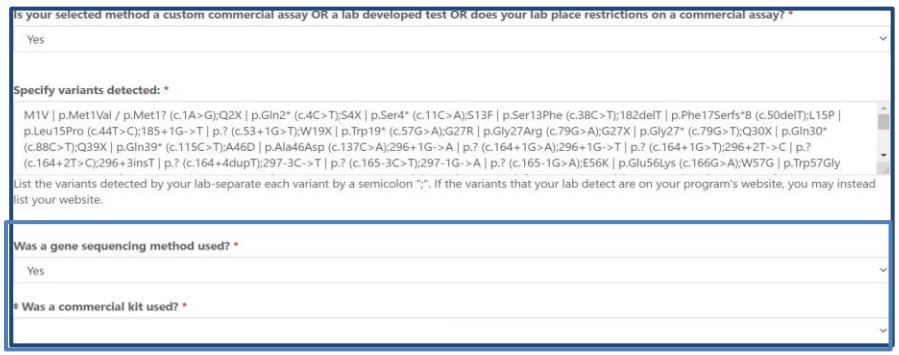
8. Indicate whether a gene sequencing method was used by clicking the drop-down arrow.



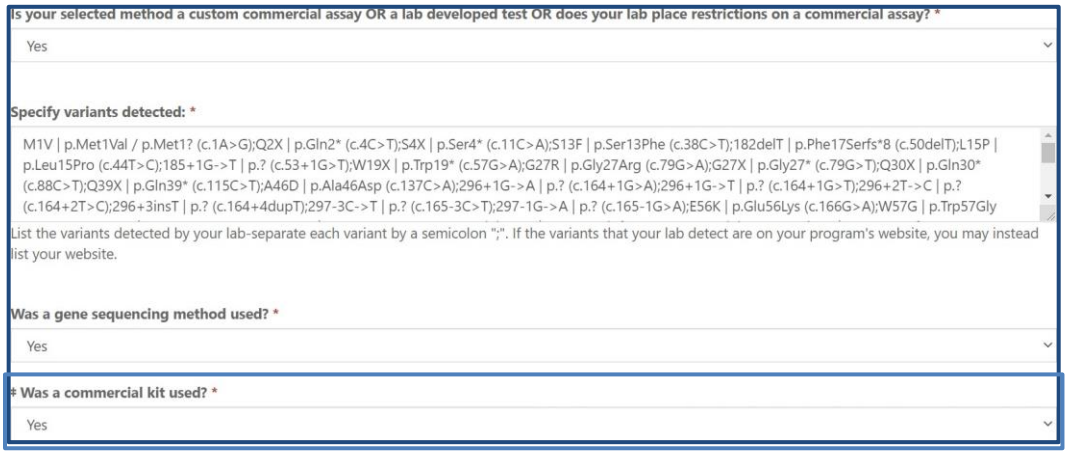
9. If a gene sequencing method was not used, no additional information is required.

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

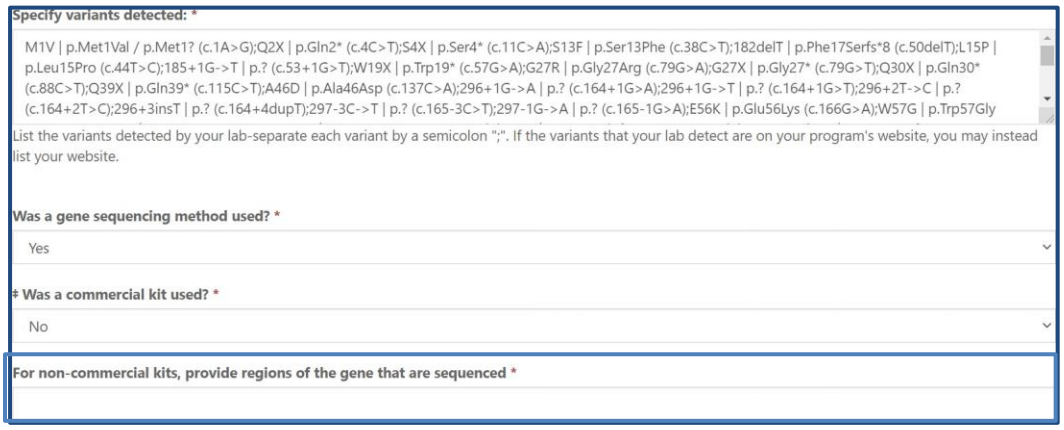
Note: A commercial kit is defined as a kit that has been designed by the manufacturer to sequence the CFTR gene.

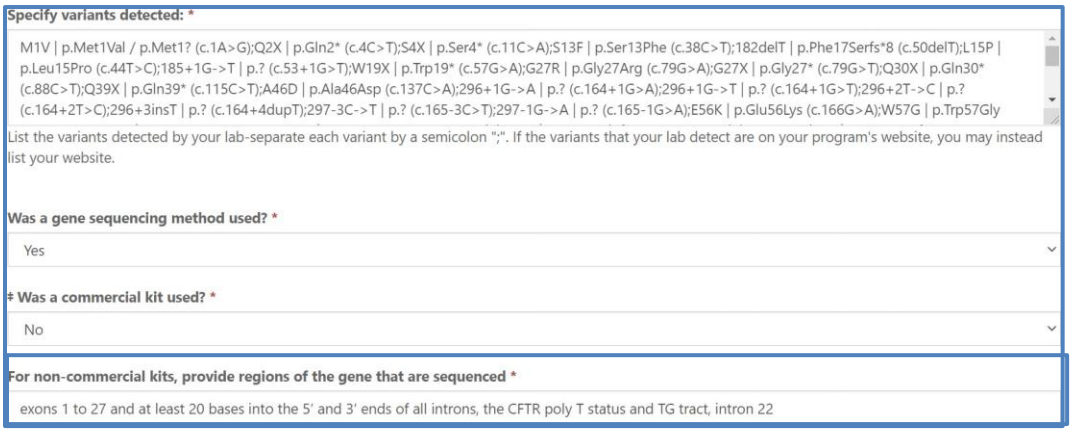


11. If a commercial kit was used, no further information is required.



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





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).

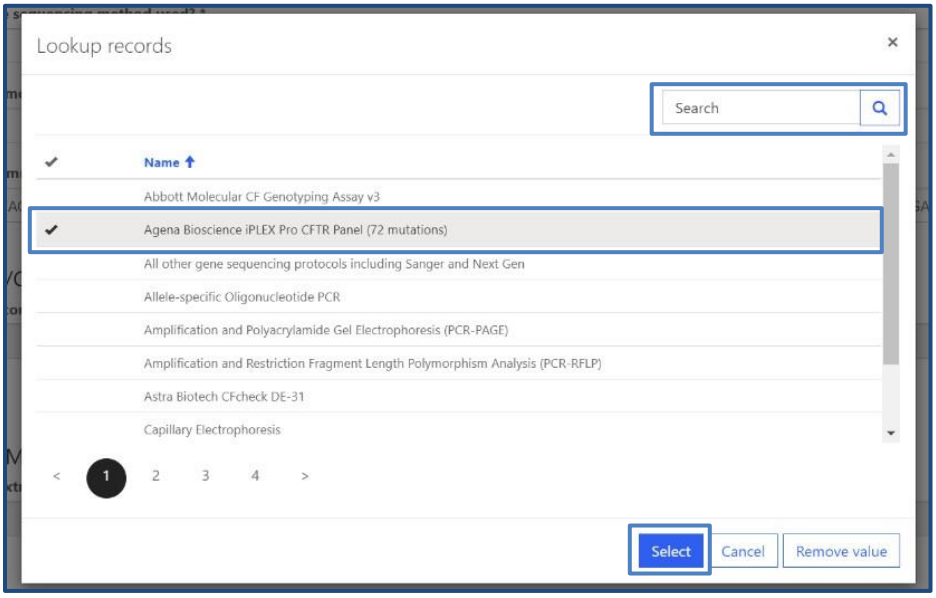
1.3 Secondary/Confirmatory Method Information

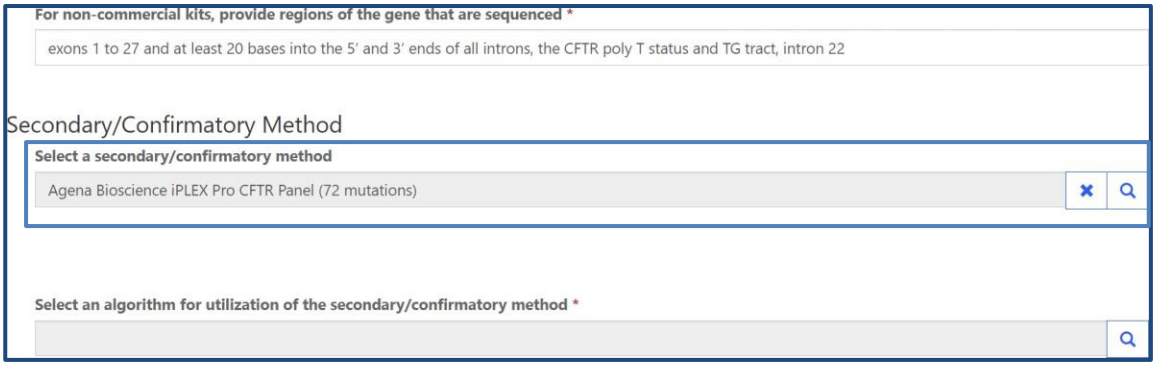
Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter method information including primary method, secondary/confirmatory method, and extraction method.

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.

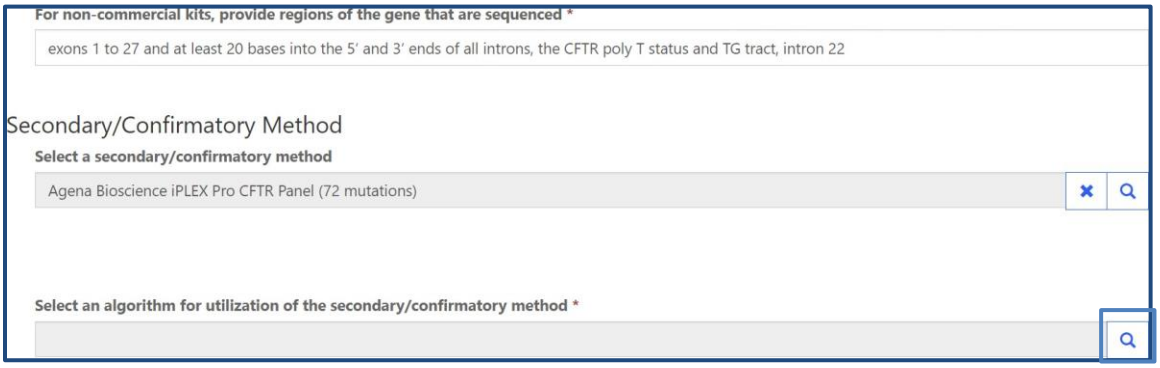
1. Select a secondary/confirmatory method by clicking on the magnifying glass.

