

# Sequence Variant Nomenclature

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## **HGVS nomenclature activities performed in 2020**

*Leiden, April 2021  
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Short report on the main activities performed in 2020 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 Organization (HUGO), the organizations officially recognizing the HGVS nomenclature standard.

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

- a draft document describing the activities, responsibilities, and membership of the SVD-WG was sent to the boards of HGVS, GV/HVP and HUGO. Ultimately, the document should be approved by these organisations.
- the working group was extended with Oleg Agafonov (DNVGL, Oslo, Norway) as a new member.
- the SVD-WG completed Community Consultation calls for SVD-WG009 (conversion) suggesting changes to the nomenclature. In addition, the “Named extension ISCN” was updated to cover the new ISCN 2020 recommendations. Other proposals are in preparation, including deletion-insertions and repeated sequences.
- May 2020 HGVS v20.05 was released (replacing version 19.01).
- members of the committee expressed an interest in having a face-to-face meeting to discuss the many issues that are on the table. Realization of this desire lacks funding; members can only meet on occasion during the annual ESHG/ASHG meeting but then only 2 or 3 SVD-WG members will normally be present. Website and nomenclature question replies will be modified to include a request to support the HGVS nomenclature work by making donations.

### ***Promotion***

VarNomen@HGVS.org e-mail

Johan den Dunnen received ~250 questions (200 in 2019) about HGVS nomenclature by e-mail all of which have been answered.

HGVS nomenclature Facebook

Based on the e-mail questions received, 20 posts (17 in 2019) were made on Facebook. In addition, some 15 questions were answered on Facebook.

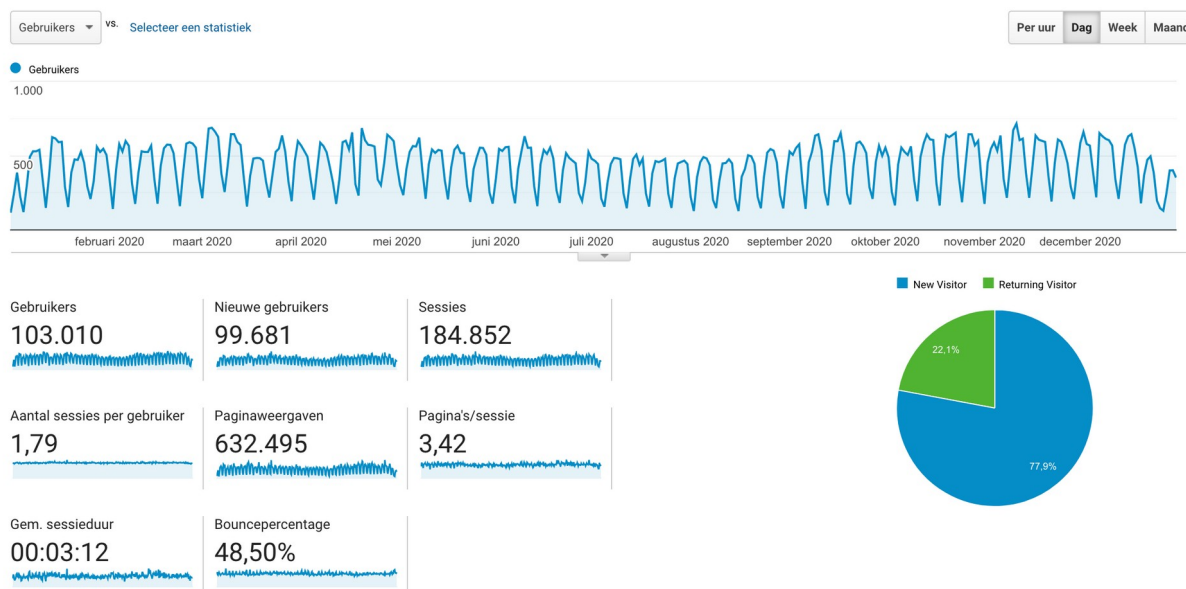
VarNomen.HGVS.org website

-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 to over 500 visitors daily (peaking at 720, see Figure). The 2020 annual total was 184,852 (154,000 in 2018).

## EJHG

For the European Journal of Human Genetics, correct use of HGVS nomenclature and mandatory data submission was checked for every paper submitted to the journal. In 2020 the total number of papers checked passed the 3,500 mark. The EJHG financially supports this work (partially employed administrative



assistant), but this funding will end July 2021.

## ISCN

The new ISCN recommendations, through the ISCN 2020 book, were released late summer 2020. Johan den Dunnen, as member of the ISCN committee, worked especially on Chapter 16: ISCN & HGVS.

## Education

- due to the pandemic no HGVS nomenclature presentations were given.

## Other business

- Raymond Dalgleish established a contact with Ian Cree, the chair of the WHO Classification of Tumours Group. Ian Cree also contacted Johan den Dunnen and showed how the new recommendations strongly recommend the use of the HGVS standard for variant descriptions. Discussions are ongoing to see whether stronger links can be made with WHO.

- Raymond Dalgleish, Pete Freeman and Johan den Dunnen, as members of the HVP Genetics Journals working group, are co-authors on 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.

- Johan den Dunnen is discussing with Karger options to produce an HGVS nomenclature book, similar to the ISCN book series. The provisional deadline is end 2021.