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DRAGEN v3.4.12 Software Release Notes



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Introduction

These release notes detail the key changes to software components for the Illumina® DRAGEN $^{\text{TM}}$ Bio-IT Platform v3.4.12 since the major release containing DRAGEN $^{\text{TM}}$ v3.4.5

If you are upgrading from a version prior to DRAGEN $^{\text{TM}}$ v3.4.5, please review the release notes for DRAGEN $^{\text{TM}}$ v3.4.5 for a list of features and bug fixes introduced in that version.

DRAGEN™ v3.4.x Installers and Release Notes are available here: https://support.illumina.com/downloads/illumina-dragen-v3-4-installers.html

The 3.4.12 software package includes:

- DRAGEN™ SW Intel Centos 6 dragen-3.4.12.el6.x86_64
- DRAGEN™ SW Intel Centos 7 dragen-3.4.12.el7.x86_64
- DRAGEN™ SW IBM PPC Centos 7 dragen-3.4.12.el7.ppc64le.run

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Highlights

DRAGEN™ v3.4.12

The v3.4.12 patch release contains the following changes:

Improvements

- o VC: Added support to generate both VCF and gVCF in the same analysis run
 - VCF and gVCF now produce the same calls
 - New option --vc-enable-vcf-output can be used to enable VCF output in a qVCF run
- VC: Improved FRD/BQD algorithm
 - Improved accuracy in gVCF mode, and helps in generating the same calls in VCF and gVCF
- Metrics: Include mapq=0 reads in coverage reports counting, when mapq<0 filter is used

Bug fixes

- Alignment output
 - Fix for a bug in the generation of NM tags in the alignment output
- Joint Caller
 - Denovo post-filter vc metrics were not updated correctly. This issue was introduced in v3.4.5 and fixed here.
 - Do not update GT based on pedigree for indels
 - Sex chromosome GT fields were incorrectly updated based on pedigree, in cases where the sample ploidy is not known.
 - A crash in Joint Caller when input FORMAT/DP field is not valid
- Duplicate marking
 - Add duplicate marking to unmapped reads. DRAGEN previously never marked unmapped reads as duplicates
 - Non-properly paired reads now mark the expected pair as a duplicate. Mark unmapped mates of duplicates as duplicates, for half-mapped pairs.
- o Fix for a rare crash in SV metric reporting
- AWS: Fix for an issue where the FPGA DRAM gets corrupted after several hundred back-to-back analysis runs on a single F1 instance, leading to invalid output and/or crashes.

DRAGEN™ v3.4.9

The v3.4.9 patch release addresses the following issues present in v3.4.5:

Stability improvements

- Fix for a rare intermittent hang during mapping, seen on Dell R740 servers only.
 This was due to a corruption in the FPGA DRAM that may happen during HW reset.
- Fix for a rare intermittent crash during partial reconfiguration of the FPGA, seen on Dell R4740 servers only. This was due to a race condition when dragen_lic concurrently access the card while the FPGA is reconfigured.
- Fix for a rare intermittent hang during RNA mapping.
- Fix for a rare intermittent crash in germline mode with CRAM output.
 - Fix for an intermittent run-to-run variation on the Joint Caller output on chrX.

Bug fixes

- Fix for a bug in Joint Calling for pedigrees containing multiple trios: The positions
 of the sibling were GT corrected even though the individual GT did not show a
 mendelian conflict.
- Fix for a bug in Joint Calling for pedigrees with trios: GT was updated even when the denovo quality score (DQ) is below the filter threshold.



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- o Fix for crashing when enabling CRAM output for Somatic T/N runs.
- o Fix for failing to auto detect the stranded (ISR) library-type in RNA quantification.
- o Fix for crashing during BCL conversion of 384-plex, 8 lane, unique-duals inputs.
- Fix for a missed deletion on the mitochondrial contig in the structural variant caller.
- Fix to the calculation of GC bias, by skipping decoy and alt contigs.

SW Installation Procedure

- Install the appropriate release based on your Linux OS with the command: sudo sh <dragen 3.4.12 .run file>
- Cold boot (hard reset or power cycle) may be required after installation. An updated FPGA
 image base needs to load from flash, this is only achieved with cold boot.
 - Cold boot is required when upgrading from a release prior or up to v3.3.7
 - Cold boot is **not** required when upgrading from a release after v 3.3.7
 - Cold boot is **not** required when upgrading from v3.4.5 or v3.4.9
- Installation of prior releases, after v3.4.x was installed:
 - Installation of a prior release v3.3.7 or older, will require the following three steps. The prior .mcs file needs to be flashed manually:
 - Install the prior release: sudo sh <dragen 3.3.7 .run file>
 - program flash /opt/edico/bitstream/07*/*.mcs
 - Power cycle
 - Installation of prior release v3.4.5 or v3.4.9, or any prior version after v3.3.7:
 - Run: sudo sh <dragen 3.x.x .run file>

md5checksum:

fb453c72ff992bba6c47f7cb35d641e4 dragen-3.4.12.el7.x86_64.run 639e8af047dc28603668faedf3857c5f dragen-3.4.12.el6.x86_64.run 1789b0ac4a988d85813e69c4f77d2561 dragen-3.4.12.el7.ppc64le.run