2. Choose a method then click **'Select'**.

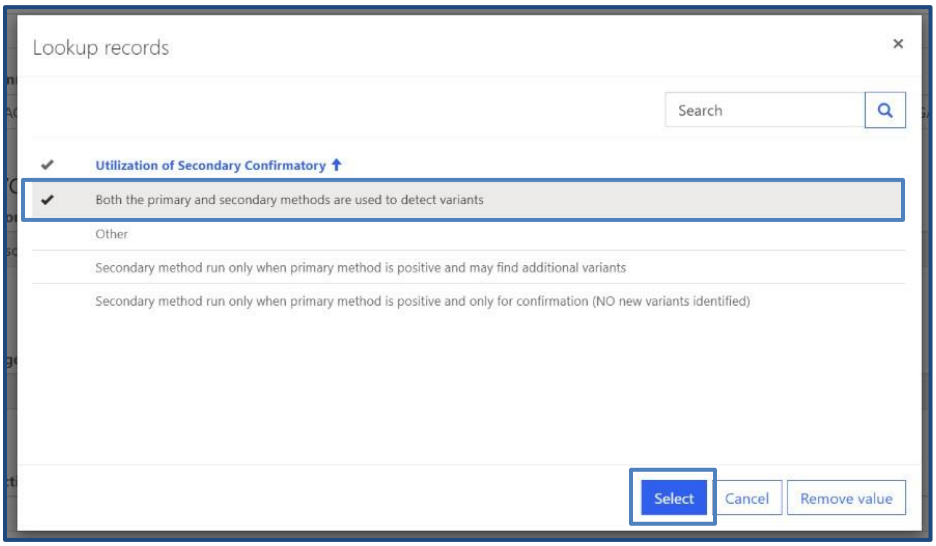


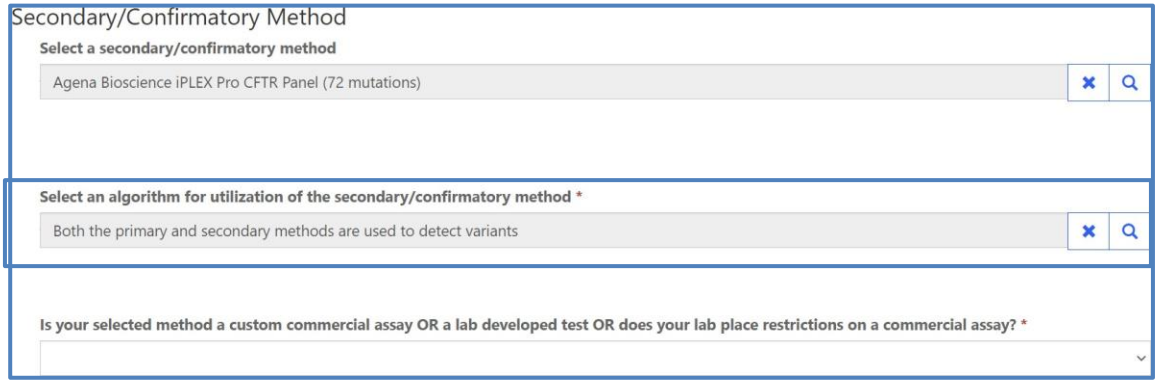


3. Select an algorithm for utilization of the secondary/confirmatory method by clicking on the magnifying glass.

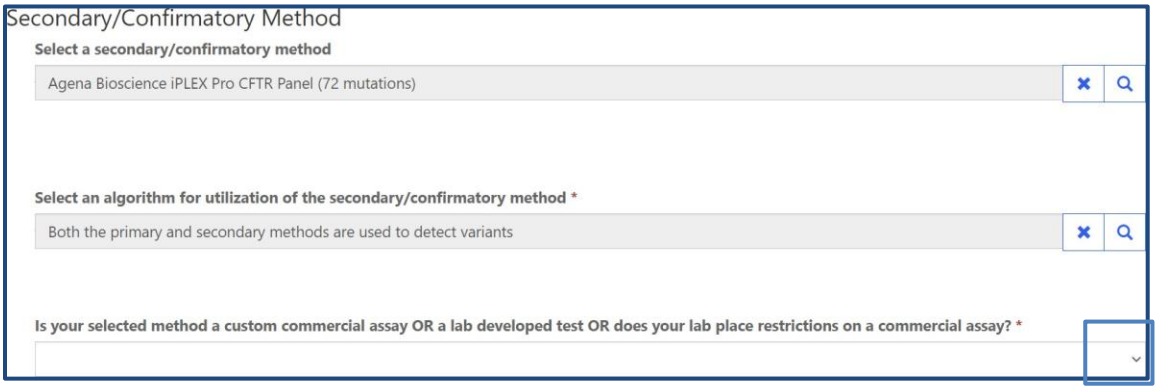


4. Choose an algorithm then click **'Select'**.

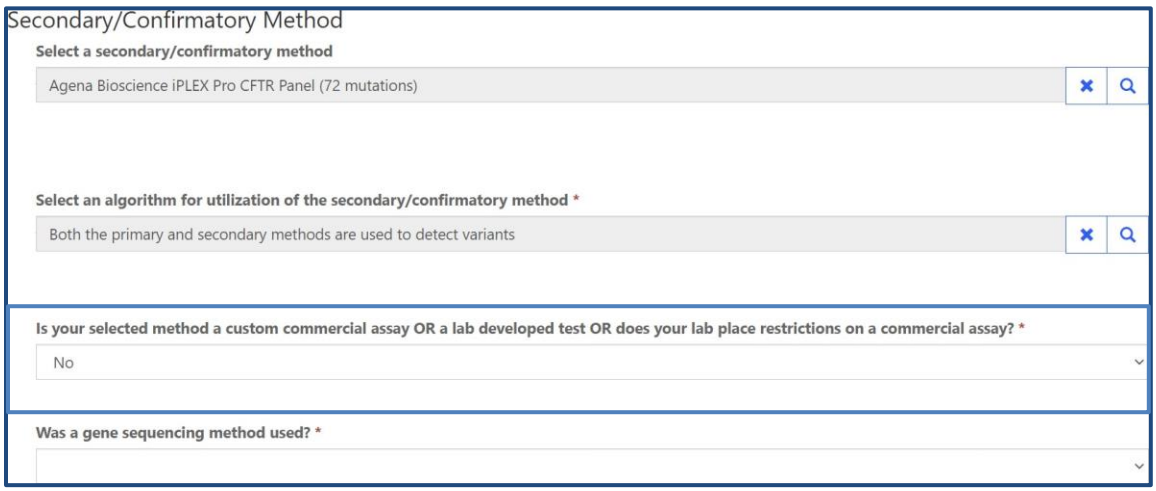




5. Click the drop-down arrow to indicate whether your selected secondary/confirmatory method is a custom commercial assay, laboratory developed test, or a commercial assay with restrictions set by your lab.

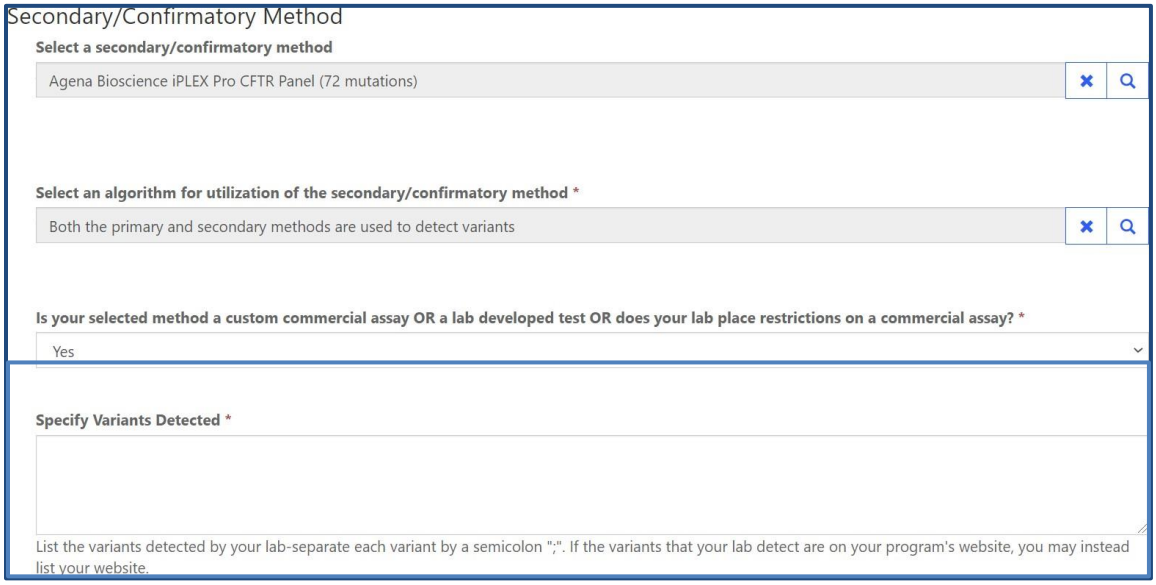


6. If no restrictions or laboratory specific customizations were made, no additional information is required.

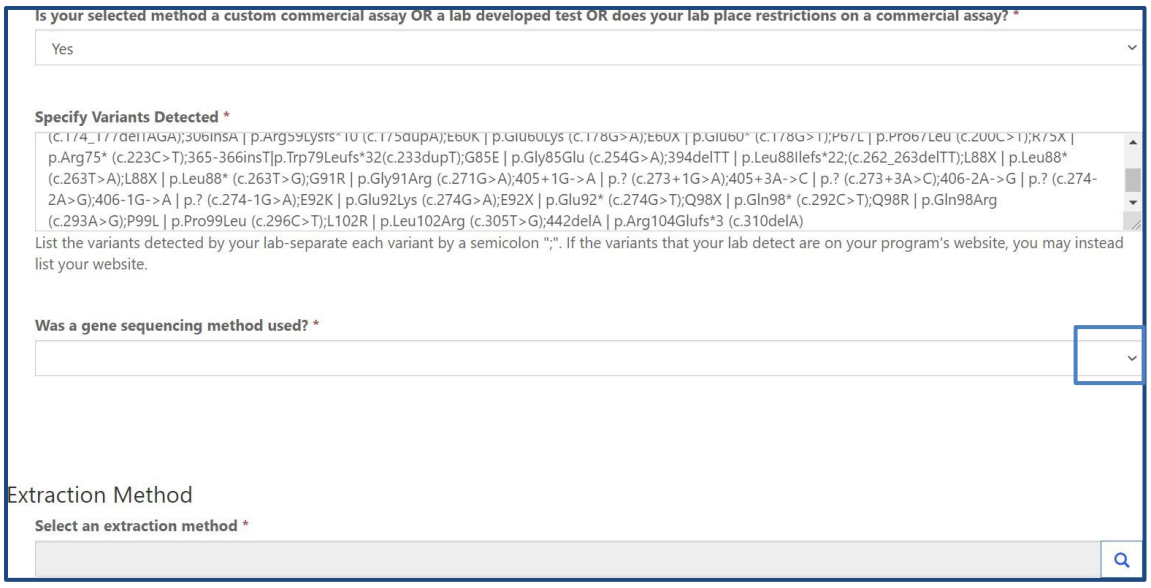


7. If restrictions were placed or laboratory specific customizations were made, you are required to specify the variants detected. See section 1.5 for additional information on a helpful tool for correctly formatting this information.

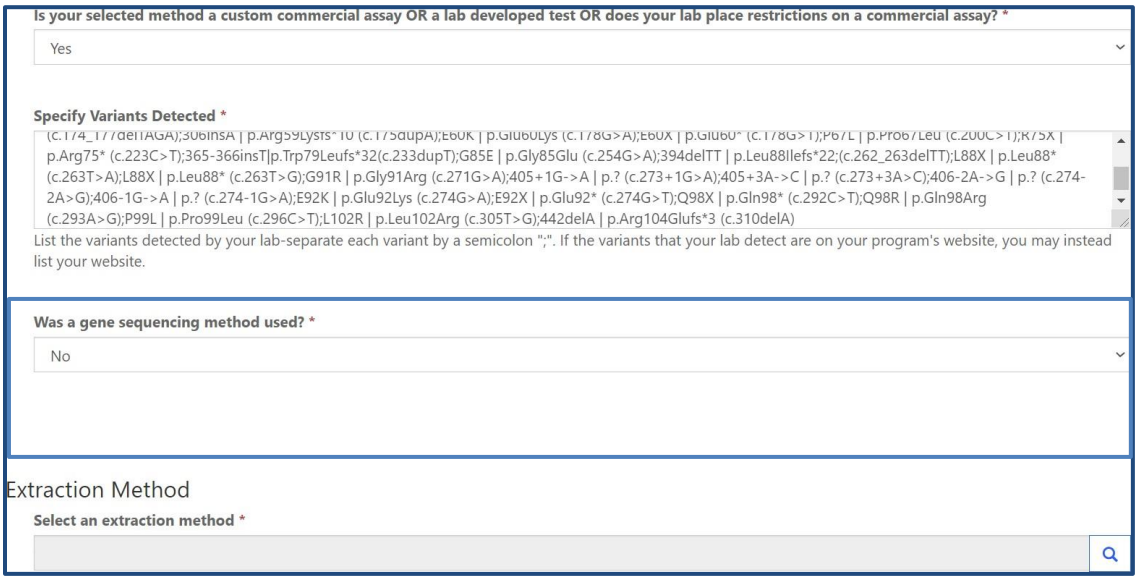
   NOTE: List the variants detected by your lab, separate each variant by a semicolon ";".
   If the variants that your lab detect are on your program's website, you may instead reference your website.



