




illumina Local Run Manager CF 139-Variant 2.0 Analysis Module Software User Guide

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Sample Information

Sample ID	Total PF Reads	Percent Q30 Bases
Sample1	3,415,516	95.43%

Amplicon Summary

Manifest	Number of Amplicon Regions	Total Length of Target Regions
rna_manifest	21,501	2,303,030 bp

The number of amplicon regions and total length do not include the expected off target regions that are included in the read and base level statistics.

Read Level Statistics

Read	Total Aligned Reads	Percent Aligned Reads
1	1,484,072	86.90%
2	1,484,072	86.90%

Base Level Statistics

Read	Percent Q30 Bases	Total Aligned Bases	Percent Aligned Bases	Percent Mismatches
1	96.83%	154,136,526	78.77%	0.18%
2	94.04%	154,132,280	78.40%	0.26%

The values for aligned bases exclude bases aligning to the probe sequences.

Coverage Summary

Amplicon Mean Coverage
134.1

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Local Run Manager CF 139-Variant 2.0 Analysis Module Workflow Guide

Overview

The Local Run Manager CF 139-Variant 2.0 analysis module is for use with TruSight Cystic Fibrosis 139-Variant Assay. The assay detects 139 clinically relevant cystic fibrosis disease-causing mutations and variants of the cystic fibrosis transmembrane conductance regulator (CFTR) gene in genomic DNA isolated from human peripheral whole blood specimens. The analysis module performs secondary analysis and report generation from sequencing runs that use TruSight Cystic Fibrosis. See the package insert TruSight Cystic Fibrosis Kit (Document # 1000000097720).

The analysis module evaluates short regions of amplified DNA, or amplicons, for variants. Focused sequencing of amplicons enables high coverage of particular regions across a large number of samples.

About This Guide

This guide provides instructions for setting up run parameters for sequencing and analysis for the CF 139-Variant 2.0 analysis module. Use of the software requires basic knowledge of the current Windows operating system and web browser-based user interface. For information about the Local Run Manager dashboard and system settings,

see the Local Run Manager Software Reference Guide for MiSeqDx (document # 1000000011880).

Enter Run Information

Set Parameters

1. Log in to Local Run Manager.
2. Select Create Run, and then select CF 139-Variant 2.0.
3. Enter a run name that identifies the run from sequencing through analysis.
Use alphanumeric characters, spaces, underscores, or dashes (40 characters or less).
4. [Optional] Enter a run description.
Use alphanumeric characters, spaces, underscores, or dashes (150 characters or less).
5. Enter the lot number and expiration date for the library prep kit.
Specify Samples for the Run
Specify samples for the run using one of the following options.
 - Enter samples manually—Use the blank table on the Create Run screen. Suggested sample wells are highlighted.
 - Import samples—Navigate to an external file in a comma-separated values (*.csv) format. A template is available for download on the Create Run screen.

Enter Samples Manually

1. Enter a unique sample name in the Sample Name field.
Use alphanumeric characters, dashes, or underscores (40 characters or less).
2. Right-click and select positive and negative control samples.
In order to save a run, it must have at least one positive and one negative control.
3. [Optional] Enter a sample description in the sample Description tab.
Use alphanumeric characters, dashes, or underscores (50 characters or less).
4. [Optional] Select an Index 1 adapter from the Index 1 (i7) drop-down list.
This step is optional because the i7 and i5 index combinations autopopulate with a default layout.
5. [Optional] Select an Index 2 adapter from the Index 2 (i5) drop-down list.
This step is optional because the i7 and i5 index combinations autopopulate with a default layout.
6. Select the Print icon to display the plate layout.
7. Select Print to print the plate layout as a reference for preparing libraries.
8. [Optional] Select Export to export sample information file.
9. Select Save Run.
If less than 24 samples have been entered, the Insufficient Sample window will display. Select Proceed to continue, or select Cancel to edit the samples.

CAUTION

Proceeding with less than 24 samples is not validated by Illumina. See TruSight Cystic Fibrosis Package Insert (Document # 1000000097720) for more information.

Import Samples

Sample information can be imported from two types of files:

- A sample information file that was previously exported from the CF 139-Variant 2.0 module using the Export feature.
- A template file, which can be generated by selecting Template on the Create Run screen. The template file contains the correct column headings for import, with placeholder information in each column. Use an external editor to customize the template file:
- Add sample information for each sample in the run.
- Once all sample information has been added, delete any remaining placeholder information in unused cells.
- Save template file.

