

Universal Analysis Software v2.5.0_20220811.201258Z

Release Notes

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Introduction

These release notes describe changes to components of the Universal Analysis Software v2.5.0_20220811.201258Z and known issues. If you are updating from a version prior to 2.4.20211.013.235212Z, review the release notes for previous version for information on the changes introduced in that release. See the applicable guides for instructions on using the software.

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New Features

The following features are added across all ForenSeq kits:

- Added global feature to select and bookmark samples across all ForenSeq kits
- Enabled high level data summary across all ForenSeq kits
- Added new categories, sorting and filtering capabilities for all ForenSeq kits
- Updated main navigation rail to include universal search and support access to all Projects, Runs, and Samples across all ForenSeq kits
- Added new bulk download feature to enable download of multiple sample reports simultaneously across all ForenSeq kits
- Added ability to create custom project reports and individual reports that automatically download and zip from the Sample List page, across all ForenSeq kits.
- Added ability to copy samples or easily create a new project to move samples from project to project.

The following features are added for the ForenSeq Imagen Kit:

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- Two assay types and their Analysis methods
 - -ForenSeq Imagen assay and ForenSeq Imagen Analysis Method to support phenotypic estimations
 - -ForenSeq Imagen Geo assay and ForenSeq Imagen Geo Analysis Method to support phenotypic and biogeographic ancestry estimations

The following features are added for the ForenSeq MainstAY SE kit:

- Assay type with its Analysis Method
 - ForenSeq MainstAY SE assay and ForenSeq MainstAY SE Analysis Method

The following features are added for the ForenSeq Kintelligence kit:

- The name for the Phenotypic SNP (pSNP) N29insA have been updated to rs312262906 in the user interface and the relevant UAS reports
- SNPs previously called using the reverse strand have been switched to the forward strand in the user interface and relevant reports, in order to be compliant with NCBI changes to dbSNP and HIRISplexS
 - No changes were made to the GEDmatch PRO reports that already only use forward strand nomenclature and no changes are expected for GEDmatch PRO match results.
 - List of SNPs impacted are tabulated in Table 1 in Appendix A
- Updated the quality flags for the positive amplification control (NA24385) and negative amplification controls as below
 - Positive amplification control is orange with less than 10185 correct SNP loci calls
 - Negative amplification control is orange with more than 21 SNP loci calls

Defect Repairs

The following bug fixes and repairs were introduced for ForenSeq mtDNA kits:

- Updated EMPOP report from the mtDNA Whole Genome analysis for position 3107 previously reported as a deletion.
- Updated EMPOP report for no coverage positions from mtDNA Whole Genome analysis, previously reported as "N", to enable EMPOP compatibility.
- Updated variant calling to be consistent with SWGDAM Interpretation Guidelines for Mitochondrial DNA Analysis by Forensic DNA Testing Laboratories –includes realignment of 16189del and 310del
- Updated the NUMT (nuclear mtDNA insertions) filter to reduce possibility of filtering actual calls.
- Updated Trimming rules
 - Updated conditions for checking the presence of upstream and downstream primers.

- Added trimming rule (during primer trimming step) in position 1490 and 8442 due to higher error rate in some samples.
- Updated alignment adjustment around 'T' in 303-315 and 16184-16193 C-stretches and to be consistent with SWGDAM Interpretation Guidelines for Mitochondrial DNA Analysis.
- Updated GEDmatch PRO report to reflect the same typed SNPs in the UAS user interface, post user edits.


Known Defects

- None

Additional Notes

- None

Version History

Version	Date	Description of Change
A	Aug 2022	Initial Release
B	Jan 2023	Updates to reflect 

Appendix A

Table 1: List of SNPs calls previously called on the reverse strand

Changed SNP		NA24385 Call		Changed SNP		NA24385 Call	
SNP ID	SNP Category	UAS v2.2-2.4	UAS v2.5+	SNP ID	SNP Category	UAS v2.2-2.4	UAS v2.5+
rs1294331	Identity	G,A	C,T	rs3737576	Biogeographical Ancestry	A,A	T,T
rs1413212	Identity	A,G	T,C	rs2814778	Biogeographical Ancestry	A,A	T,T
rs993934	Identity	T,C	A,G	rs1876482	Biogeographical Ancestry	C,C	G,G
rs1357617	Identity	T,A	A,T	rs3827760	Biogeographical Ancestry	T,T	A,A
rs2399332	Identity	A,A	T,T	rs1229984	Biogeographical Ancestry	G,G	C,C
rs1355366	Identity	G,G	C,C	rs3811801	Biogeographical Ancestry	C,C	G,G
rs2046361	Identity	A,A	T,T	rs870347	Biogeographical Ancestry	T,T	A,A
rs1979255	Identity	C,C	G,G	rs2196051	Biogeographical Ancestry	T,C	A,G
rs251934	Identity	C,C	G,G	rs1871534	Biogeographical Ancestry	C,C	G,G
rs338882	Identity	C,T	G,A	rs3814134	Biogeographical Ancestry	T,T	A,A
rs1336071	Identity	G,G	C,C	rs1079597	Biogeographical Ancestry	G,G	C,C

rs214955	Identity	G,G	C,C	rs1572018	Biogeographical Ancestry	A,G	T,C
rs727811	Identity	C,C	G,G	rs1800414	Biogeographical Ancestry	A,A	T,T
rs763869	Identity	C,C	G,G	rs4411548	Biogeographical Ancestry	G,A	C,T
rs1463729	Identity	G,A	C,T	rs2593595	Biogeographical Ancestry	T,T	A,A
rs735155	Identity	G,G	C,C	rs2042762	Biogeographical Ancestry	A,A	T,T
rs3780962	Identity	T,C	A,G	rs3916235	Biogeographical Ancestry	A,G	T,C
rs10488710	Identity	G,C	C,G	rs310644	Biogeographical Ancestry	A,G	T,C
rs2076848	Identity	A,A	T,T	rs1800407	Phenotype	G,G	C,C
rs2920816	Identity	C,C	G,G				
rs2111980	Identity	G,G	C,C				
rs1335873	Identity	T,T	A,A				
rs1886510	Identity	C,T	G,A				
rs354439	Identity	A,A	T,T				
rs1454361	Identity	A,A	T,T				
rs722290	Identity	C,G	G,C				
rs1821380	Identity	G,C	C,G				
rs729172	Identity	C,C	G,G				
rs1382387	Identity	T,T	A,A				
rs1736442	Identity	A,G	T,C				
rs1024116	Identity	A,A	T,T				
rs719366	Identity	C,T	G,A				
rs445251	Identity	C,C	G,G				