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



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

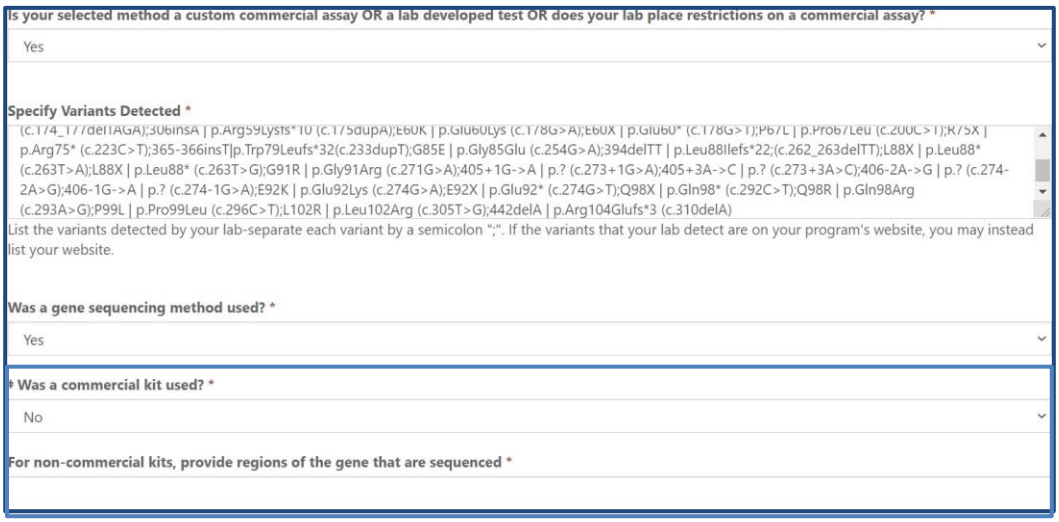


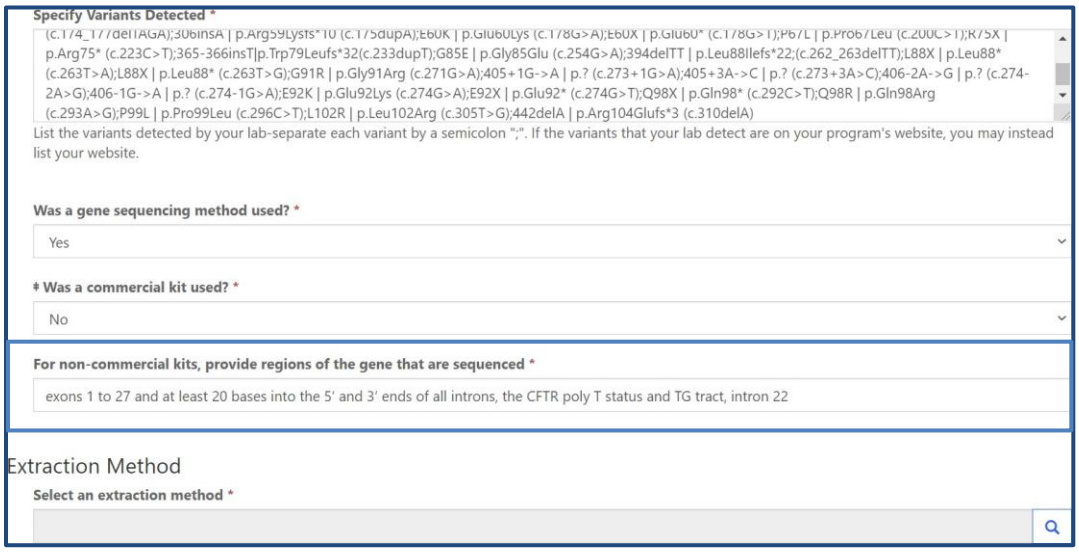
10. If a gene sequencing method was used, indicate whether a commercial kit was used.

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

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

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

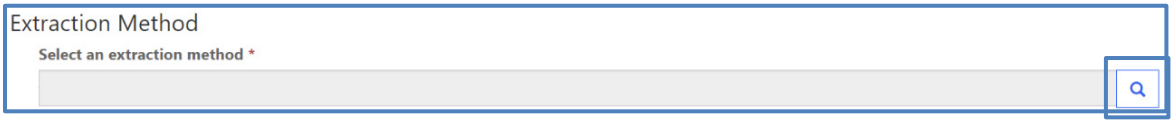




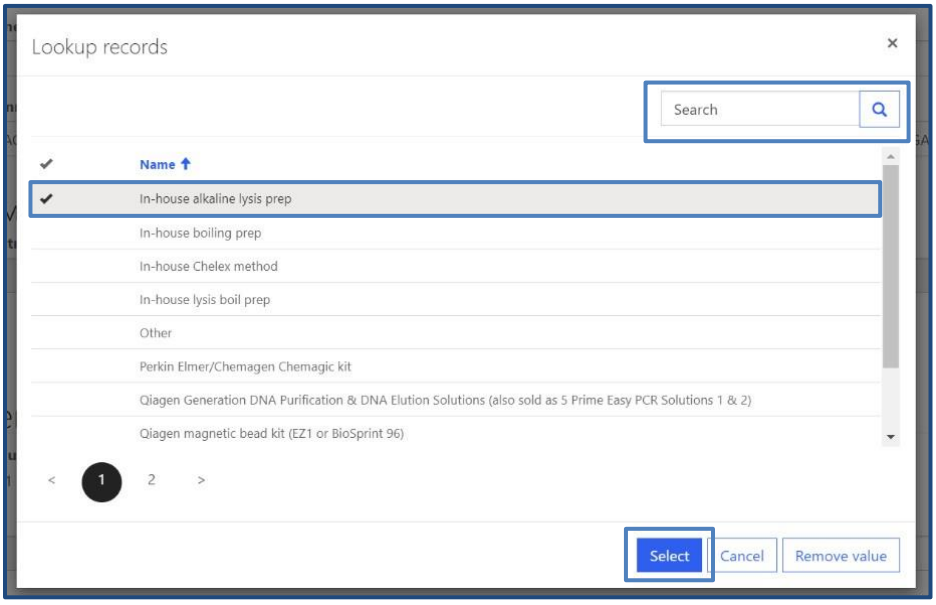
1.4 Extraction Method Information

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.

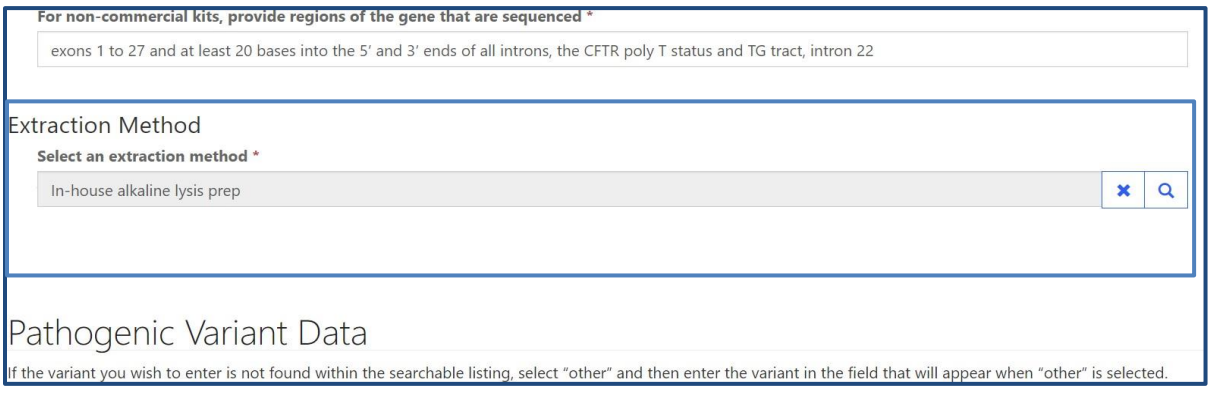
1. To select an extraction method, click on the magnifying glass and select a method from the search box.



2. Choose a method then click 'Select'.



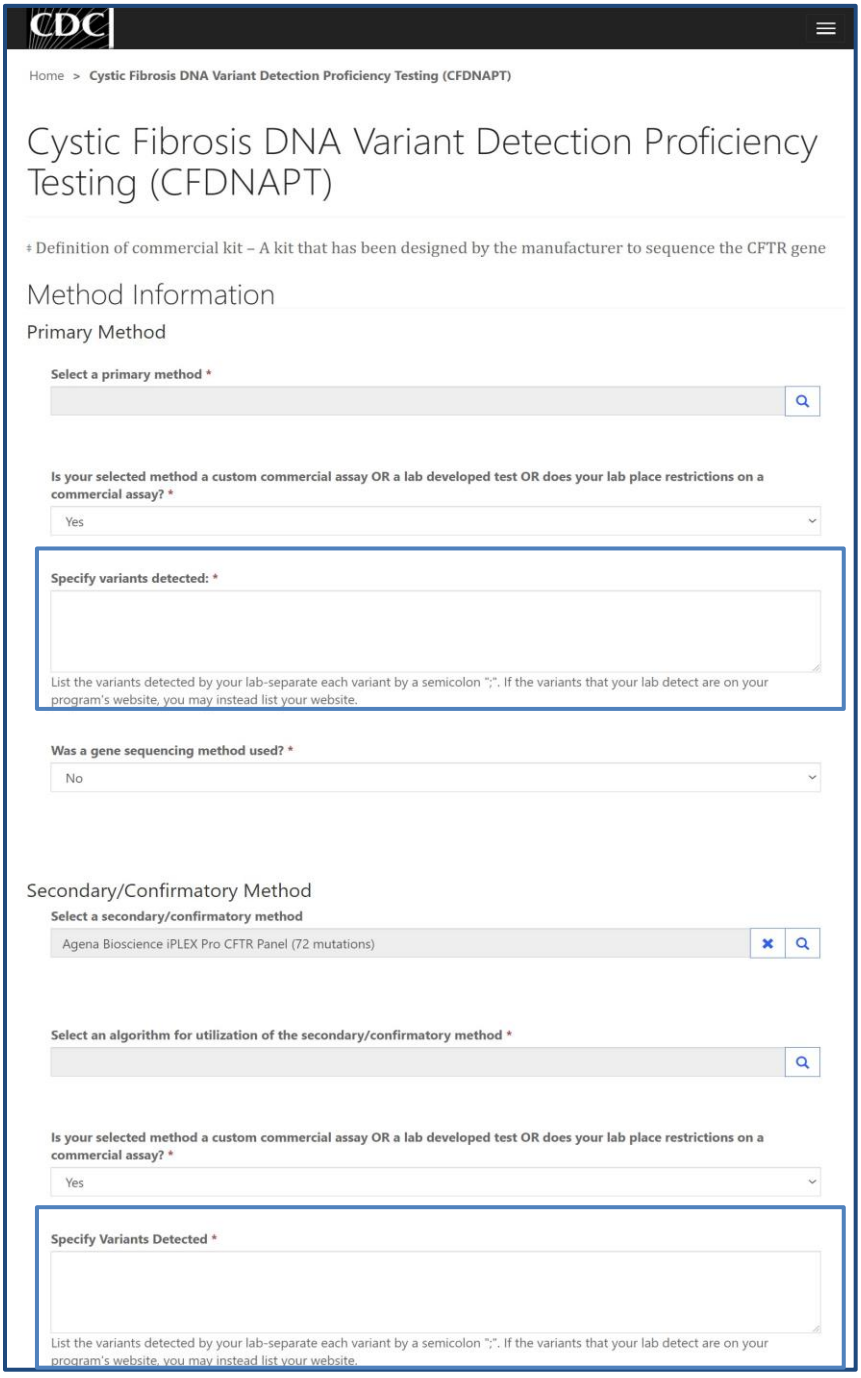
3. Continue to section 1.6 for guidance on reporting pathogenic variant data.



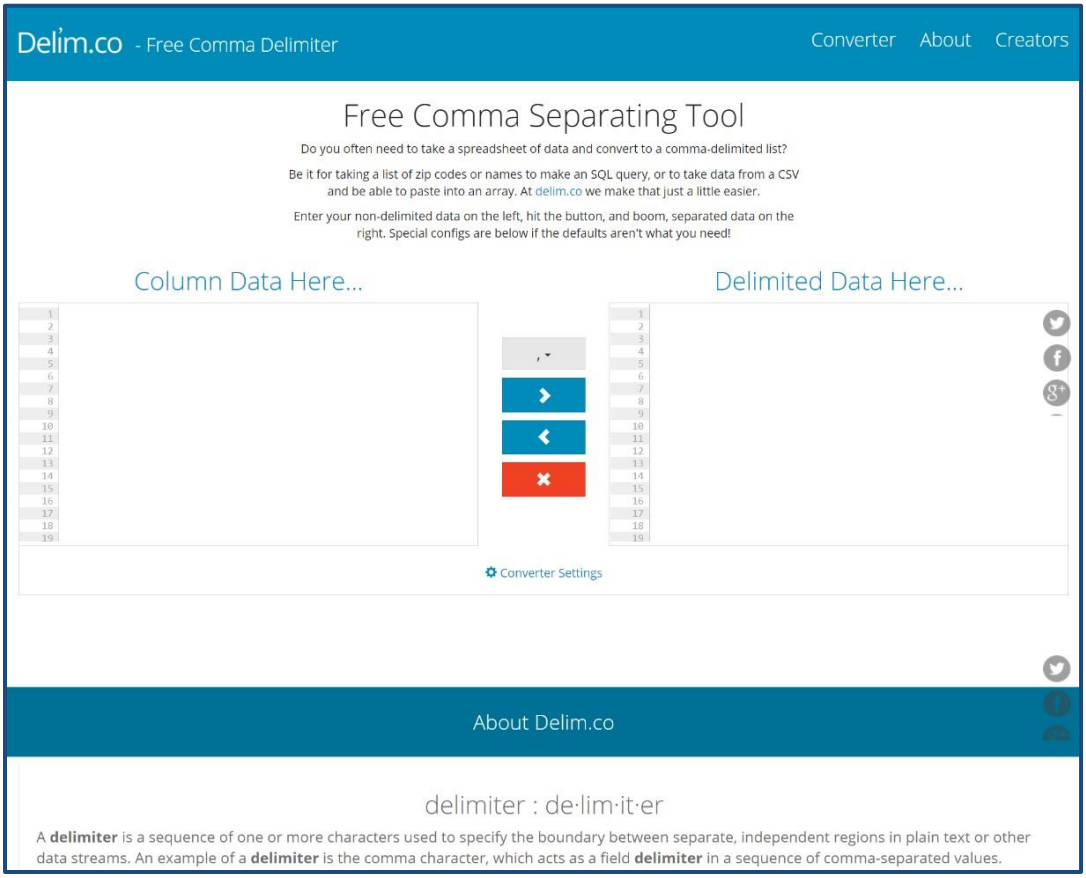
1.5 Specifying Variants Detected

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.

