

HGVS nomenclature activities performed in 2019

*Leiden, February 2020
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Short report on the main activities performed in 2019 on the topic of the “HGVS recommendations for the description of sequence variants (HGVS nomenclature)”. The report will be shared with the Human Genome variation Society (HGVS), Global Variome/Human Variome Project (GV/HVP) and the Human Genome Organisation (HUGO), the organizations officially recognizing the HGVS nomenclature standard.

Sequence Variant Description working group (SVD-WG)

- discussions to get new SVD-WG members proceeded but GV/HVP did not initiate the official steps required. The working group was extended with Peter Causey-Freeman (Leicester, UK) as a new member.
- the SVD-WG completed Community Consultation calls for SVD-WG007 (RNA fusion) and SVD-WG008 (RefSeq) suggesting changes to the nomenclature. Other proposals are in preparation, incl. deletion-insertions, repeated sequences and HGVS/ISCN.
- end 2018 HGVS v19.01 was released (replacing version 15.11). The SVD-WG did not release a new HGVS nomenclature version. Version 19.01 is still valid.
- members of the committee expressed an interest to have a face-to-face meeting to discuss the many issues that are on the table. Realization lacks funding; members can only meet on occasion during the annual ESHG/ASHG meeting but then rarely 2 or 3 SVD-WG members will be present.

Promotion

VarNomen@HGVS.org e-mail

Johan den Dunnen received ~200 questions about HGVS nomenclature by e-mail all of which have been answered.

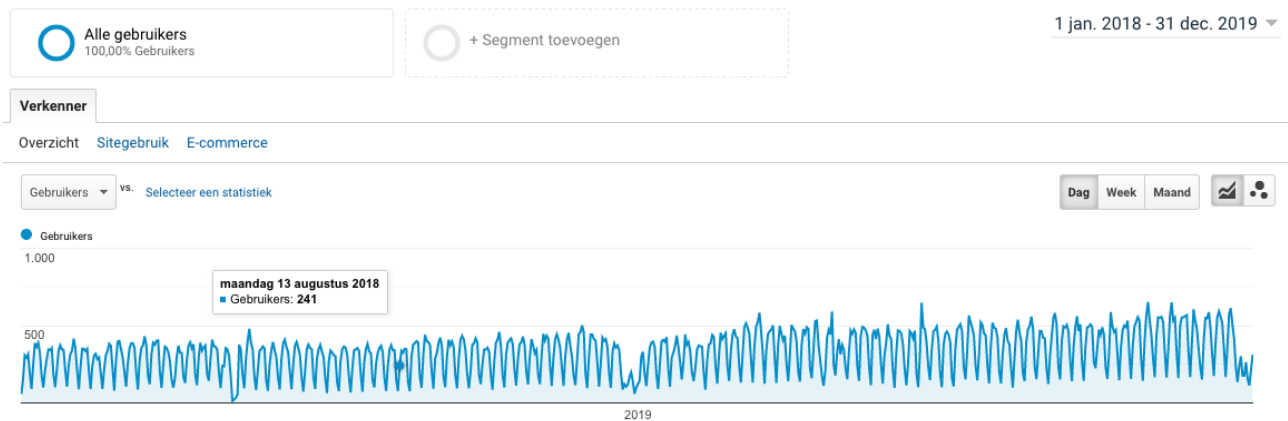
HGVS nomenclature facebook

Based on the e-mail questions received, 17 posts were made on facebook (mirrored on HGVS twitter Twitter). In addition some 20 questions were answered on facebook.

VarNomen.HGVS.org website

-many improvements were made to the HGVS nomenclature website, most triggered by questions or remarks received by e-mail. Correcting errors, adding more examples and improving explanation.

- the HGVS nomenclature website visits increased from ~400 to ~500 visitors daily (peaking at 650, see Figure). The 2019 annual total was 154,000 sessions (121,000 in 2018). The dip in visits in Jan. 2019 was caused by a problem with the HGVS website (www.HGVS.org).



EJHG

For the European Journal of Human Genetics proper use of HGVS nomenclature and mandatory data submission was checked for every paper submitted to the journal. In 2019 the total number of papers checked over time passed the 3,000 mark. The EJHG financially supports this work (partially employed administrative assistant).

ISCN2020 book

Johan den Dunnen participated in the ISCN meeting (Copenhagen, Denmark) preparing the ISCN2020 book. Chapter 16, ISCN & HGVS, was updated.

Education

- HGVS nomenclature presentations were given at several national and international meetings / courses / workshops: Breda, Leiden and Rotterdam (Netherlands), Paris (France), Moscow (Russia), Seoul (Korea), Copenhagen (Denmark)

Other business

- since the HGVS board was not able to come to a consensus decision, an anticipated collaboration on HGVS nomenclature with DNV-GL (Norway) could not be started. End 2019 the HGVS board decided "*HGVS nomenclature is an open, community-owned standard so one does not need permission to work with it*". Contacts with DNV-GL will be re-initiated early 2020.

- a HGVS nomenclature book, similar to the ISCN book series, has been an option for many years. Contacts with Wiley were never successful. Karger expressed an interest and discussions have been initiated (provisional deadline 2021).

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