From: Julie Eggington <jeggington@genomicinterpretation.org>
Date: Tuesday, 7 January 2020 at 18:34
To: Bryan Gall <bgall@genomicinterpretation.org>
Cc: Peter Causey-Freeman <peter.causey-freeman@manchester.ac.uk>, "Dalgleish, Raymond W.M. (Prof.)" <raymond.dalgleish@leicester.ac.uk>
Subject: Re: VariantValidator query (: hgvs 1.1.3 uta_20180821)

Hi Peter,

Our mission is to drive quality in clinical genetics, genomics and precision medicine. We are deeply concerned about a lot of sloppy science that happens within clinical labs, and we work with industry stakeholders to find ways to improve quality expectations. Our biggest project launch will be later in 2020 when we launch ELEVATEGenetics to assist Blue Shield of California in their lab/test selection. We will be working with labs and health insurance companies to assess labs against more stringent quality standards, thus connecting reimbursement more directly to quality like never before. I've talked to Reece Hart about it, who I believe you know.

So what has kept us afloat is as we've been developing ELEVATEGenetics is a lot of volunteers, as well as a small contract with a drug development company that keeps a disease registry of leukemia. The disease registry consists of many patients across the USA. Their doctors send in all types of info on the patients. The doctors have started sending in genomic profile reports. The drug development company has contracted with us to take those reports, and catalog them in a big spreadsheet. We have been using Variant Validator to check the nomenclatures, and to translate them into genomic coordinates (build 38). We've been disappointed at the nomenclature errors we've been finding in reports, particularly due to left and right alignment issues.

Kind regards,

Julie

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