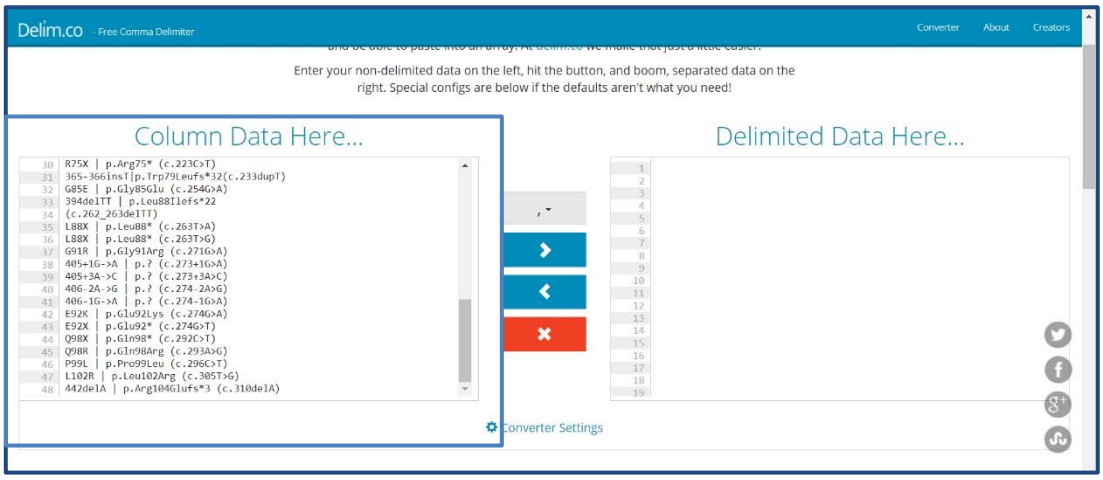
The variants detected by your lab should be listed in the indicated text boxes. **Format each variant by separating each with a semicolon ";".** Alternatively, if the variants that your lab detect are on your program's website, reference your website.



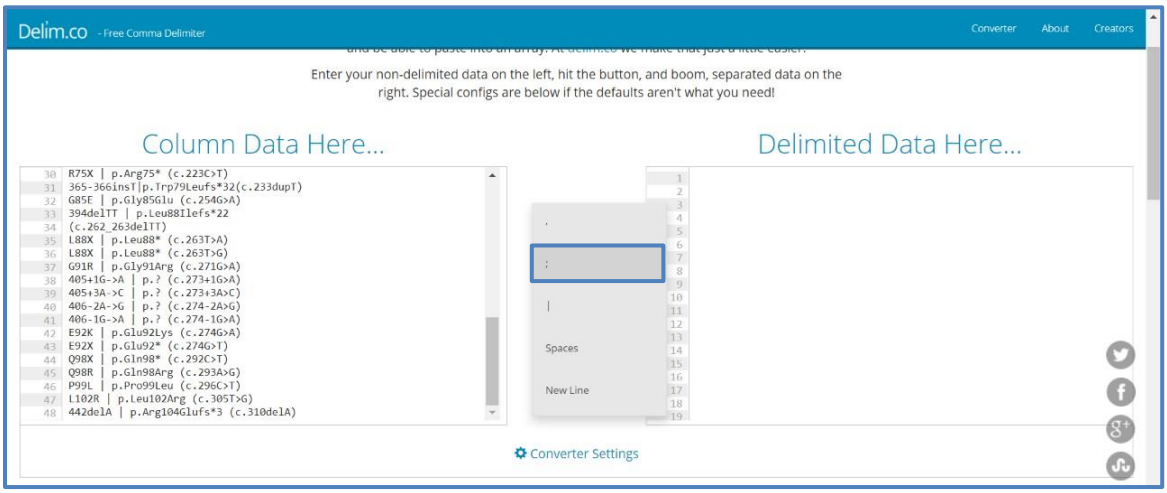
To assist with formatting the variant list, the delim.co website can be used as a tool to generate the formatted list. The tool can be found here.



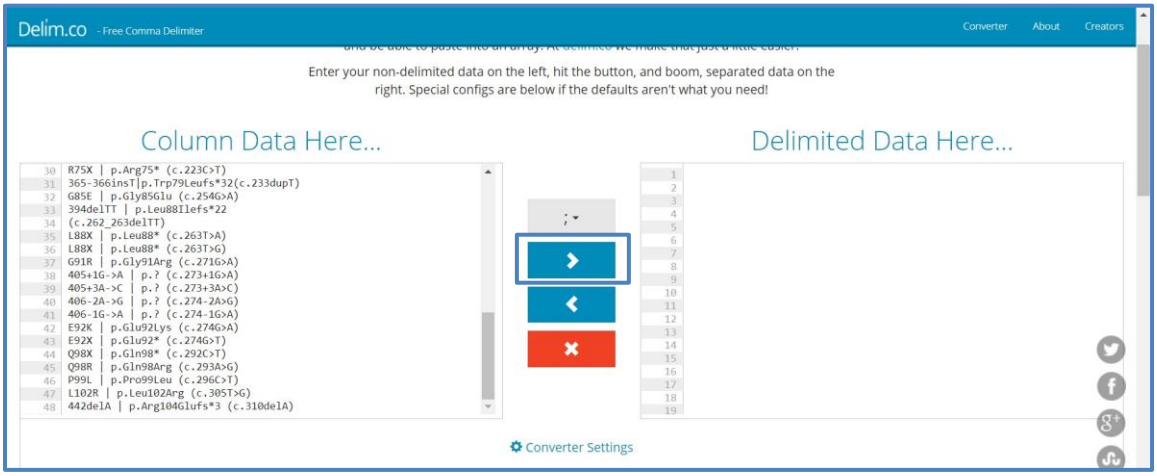
1. Paste the unformatted variant list in the box labeled ‘Column Data Here’.



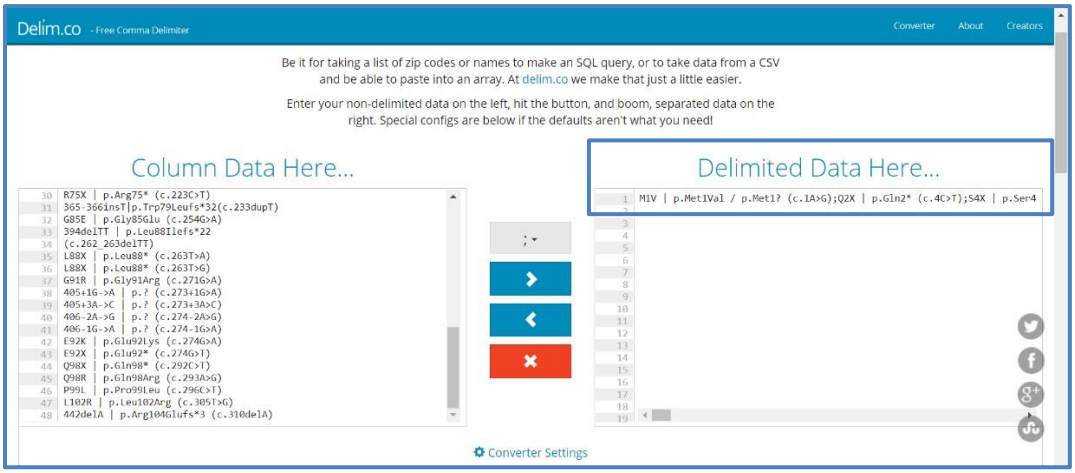
2. Select the semicolon option for the drop-down menu.



