



EMQN Office

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24/01/23

To Whom it may concern,

I am writing to convey the utility of Variant Validator (VV) to our organisation, EMQN CIC, a global leading quality assurance provider in genomics, and the community of more than 3000 laboratories we currently serve. We routinely use the VV 'Validator' tool to ensure we are publishing the validated genotypes of samples supplied to participants under our external quality assessment (EQA) schemes, according to correct HGVS nomenclature. Our services aim to educate the genomics community on the correct reporting of variation identified within the human genome in the context of diagnostic reporting, so it is imperative that we demonstrate accurate use of this widely adopted variant nomenclature system. Variant Validator enables us to verify the correct HGVS representation of a variant efficiently and with confidence that the output is exact, since VV utilizes the HGVS Python package. Further, this year we announced our support for the community's transition from use of Locus Reference Genome (LRGs) reference sequences to Matched Annotation from NCBI and ENBL-EBI (MANE) transcripts. The VV 'Genes to Transcripts Tool' has been crucial in this process, enabling us to quickly identify MANE Select and MANE Plus Clinical transcripts, for genes covered by our EQA schemes. We will continue to recommend use of Variant Validator by our colleagues and community.

Yours faithfully,

Dr Rachel L. Taylor

Scientific Programme Manager, EMQN CIC