To import sample information:

1. Select Import Samples, and then browse to the file and select it.
2. Select the Print icon to display the plate layout.
3. Select Print to print the plate layout as a reference for preparing libraries.
4. [Optional] Select Export to export sample information to an external file.
5. Select Save Run.

If less than 24 samples have been entered, the Insufficient Sample window will display. Select Proceed to continue, or select Cancel to edit the samples.

CAUTION

Proceeding with less than 24 samples is not validated by Illumina. See TruSight Cystic Fibrosis Package Insert (Document # 1000000097720) for more information.

Edit a Run

For instructions on editing the information in your run before sequencing, see Local Run Manager Software Reference Guide for MiSeqDx (document # 1000000011880).

Analysis Methods

For a description of the analysis methods for TruSight Cystic Fibrosis, see the package insert TruSight Cystic Fibrosis Kit (Document # 1000000097720).

View Run and Results

1. From the Local Run Manager main screen, click the run name.
2. From the Run Overview tab, review the sequencing run metrics.
3. [Optional] Select the Copy to Clipboard icon to copy the output run folder path.
4. Select the Sequencing Information tab to review run parameters and consumables information.
5. Select the Samples and Results tab to view the analysis results.

The tab provides a sample list that summarizes the call rate, performance, and information on controls.

If analysis was repeated, expand the Select Analysis drop-down list and select the appropriate analysis.

6. [Optional] Double-click the Comment field to enter a comment about a sample in the list. Click Save Changes.
7. Select samples in the list to display detailed variant information. Sample variant information displays below the sample list.

NOTE Each time changes are saved, an analysis results file will be generated in the Alignment folder. This file includes a time stamp appended to the file name, with the format YYMMDD_HHMMSS.

For more information on the Run Overview and Sequencing Information tabs, and how to requeue analysis, see

the Local Run Manager Software Reference Guide for MiSeqDx (document # 1000000011880).

Lot Tracking Data File

Once analysis is complete, choose one of the following options to view the LotTracking.txt file.

- From the Sample and Results tab, click Export Lot Tracking Data.
- Open the file in the Alignment folder.
The path to the Alignment folder is displayed on the Samples and Results tab in the Analysis Folder field.
- [Optional] Select the Copy to Clipboard icon to copy the analysis folder path.

When analysis is complete, the lot tracking data file is written to the Alignment folder for the run. For example: MiSeqAnalysis\<Run Folder Name>\Alignment_N\YYMMDD_HHMMSS. N is a sequential number that increases if the analysis was requeued. YYMMDD_HHMMSS is the timestamp of the run.

Results Report

After secondary analysis is complete, analysis results are summarized on the Samples and Results tab. An analysis results (output) file for the assay also summarizes results in a one tab-delimited text file called TruSightCF139VariantAssay.txt.

Results in the analysis results file include the information found on the Sample and Results tab. Choose one of the following options to view the analysis results file TruSightCF139VariantAssay.txt:

- From the Sample and Results tab, click Export Data.
- Open the file in the Alignment folder.
The path to the Alignment folder is displayed in the Analysis Folder field of the Samples and Results tab.

NOTE

Select the Copy to Clipboard icon to copy the analysis folder path.

The analysis files for each sequencing run are stored on instrument, with files saved to MiSeqAnalysis\<Run Folder Name>\Data\Intensities\BaseCalls and MiSeqAnalysis\<Run Folder Name>\Alignment_N\YYMMDD_HHMMSS. N is a sequential number that increments if the analysis was requeued. YYMMDD_HHMMSS is the timestamp of the run.

Analysis Result File Information

The analysis results file TruSightCF139VariantAssay.txt includes three sections: the file header, sample information, and sample variants information. The file header is listed first, and includes general information about the run. Note that there is only one file header per run. In contrast, sample information and sample variants information are included for every sample used in the analysis. These sections are paired together for each sample.

The following table provides a description of each row in the file header, which includes information about the run: Table 1 File Header (Run Information)

- Test Describes the test that was performed. Run ID Run ID that was generated by the MiSeq Operating Software (MOS) at the beginning of the sequencing run.
- Run Date Date (YYMMDD) that the sequencing run was started in MOS.
- Analysis Version Software version that was used for analysis.