3. Select the blue right pointing arrow.

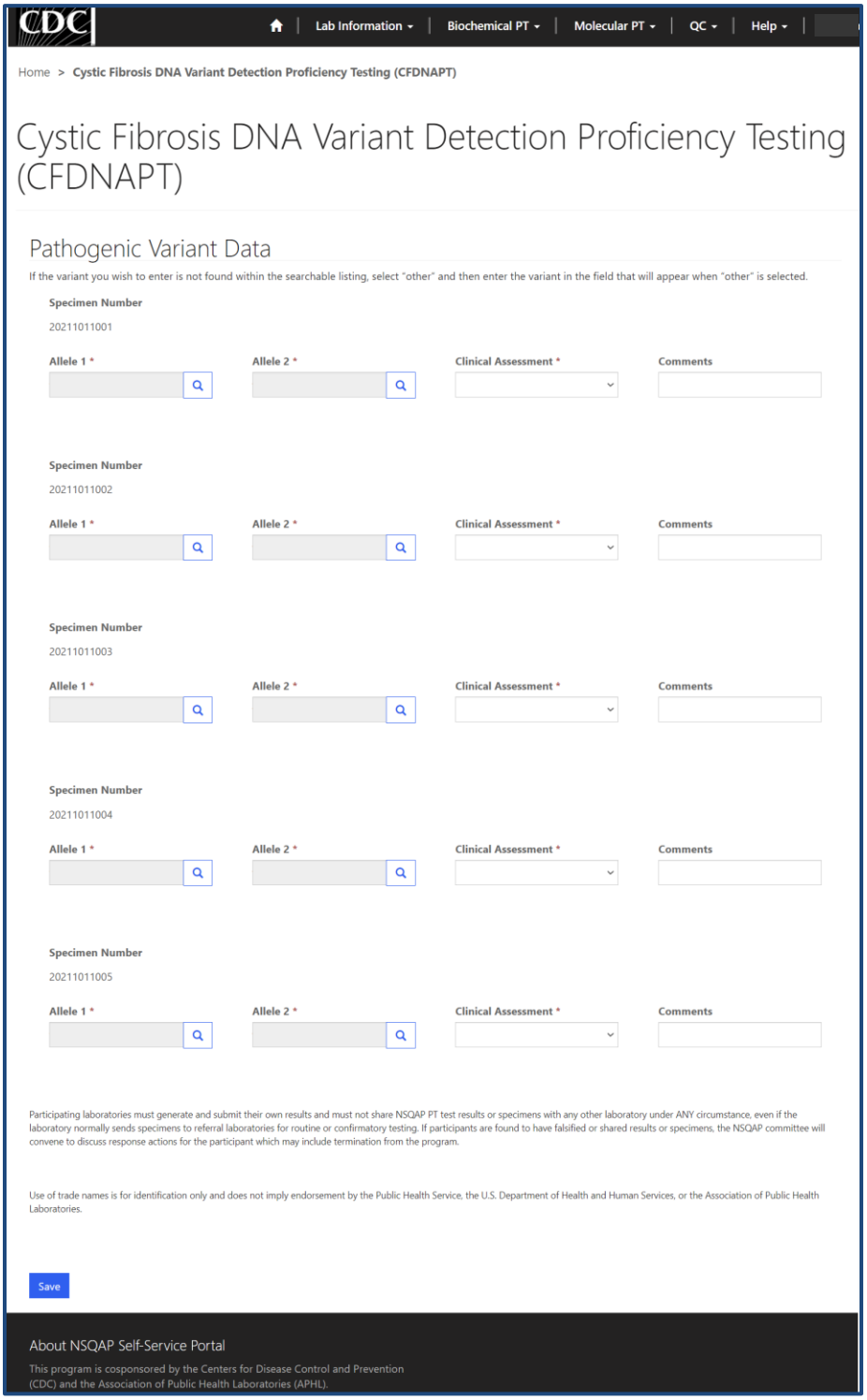


4. Copy formatted data and paste into the NSQAP CFDNAPT Portal page.

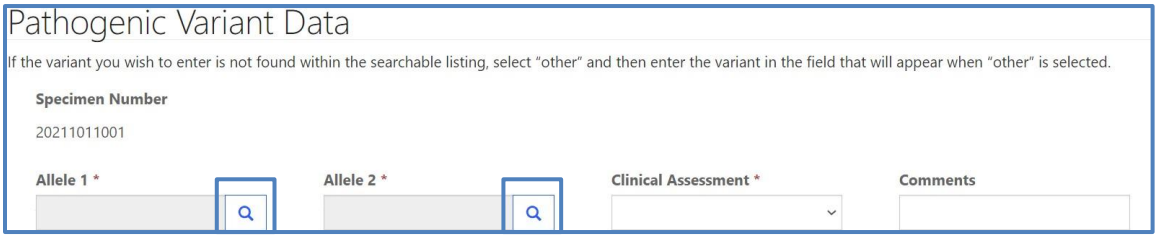


1.6 Specimen Results - Pathogenic Variant Data Entry

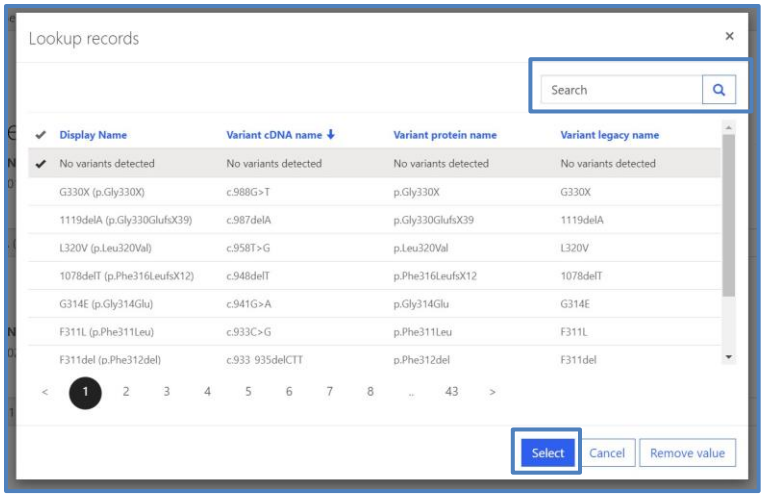
Navigate to the page titled 'Cystic Fibrosis DNA Variant Detection Proficiency Testing (CFDNAPT)' to enter CFDNAPT pathogenic variant results including allele 1, allele 2, and clinical assessment for each specimen. Navigation details can be found in section 1.1.

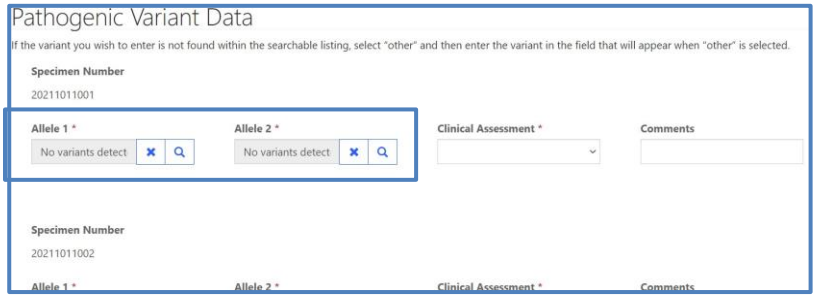


1. Click the magnifying glass to select a variant value for ‘Allele 1’ and ‘Allele 2’ for each specimen.

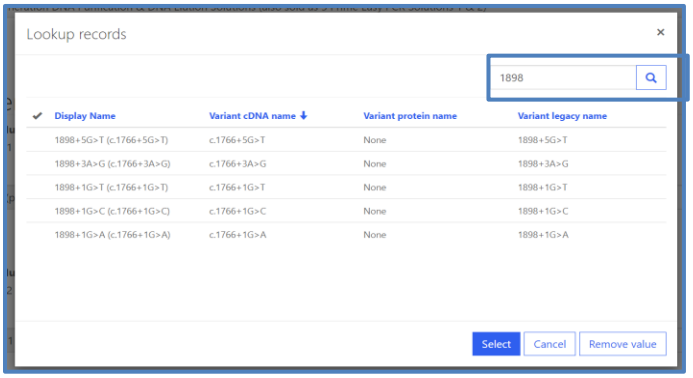


2. Search for variants using the search box. Click ‘Select’.

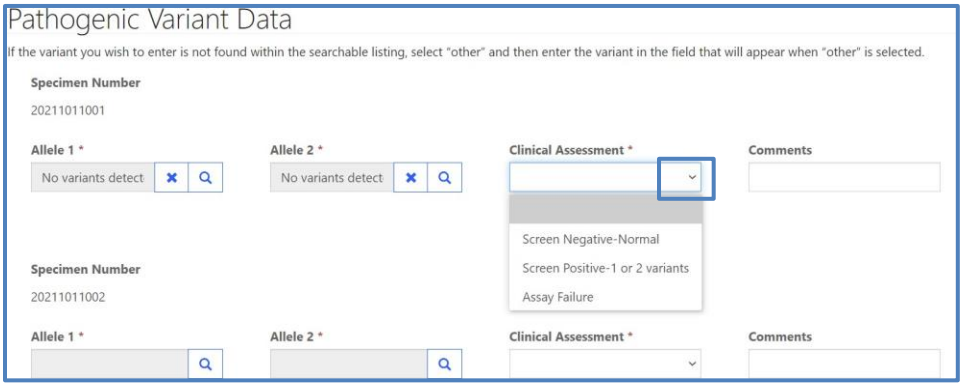




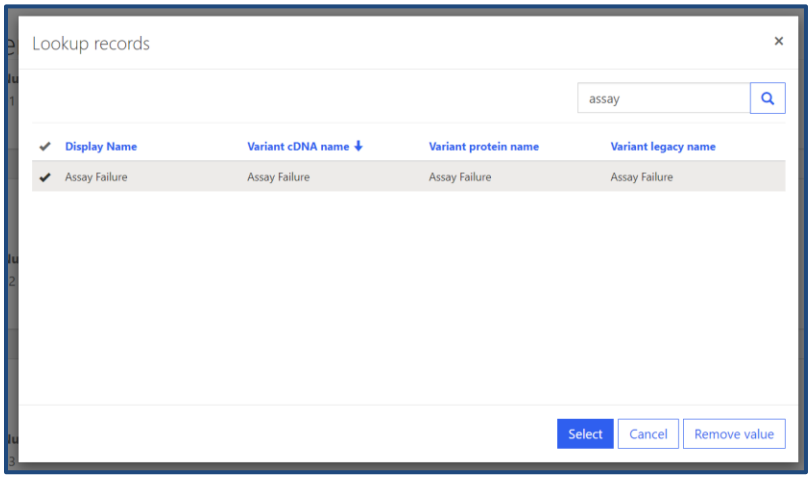
3. Variants can be quickly located by typing the variant name in the provided search box.

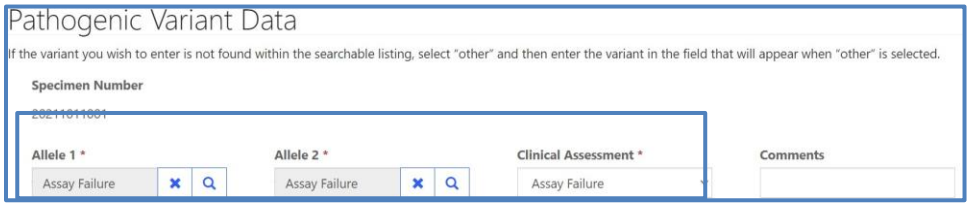


4. Choose a clinical assessment code for each specimen by clicking the drop-down arrow.

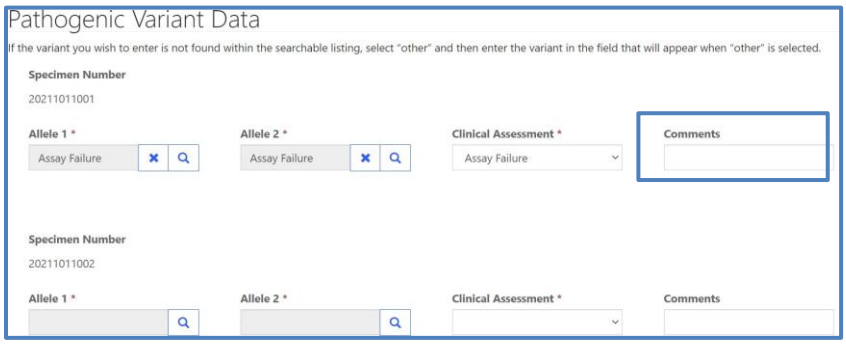


5. If 'Assay Failure' is chosen as the clinical assessment, choose it for both Allele 1 and Allele 2.





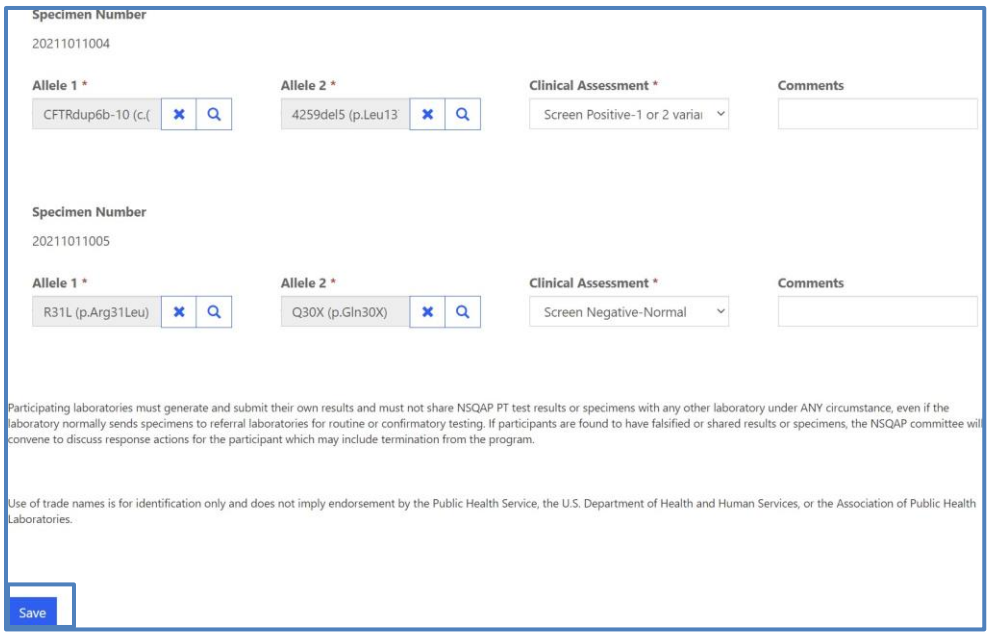
6. If necessary, enter any comments into the appropriate comment box.



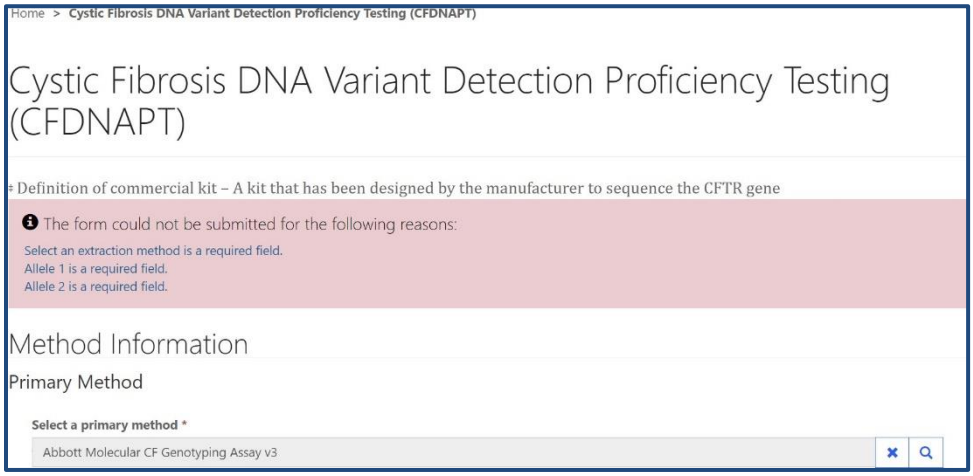
1.7 Save

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.



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.



