Dalgleish, Raymond W.M. (Prof.)

From:	Simon Denil <Simon.Denil@uzbrussel.be>
Sent:	04 December 2020 11:51
To:	'admin@variantvalidator.org'
Subject:	note of appreciation

Dear Dr. Causey-Freeman,
Dear VariantValidator development team,

With this email I would like to express both for myself and on behalf of my institute, our profound gratitude and appreciation for the efforts you have invested in creating, maintaining and making publicly available the VariantValidator suite of tools.

At our institute, the Centre for Medical Genetics at the Brussels University Hospital, we process all kinds of sequencing data. The samples are usually derived from patients that come into the hospital with all manner of indications. We have recently improved our internal processes to do a better job of tracking genetic variant information over time (since new insights inevitably come along). VariantValidator has been indispensable in this effort.

As you are undoubtedly aware, simply matching variants by VCF-style chr:start:stop(:ref:alt) information, will yield a lot of false negatives in a search. Variant callers are updated, different callers may be used for different applications, … To address these issues we have created a private database that tracks distinct observed variants across all relevant internal data sources. The key break-through in this application is that fully compliant genomic HGVS notation is used as the primary key, thus allowing unambiguous matching between datasets and correcting of human error that may have arisen from manual data entry.

We would also like to thank you for your advice which helped us set up a local copy of VariantValidator on our own computer systems. Even though running these variant validation queries is unlikely to yield easily identifiable information, it is considered patient data and therefore should not leave our internal network.

We are now validating thousands of variants every week with VariantValidator. Even though this does not show up in your usage statistics for the public instance, please know that the tool is performing valuable work out there in the world. In the near future I also hope to promote the tool for research purposes which should results in few citations coming your way as a more tangible token of appreciation.

Kind regards and seasonal wishes,

Simon Denil, PhD

Simon Denil
BioIT – Centrum voor Medische Genetica

Simon.Denil@uzbrussel.be

Laarbeeklaan 101 – 1090 Brussel

Tel 02 474 90 51

www.uzbrussel.be

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