The following table provides a description of each row in the sample information sections: Table 2 Sample Information

- **Sample ID** The sample name provided when the run was created, combined with the analysis ID assigned by the Local Run Manager software. If you requeue a run, the sample name part of the Sample ID stays the same, while the analysis ID changes.
- This field is in the *.txt file only.

Sample Name The sample name provided when the run was created.

Row Heading	Description
Control	The type of control as provided when the run was created. Values are positive or negative. A blank field indicates sample only.
Comment	An optional text field for comments. Comments from a previous analysis run do not carry over to the next analysis run.
Performance	Pass or Fail rating based on the call rate. For a positive control sample: <ul style="list-style-type: none"> • PASS — with a call rate \geq 99% • FAIL — with a call rate < 99% For a negative control sample: <ul style="list-style-type: none"> • PASS — with a call rate \leq 10% • FAIL — with a call rate > 10% For a sample not labeled as a positive or negative control: <ul style="list-style-type: none"> • PASS — with a call rate \geq 99% • FAIL — with a call rate < 99%
Sample Call Rate	The number of variant positions that meet a predefined confidence value threshold divided by the total variant positions interrogated. Call rate is described on a per-sample basis and reported as a percentage that is calculated as 1 minus [number of variant positions with incomplete calls divided by the total number of variant positions sequenced].

The following table provides a description of each column in the sample variants sections: Table 3 Sample Variants Information

Column Heading	Description
Mutations (Common Name)	Common name of the Cystic Fibrosis variant, as described in the CFTR2 database. The name of the field is Mutations (Common Name) in the *.txt file.
Mutation Type	The type of variant. <ul style="list-style-type: none"> • SNV — Single Nucleotide Variant • DIV — Deletion Insertion Variant • DEL — Large deletion. • PolyTGPolyT — PolyTG/PolyT genotype in CF gene
dbSNP rsID	The dbSNP rsID of the variant, if applicable.
CFTR Gene Region	CFTR gene region (exon # or intron #) where variant is present.
Genomic Location	Genomic location of the variant within chromosome 7.
cDNA Name (HGVS)	Description of the variant at the DNA level using the coding DNA (cDNA) sequence nomenclature as recommended by the Human Genome Variation Society (HGVS). The name of the field is cDNA Name (HGVS) in the *.txt file.
Protein Name (HGVS)	Description of a variant at the protein-level using the protein sequence nomenclature as recommended by the Human Genome Variation Society (HGVS). The name of the field is Protein Name (HGVS) in the *.txt file.
Result	Variant genotype. For SNVs, DIVs, and DELs: <ul style="list-style-type: none"> • HET — Heterozygous • HOM — Homozygous For PolyTGPolyT variant, the actual genotype is reported. NOTE: PolyTGPolyT is reported only when the R117H variant is detected.
Interpretation	This field is always left blank.

Revision History

- Document Date Description of Change
- Document # 1000000100945 v01 August 2021 Updated EU Authorized Representative address.
- Document # 1000000100945 v00 December 2019 Initial release.

Technical Assistance

For technical assistance, contact Illumina Technical Support.

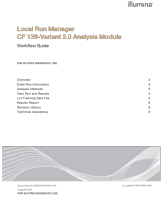
- Website: www.illumina.com
- Email: techsupport@illumina.com

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Australia	+1.800.775.688	
Austria	+43 800006249	+43 19286540
Belgium	+32 80077160	+32 34002973
China	400.066.5835	
Denmark	+45 80820183	+45 89871156
Finland	+358 800918363	+358 974790110
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Ireland	+353 1800936608	+353 016950506
Italy	+39 800985513	+39 236003759
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Netherlands	+31 8000222493	+31 207132960
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South Korea	+82 80 234 5300	
Spain	+34 911899417	+34 800300143
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Taiwan, China	00806651752	
United Kingdom	+44 8000126019	+44 2073057197
Other countries	+44.1799.534000	

Safety data sheets (SDSs)—Available on the Illumina website at support.illumina.com/sds.html. Product documentation—Available for download from support.illumina.com.

Documents / Resources

	<p>illumina Local Run Manager CF 139-Variant 2.0 Analysis Module Software [pdf] User Guide</p> <p>Local Run Manager CF 139-Variant 2.0 Analysis Module, Software, Local Run Manager CF 139-Variant 2.0 Analysis Module Software</p>
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References

- [Support Center](#)
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- [Safety Data Sheets \(SDS\)](#)
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