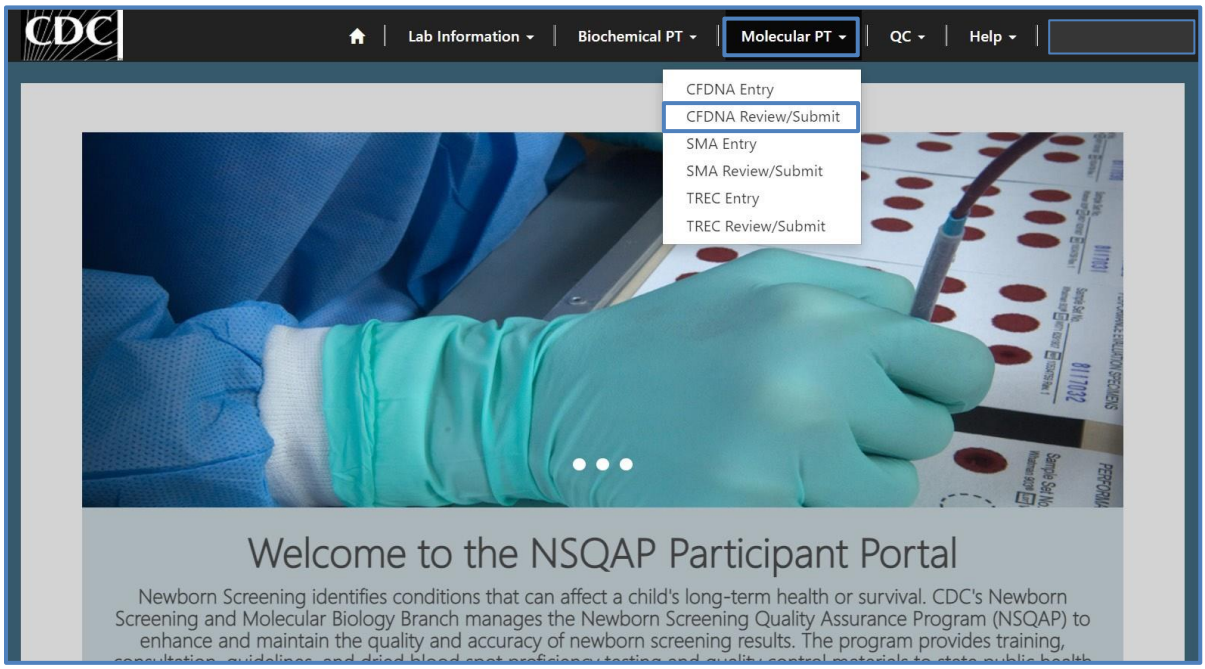
3. After you have successfully saved your results, you will be redirected to the 'CFDNA Review/Submit Page'. See section 2 for more guidance on this page.

2. CFDNAPT Review & Submit Page

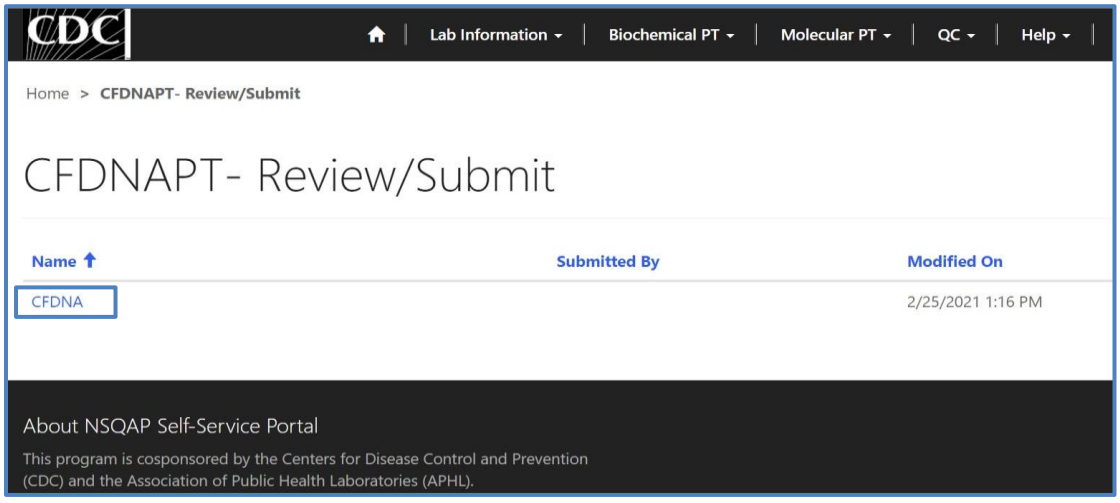
2.1 Navigation

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.

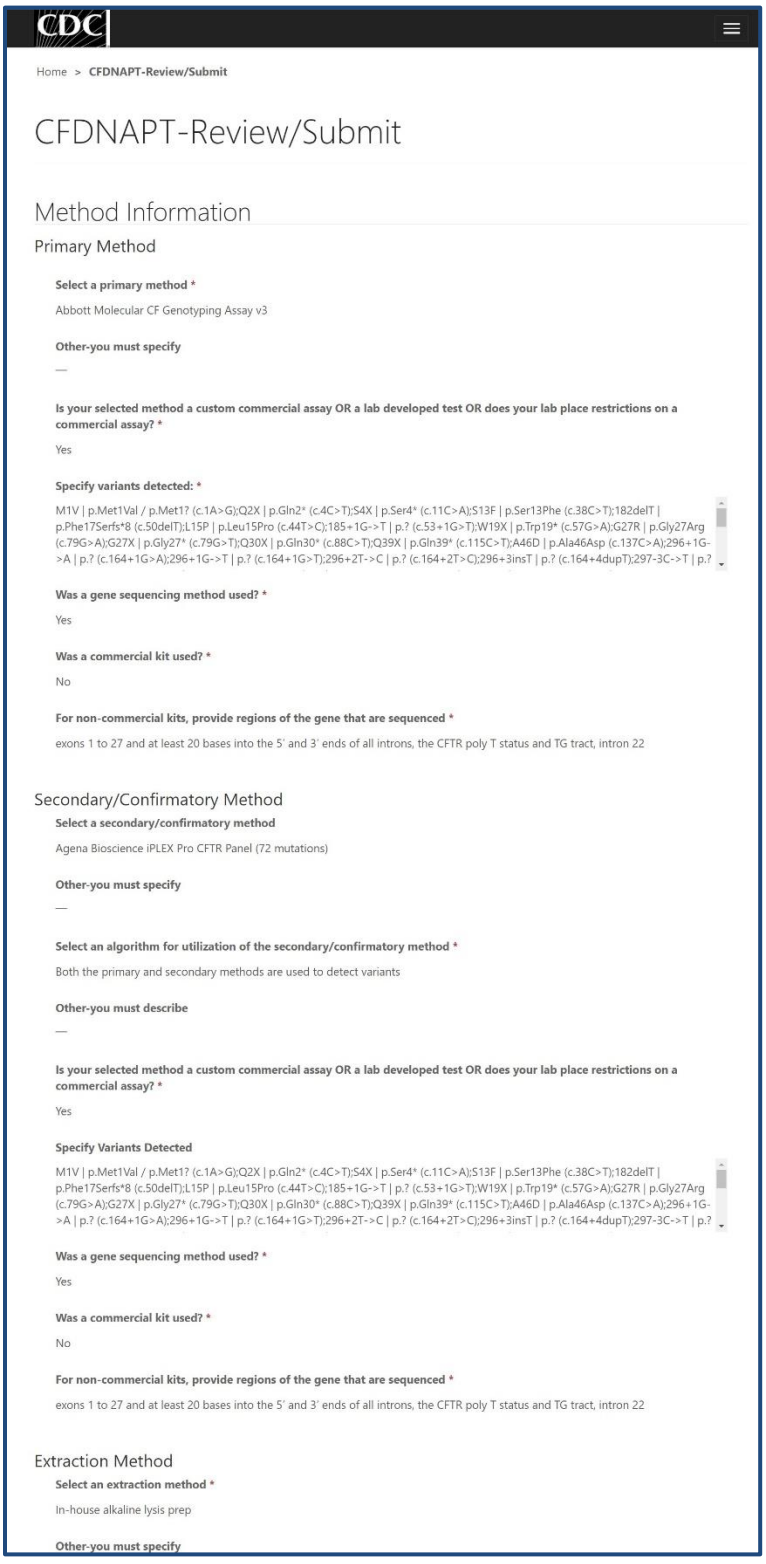


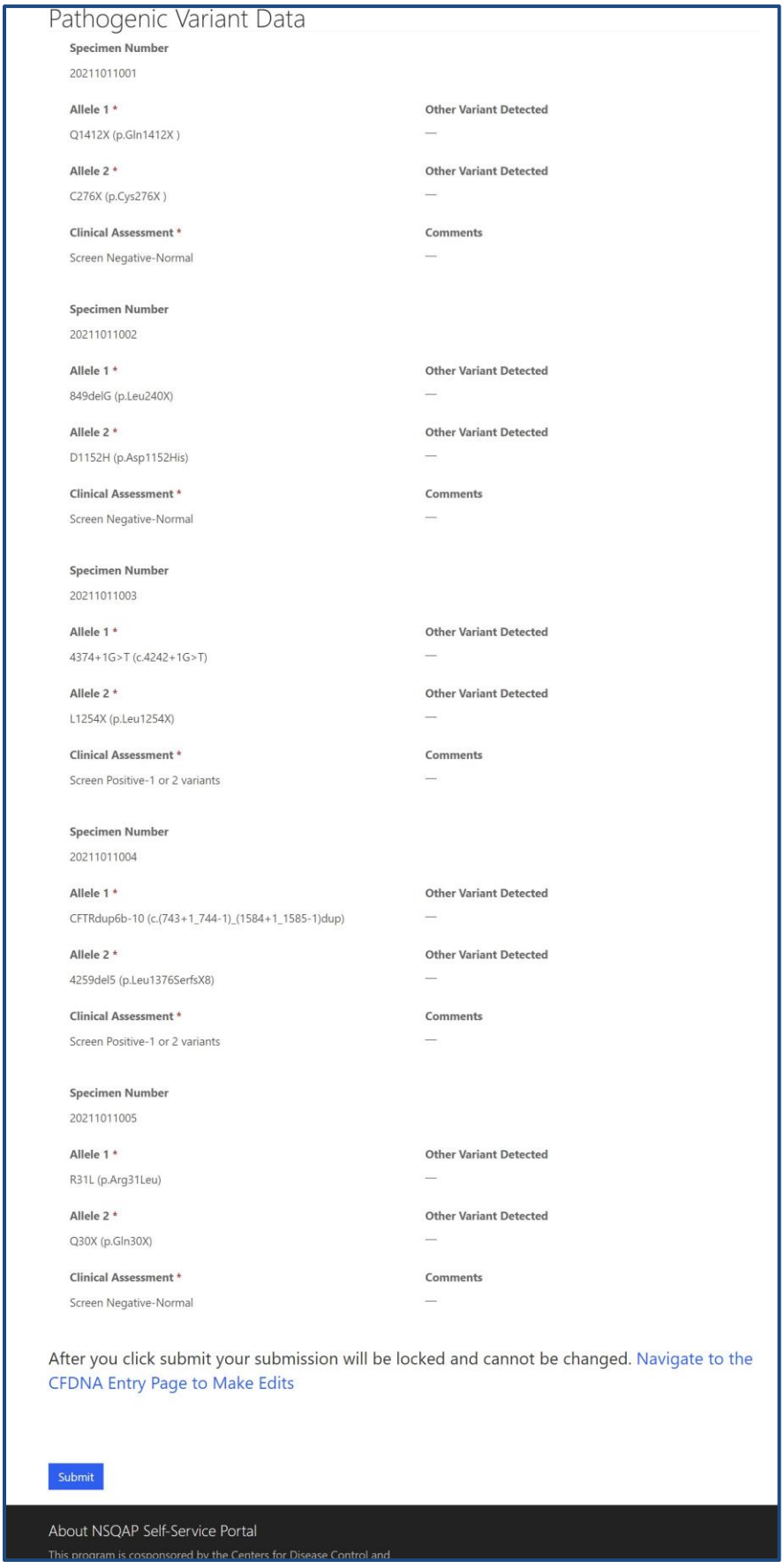
2. The CFDNA Review/Submit landing page will appear. Select **'CFDNA'** to navigate to the review and submit page.



