

On 5/07/2012 7:22 PM, P.E.M.Taschner@lumc.nl wrote:

Dear International Scientific Advisory Committee,

Following my proposal of the activity to recognize the HGVS variation nomenclature guidelines as Human Variome Project standard human variation nomenclature, I would like to propose an assessment of HGVS variation nomenclature checker software, in particular the Mutalyzer 2 package (<https://mutalyzer.nl>) which is recommended by Human Mutation to check variant descriptions prior to publication. Mutalyzer is also used by many LOVD gene variant databases during variant submission.

Recognition of Mutalyzer as a standard tool might lead to more wide-spread recommendations in the author instructions of journals and thereby help to improve the quality of variant descriptions in the literature.

Thank you very much for your time and consideration.

Best regards,

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