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. Here, we present 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.