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 *
C276X (p.Cys276X)

Other Variant Detected
—

Clinical Assessment *
Screen Negative-Normal

Comments
—

Specimen Number
20211011002

Allele 1 *
849delG (p.Leu240X)

Other Variant Detected
—

Allele 2 *
D1152H (p.Asp1152His)

Other Variant Detected
—

Clinical Assessment *
Screen Negative-Normal

Comments
—

Specimen Number
20211011003

Allele 1 *
4374+1G>T (c.4242+1G>T)

Other Variant Detected
—

Allele 2 *
L1254X (p.Leu1254X)

Other Variant Detected
—

Clinical Assessment *
Screen Positive-1 or 2 variants

Comments
—

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 *
Screen Positive-1 or 2 variants

Comments
—

Specimen Number
20211011005

Allele 1 *
R31L (p.Arg31Leu)

Other Variant Detected
—

Allele 2 *
Q30X (p.Gln30X)

Other Variant Detected
—

Clinical Assessment *
Screen Negative-Normal

Comments
—

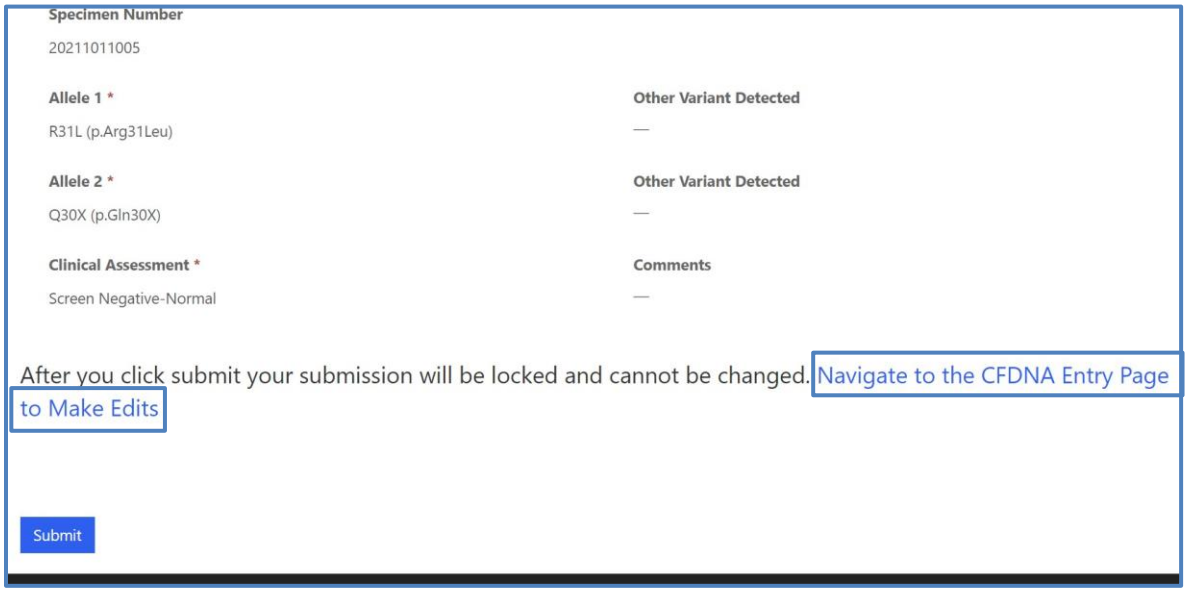
After you click submit your submission will be locked and cannot be changed. Navigate to the CFDNA Entry Page to Make Edits

Submit

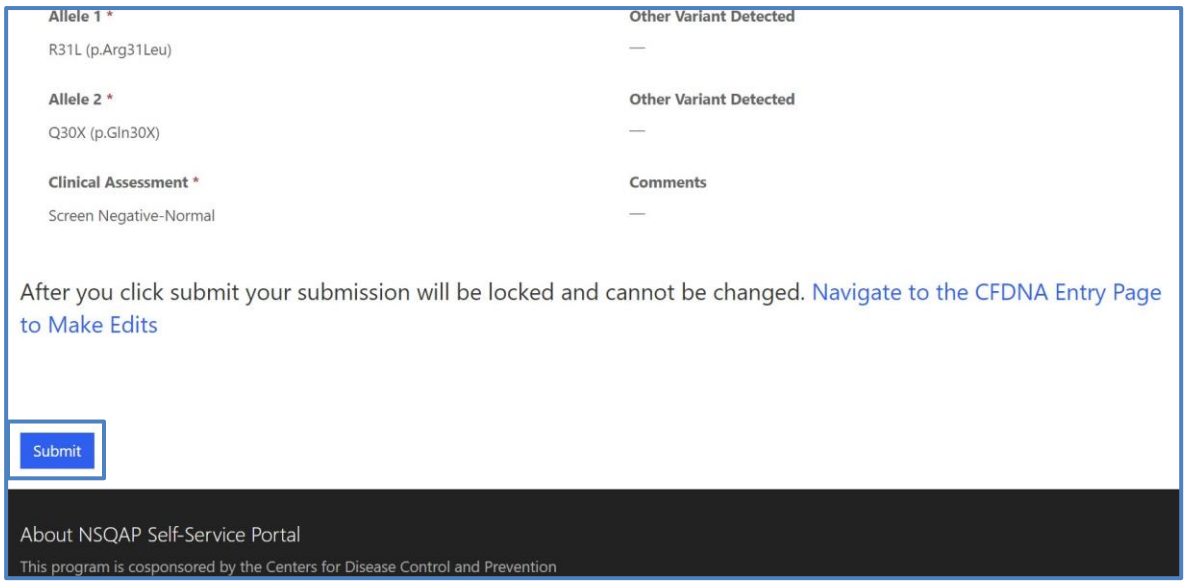
About NSQAP Self-Service Portal

This program is cosponsored by the Centers for Disease Control and

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'**.

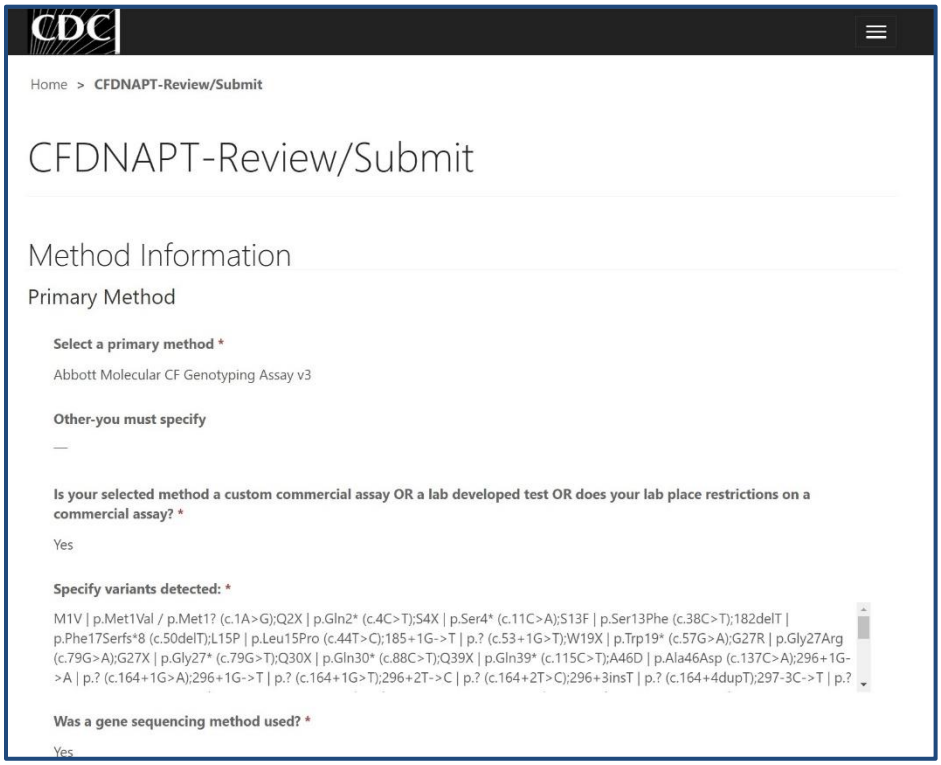


2. If no further edits are needed, results can be submitted by clicking the 'Submit' button. See section 2.3 for additional details.

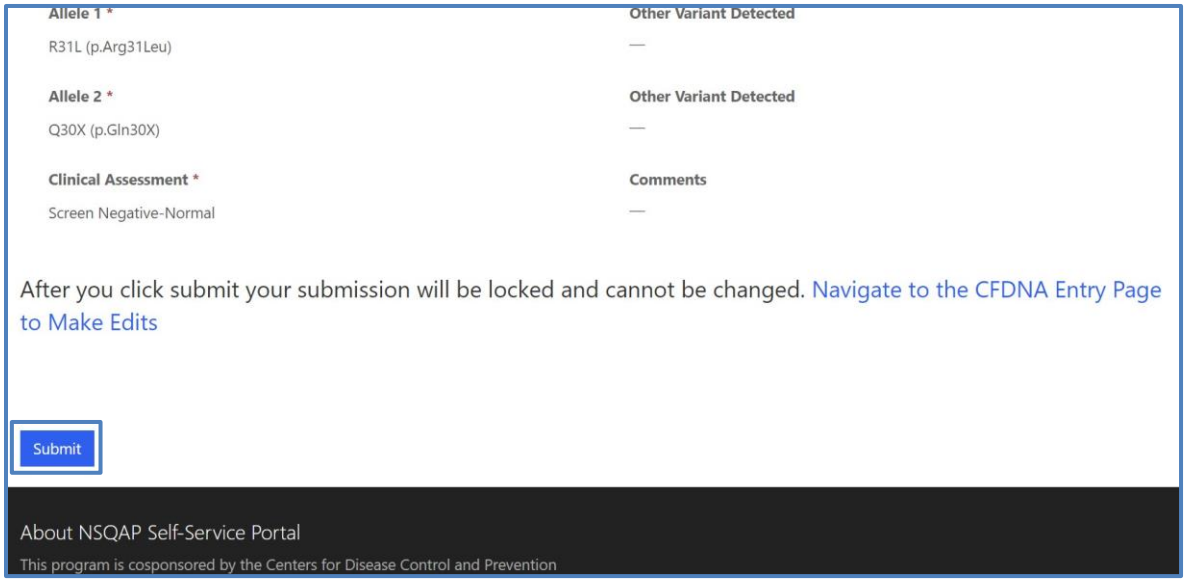


2.3 Submit

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

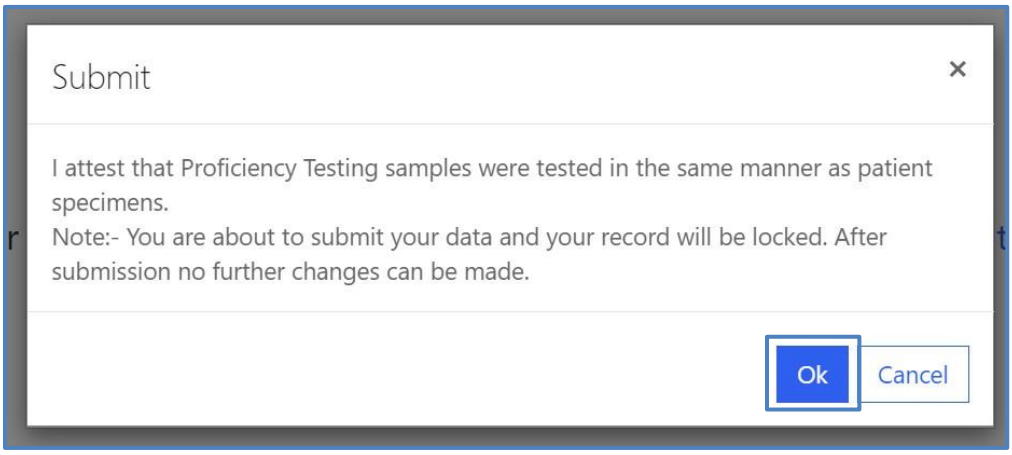


2. After reviewing the CFDNA review and submit page, click on the '**Submit**' button located at the bottom of the page.

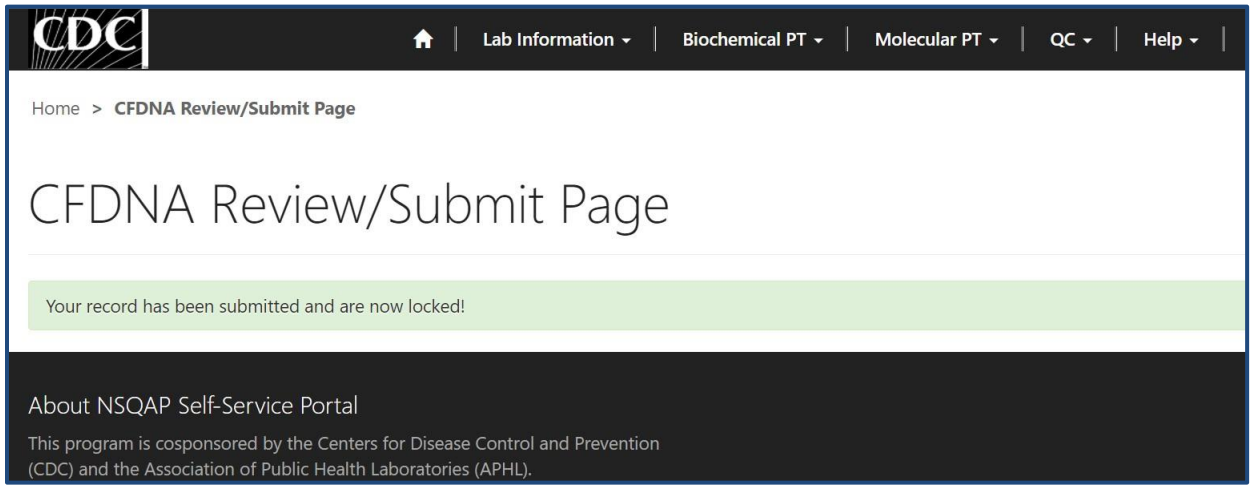


3. You will be prompted to confirm that you are ready to submit. Click **'Ok'** to confirm and submit your CFDNAPT results.

