

# Sequence Variant Nomenclature

*please support the HGVS nomenclature work. DONATE at:  
[https://www.paypal.com/donate/?hosted\\_button\\_id=DHJVLF3Z2TA2U](https://www.paypal.com/donate/?hosted_button_id=DHJVLF3Z2TA2U)*



## HGVS nomenclature activities performed in 2021

*Leiden, January 2022  
Johan T. den Dunnen*

Short report on the main activities performed in 2021 on the topic of HGVS nomenclature (the “HGVS recommendations for the description of sequence variants”). The report is written on behalf of the HGVS Variant Nomenclature Committee (HVNC), a committee working under the auspices of the Human Genome Organization (HUGO), as a member of the HUGO Nomenclature Standards Committee (HNSC).

### *Sequence Variant Description working group (SVD-WG)*

- the HVNC finalised the Terms of Reference and send it to the HUGO board for approval.
- in 2021 the HVNC welcomed two new members. Ros Hastings (Oxford, UK) succeeded Jean McGowan-Jordan as the ISCN representative in the committee. Terence Murphy (NCBI, USA) succeeded Donna Maglott as representative of the major databases (EBI/NCBI).
- members of the committee met for the first time in an online meeting (Oct.2021). Topics discussed included the future organisation of the committee within HUGO (Terms of Reference), membership of the HVNC, call for new members (Ja./Feb.2022) and proposal SVD-WG010.
- the committee finalised an update of the accepted “Reference Sequences”. With active input from committee members Pete Causey-Freeman, Raymond Dalglish, Reece Hart and extensive discussions with EBI and Genomics England LTD, Ensembl reference sequences (ENSG, ENST, ENSP) are now accepted as approved reference sequences.
- the SVD-WG completed the Community Consultation call for SVD-WG010 (var distance) suggesting changes to the nomenclature. Given the comments received, the HVNC decided the proposal needs further improvements before it can be accepted. A modified proposal is in preparation.
- no new versions of the HGVS nomenclature were released. The current version of the HGVS nomenclature remains v20.05.

## Promotion

### - HGVS online Q&A

June 2021, Johan den Dunnen started a series of bi-monthly online HGVS nomenclature Q&A sessions. So far four one hour sessions have been completed. Each sessions starts with a 5 min. introduction, followed by a 40-45 min Q&A session and ending with 10-15 min. in which participants can ask direct questions about HGVS nomenclature. The interactive Q&A sessions use the DirectPoll system ([www.DirectPoll.com](http://www.DirectPoll.com)), allowing participants to answer about 15-20 questions per session. In addition, using so called Socrative classrooms ([www.Socrative.com](http://www.Socrative.com)), questions and answers have been made available for users which were not able to join the live online session. Up to over 200 users per classroom have so far used this option to test their knowledge on HGVS nomenclature.

### - VarNomen@HGVS.org e-mail

The chair received ~150 questions about HGVS nomenclature by e-mail, all of which have been answered.

### - HGVS nomenclature Facebook

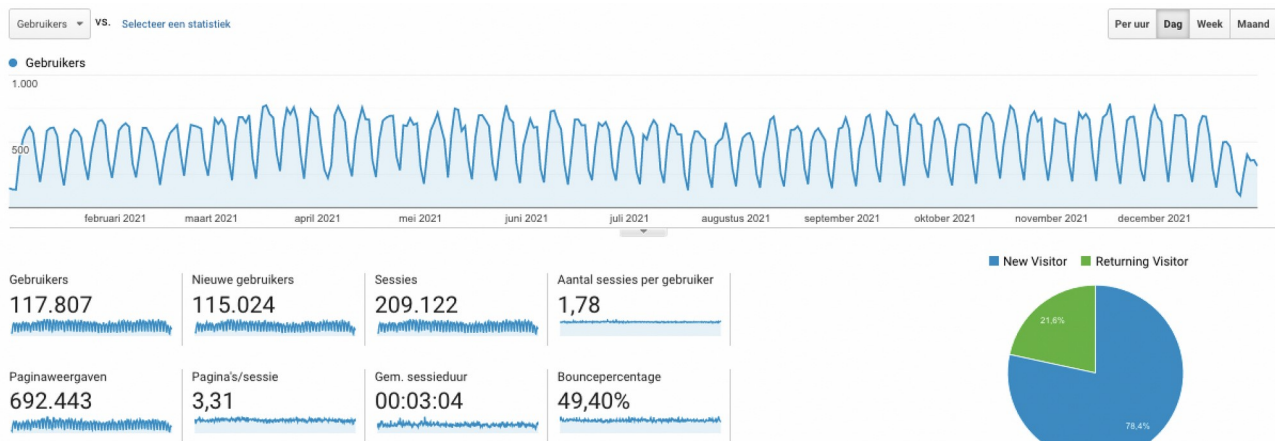
Based on the e-mail questions received, 26 posts (20 in 2020) were made on Facebook. In addition, some 10 questions received on Facebook were answered.

### - VarNomen.HGVS.org website

Many improvements were made to the HGVS nomenclature website (mainly correcting mistakes, adding examples, general updates), most triggered by questions or remarks received by e-mail.

### - web site statistics

The HGVS nomenclature website visits increased again, about 15-20% in 2021. Daily visits peaked at 783 in November (2020 peak 720). Annual totals for 2021 are 692,443 pageviews (632k in 2020) in 209,112 sessions (185k in 2020), from 117,807 visitors (103k in 2020).



### - EJHG

Collaboration with the European Journal of Human Genetics to correct the use of HGVS nomenclature and check mandatory data submission for every paper submitted to the journal ended in June 2020.

## Education

Besides the bi-monthly Q&A sessions, lectures / demos / workshops on HGVS nomenclature were contributed to the GV/HUGO "Variant Effect Prediction training course", the UNESCO-funded "Online training course in curation of human gene/disease databases" and a training course in Breda (Netherlands).

***Other business***

- Raymond Dagleish, Pete Freeman and Johan den Dunnen, as members of the HVP Genetics Journals working group, co-authored a paper promoting the use of HGVS nomenclature: Higgins et al. (2021). "Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals". Hum. Mutat. 42(1), 3-7.

***Current members HGVS Variant Nomenclature Committee (HVNC)***

- Oleg Agafonov
- Raymond Dagleish
- Johan den Dunnen (chair)
- Peter Causey-Freeman
- Reece Hart
- Ros Hastings (ISCN)
- Terence Murphy
- Anne-Francois Roux
- Peter Taschner

*Report made by Johan den Dunnen, chair of the SVD-WG*