Molecular Diagnostic Laboratory

Instrument, Assay and Software Verification Form

Alberta Health Services
Edmonton Zone

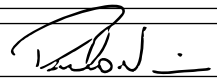
Verification type: Software (Appendix D)

Instrument verification should occur after a piece of equipment has been repaired on site or sent out for repair. As well the performance of a new instrument when identical to one already found in the laboratory should be verified before being employed in routine diagnostic work.

Assay verification should occur after minor modifications to a test have been employed (i.e. reaction volume, primer concentration, annealing temperature etc). At a minimum it must be ensured that a positive and normal control perform consistent with the old method and as expected.

Software verification should occur after the release of a new version or update to an existing in use software application. In some cases there may be a set panel of samples to run through the verification i.e. sequence validation panel (10 samples) for SeqPilot updates.

Software to be verified:	NGS Pipeline (v1.19.3, SNPs and short indels only)
Reason for verification:	Validation of the SNP/short indel detection by the pipeline, with report on accuracy from alignment, duplicate removal, realignment, base recalibration and variant calling by three independent callers, GATK4, Freebayes and Platypus.
Summary of testing and outcomes:	Validation was performed with 142 whole exome samples obtained from the European Genome-Phenome Archive, dataset EGAD00001001462 (doi:10.12688/f1000research.8219.2), with 402 Sanger-validated SNPs and short indels (< 30 nt). Code was slightly modified to accommodate sample IDs and carried up to the merged VCF step in the pipeline. Due to the number of variants that were not checked by Sanger sequencing in the study, it is impossible to assess specificity and sensitivity of the test, while only accuracy can be measured. All callers found 380 of 382 variants (of the 402, 20 were not found in the BAM file or had quality/coverage/problems), an accuracy of 99.48%, while 2 callers were able to determine 382 of 382 variants (100%).
Test Number(s):	142 Samples are stored in 192.168.1.169 external disk, result files are located in the pipeline repository under the validation/snps directory
Disorders affected:	Cancer

Verified by:	Paulo Nuin	Date (dd/mm/yyyy):	29/01/2019	Signature:	

Comments:					
Reviewed by:		Date (dd/mm/yyyy):		Signature:	

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