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Local Run Manager RNA Amplicon Analysis Module Release Notes
Part Number: 1000000048807, Ver. 02.1
Effective Date: 01-Sep-2022

Page **1** of **4**

Local Run Manager

RNA Amplicon Analysis Module v2.0.2

Release Notes

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Page **2** of **4**

Introduction

These Release Notes detail notable items for the Local Run Manager RNA Amplicon Analysis Module v2.0.2 release.

Please note that the RNA Amplicon Analysis Module v2.0.2 requires Local Run Manager Framework v2.0.0 or higher and is not compatible with Local Run Manager Framework v1.3.1 or lower.

For more information about this analysis module and how to use it, refer to the Local Run Manager RNA Amplicon Analysis Module Workflow Guide, available in the Documentation section of Local Run Manager Support Page.

http://support.illumina.com/sequencing/sequencing_software/local-run-manager/documentation.html

NEW FEATURES:

- Added ability to perform analysis with additional Advanced Settings (see table below) for Ampliseq Panel.

Setting Name	Value Options	Description
Clipping Penalty	The default value is 50.	BWA MEM clipping penalty.
Maximum Fusion Indel Size	Any value >=0 The default value is 100	Maximum indel size allowed in an alignment to a fusion target.
Short-read Length Threshold	Any value 1-35 The default value is 30.	The threshold determines whether an alignment is short.
Maximum Short-read Substitution Fraction	Any value 0-1 The default value is 0.1.	Maximum substitution fraction allowed for a short alignment.
Maximum Substitution Fraction	Any value 0-1 The default value is 0.04.	Maximum substitution fraction allowed for a non-short alignment.
Maximum Soft-clip Fraction	Any value 0-1 The default value is 0.1.	Maximum soft-clip fraction allowed for an alignment in the target region (excluding probes).
Minimum Mapping Quality	Any value >=0 The default value is 1.	Minimum mapping quality allowed for an alignment.
Require Both Reads Mapped	0 or 1 Default to 0.	Require that both read 1 and read 2 are mapped.
Minimum Singleton Coverage Fraction	When the Require Both Reads Mapped is • set to 0, any value 0 and 1. Default to 0.75. • set to 1.	Minimum target coverage fraction required for a singleton alignment.

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Local Run Manager RNA Amplicon Analysis Module Release Notes
Part Number: 1000000048807, Ver. 02.1
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Page **3** of **4**

DEFECT REPAIRS:

- Fixed "Analysis aborts early with error" issue when Resources requested are greater than maximum allowed in iSeq.

- Fixed "Aggregated Large Log files causing secondary analysis failure" in MiSeq

KNOWN ISSUES:

- Display: Gene Normalization Dropdown list is truncated.

PREVIOUS RELEASE NOTES

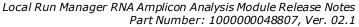## RNA AMPLICON ANALYSIS MODULE V2.0.1

NEW FEATURES:

- Added ability to perform analysis of External RNA Controls Consortium (ERCC) spike-in RNAs as part of settings on user interface.
  - ERCC targets are part of the reporting files output.
  - Merged manifests are now always encrypted with ERCC targets defined in the manifest.

- Lowered the detection threshold for RNA fusion events.

- Added the option to enable Gene Based Normalization. (option is disabled by default)

- Now, the output for RNA fusions are tabix-indexed VCF and CSV files and exons are skipped.

- Added a column of "Number of detected exon variants" to the summary CSV file.

DEFECT REPAIRS:

- Fixed "array index out of bound" error when writing fusion output files.

- Fixed intermitted issue with the report links.

- Fixed app crashing when the first segment of a fusion target aligns to the reference with a deletion or skipped region.

- Ensure that there are two entries present in fusions.vcf for each entry in fusions.csv.

- Overlapping target regions are now merged to calculate total length.

KNOWN ISSUES:

- Analysis for TruSeq Targeted RNA Expression Library Prep Kit are not supported when used with the associated panel manifests. List:
  https://support.illumina.com/downloads/truseq-targetd-rna-expression-manifest-files.html

Local Run Manager RNA Amplicon Analysis Module Release Notes
Part Number: 1000000048807, Ver. 02.1
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Page **4** of **4**

- Display: Index Column Dropdown Arrow Missing for TruSeq Targeted RNA Expression Kit

- Display: Library Prep Kit dropdown does not scale in size to match longer library prep kit names.

- Display: Report footer information contains the RNA Amplicon Analysis Worker version instead of the RNA Amplicon Local Run Manager module version.

- In rare instances, the Sample Sheet file generated during analysis may not reflect the same ordering of samples as what was entered on the "Run Setup" page.

- Report incorrectly lists PUMA Metrics version, which was replaced with bammetrics