Dalgleish, Raymond W.M. (Prof.)

From: I.F.A.C.Fokkema@lumc.nl
Sent: 08 December 2020 09:42
To: admin@variantvalidator.org
Cc: Dalgleish, Raymond W.M. (Prof.); peter.causey-freeman@manchester.ac.uk
Subject: VariantValidator and LOVD

Dear Pete and Raymond,

As you know, I have been working on building VariantValidator into LOVD to validate and automatically correct variant descriptions and mappings. We previously used the Mutalyzer service for this, which lacked validation of results during mapping and at times provided unreliable results. Furthermore, VariantValidator has been invaluable in handling variants located in regions where the genome and the transcript sequence can not fully be aligned. Since July 2020, we are using VariantValidator to verify all of our country's genome diagnostic labs' data which has markedly improved its accuracy and we were able to build and publish our LOVD hg38 genome browser track, now available at the UCSC and EBI genome browsers. I am now confident that the variant data provided by LOVD are fully standards-compliant and reliable for use in genetic testing.

Best regards,

Ivo

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Ivo F.A.C. Fokkema
Lead developer LOVD/LOVD+

LOVD - Leiden Open Variation Database
A flexible, freely available tool for collection
and display of DNA variations.
LOVD+ - LOVD for diagnostics
A free tool for gene panel and WES analysis
using configurable analyses.

Celebrating 16 years of LOVD!
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