https://www.humanvariomeproject.org/activities/sequence-variant-description-committee.html

Stimulated by the HGVS, standards have been developed regarding the description of sequence variants, the so called HGVS recommendations for the description of sequence variants ("Mutation nomenclature"). Over time, these recommendations have developed into a standard that is used world-wide, esp. in the field of human and clinical genetics. The latest version of the recommendations can be found at the official HGVS nomenclature web site (https://www.hgvs.org/varnomen).

Questions regarding the current recommendations and requests for modifications and extensions should be directed at the "Sequence Variant Description working group (SVD-WG)": mail to "Varnomen @ HGVD.org" (remove spaces). Decisions on specific topics are made by the SVD-WG, which includes a "Community Consultation" step (http://varnomen.hgvs.org/bg-material/consultation/) to gather community opinions. Mails will be answered by the current chair of the SVD-WG, Johan den Dunnen.

The consistent and unambiguous description of sequence variants is essential to report and exchange information on the analysis of a genome. In particular, DNA diagnostics critically depends on accurate and standardized description and sharing of the variants detected. The sequence variant nomenclature system proposed in 2000 by the Human Genome Variation Society has been widely adopted and has developed into an internationally accepted standard. The recommendations are currently commissioned through a Sequence Variant Description Working Group (SVD-WG) operating under the auspices of three international organizations: the Human Genome Variation Society (HGVS), the Human Variome Project (HVP), and the Human Genome Organization (HUGO). Requests for modifications and extensions go through the SVD-WG following a standard procedure including a community consultation step. Version numbers are assigned to the nomenclature system to allow users to specify the version used in their variant descriptions.

den Dunnen et al. presented the current recommendations, HGVS version 15.11, and briefly summarize the changes that were made since the 2000 publication. Most focus has been on removing inconsistencies and tightening definitions allowing automatic data processing. An extensive version of the recommendations is available online, at http://www.HGVS.org/varnomen [1].

1)

den Dunnen JT, Dalgleish R, Maglott DR, Hart RK, Greenblatt MS, McGowan-Jordan J, Roux AF, Smith T, Antonarakis SE, Taschner PE. HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Hum Mutat. 2016 Jun;37(6):564-9. doi: 10.1002/humu.22981. Epub 2016 Mar 25. PMID: 26931183.