   NOTE: 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.



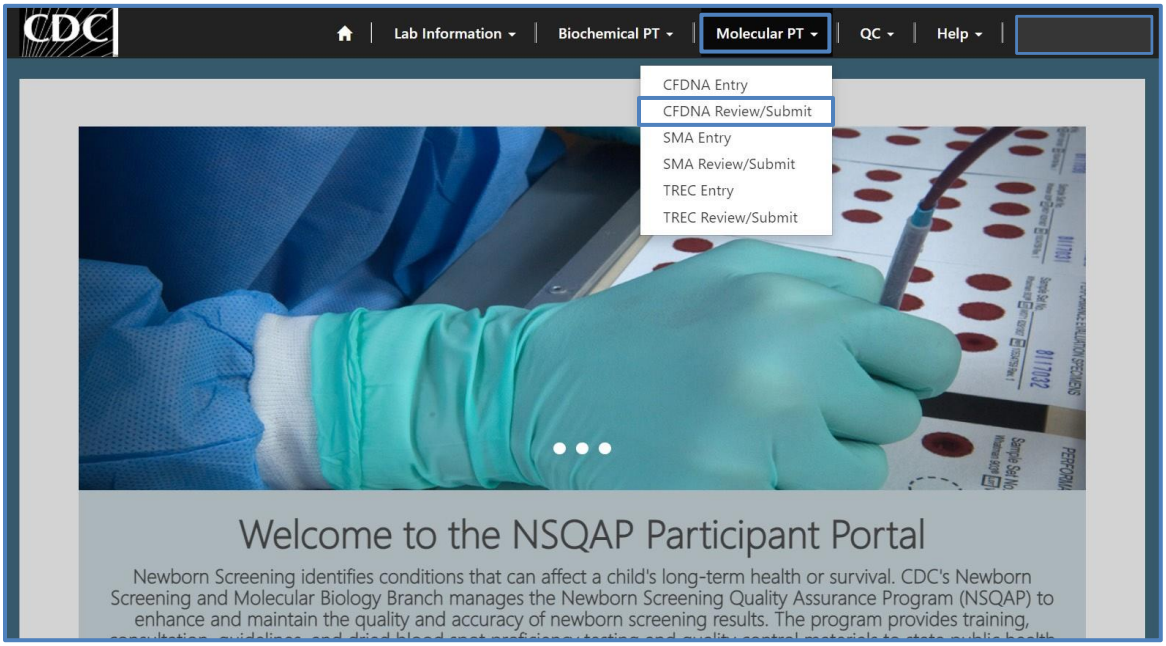
5. Once your CFDNAPT results are submitted you will no longer be able to access the 'CFDNA Entry' page. You can view your submitted data in a read-only format by accessing the review and submit page (see sections 2.1 and 2.2).

2.4 Save Data – Pdf Format

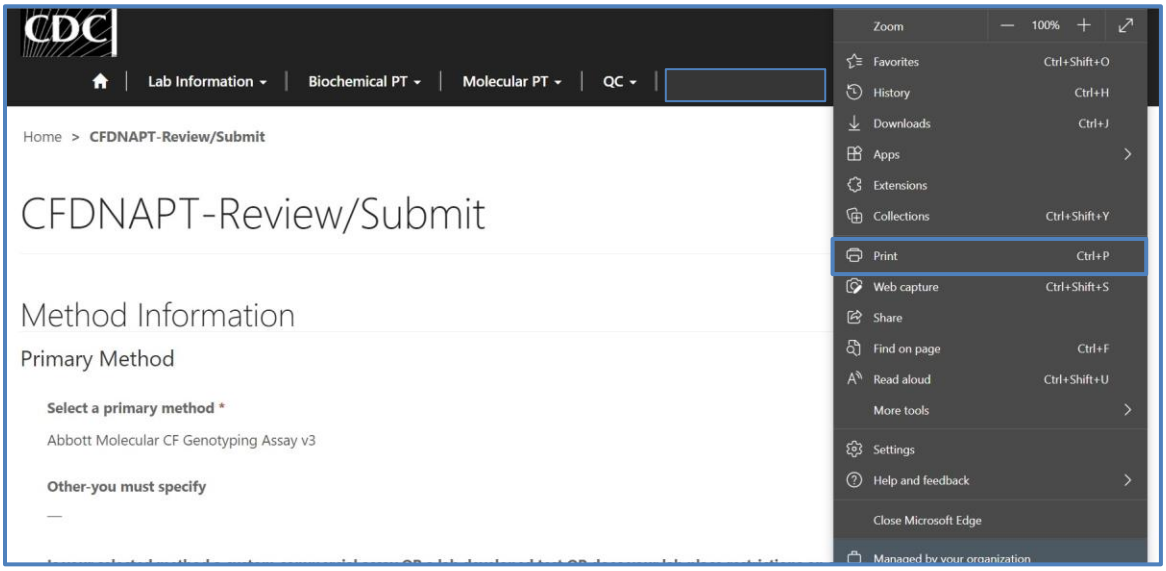
Submitted data can be saved in a pdf format by using the 'Save a PDF' function included in your web browser.

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

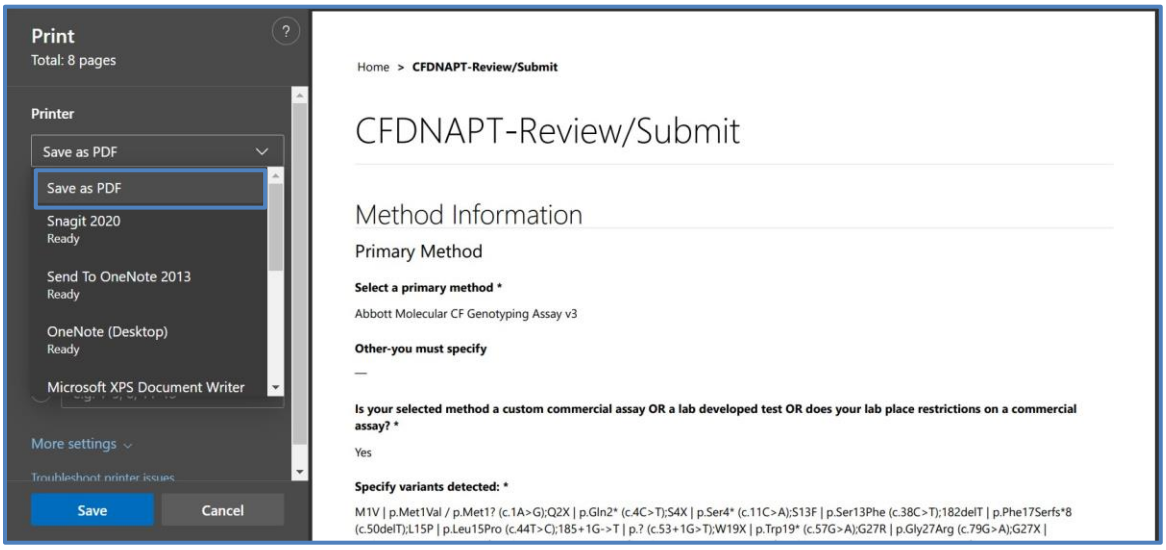
1. Navigate to the review and submit page as described in section 2.1.



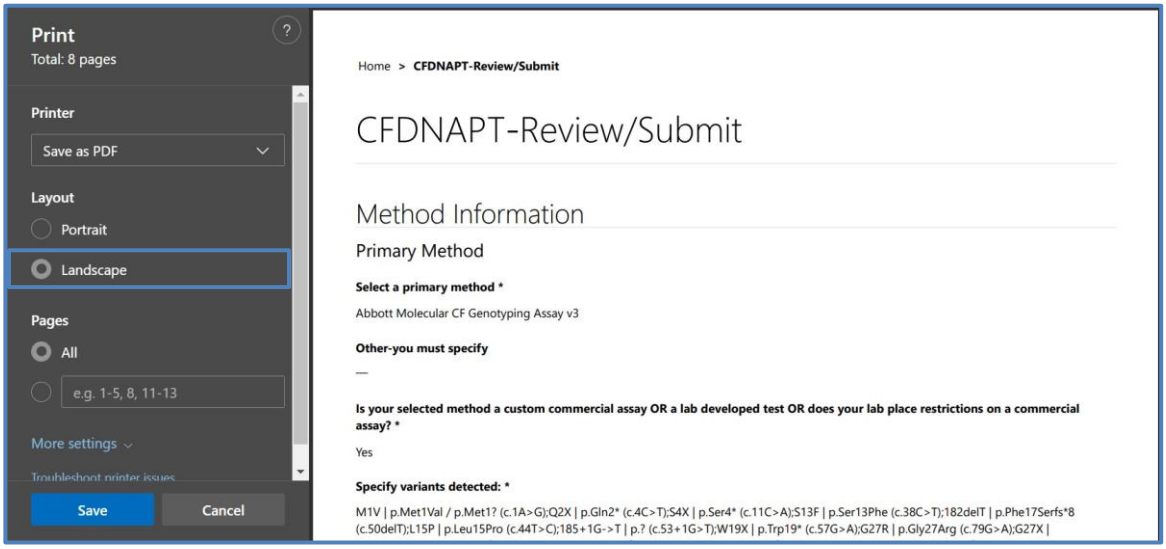
2. Locate the "Print' function on your web browser.



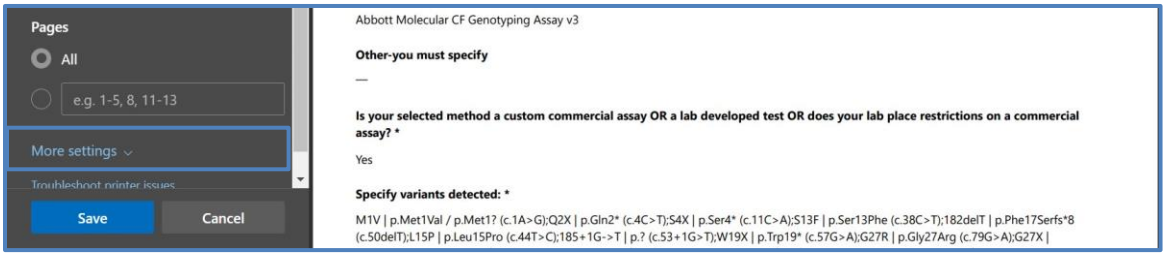
3. Select ‘Save as PDF’.



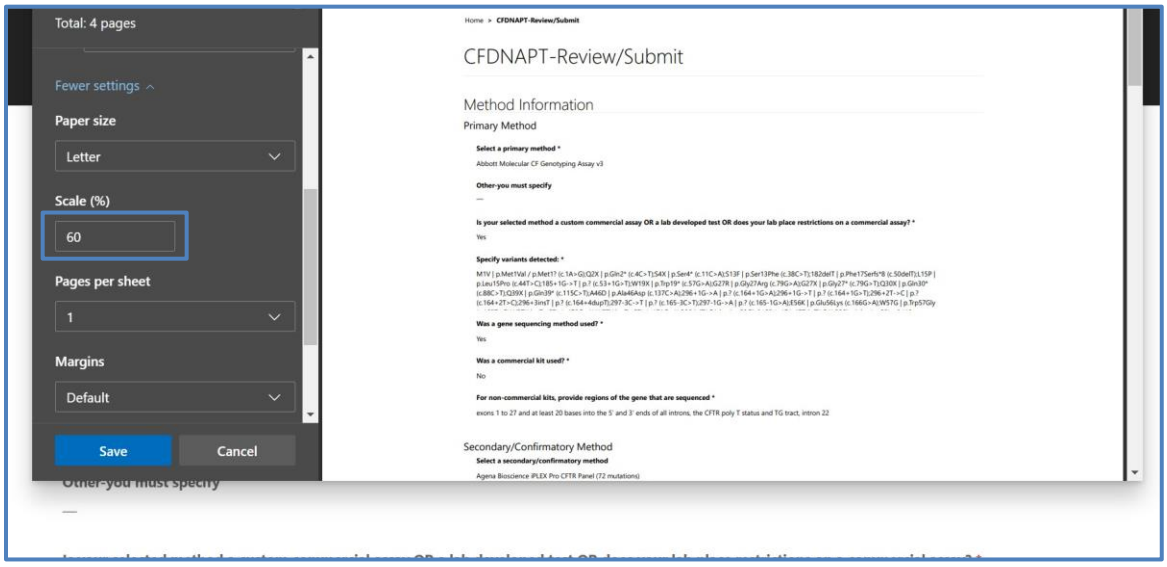
4. Select ‘Landscape’ as the layout choice.



5. Select ‘More Settings’.



6. Adjust the scale percentage to 60%.



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