

HGVS VARIANT NOMENCLATURE COMMITTEE (HVNC)

TERMS OF REFERENCE

1. The HGVS Variant Nomenclature Committee

The HGVS Variant Nomenclature Committee (HVNC), formerly known as the HGVS SVD-WG (Sequence Variant Description Working Group), has been initiated at the request of the boards of Human Genome Variation Society (HGVS), Human Variome Project (HVP)/Global Variome (GV) and Human Genome Organisation (HUGO).

- 1.1. The scope of the work of the HVNC, as defined here, is authorised by the HUGO board.
- 1.2. A request to modify the scope of the HVNC can be made in writing to the HUGO board.
- 1.3. The HVNC Terms of Reference will be made available on the HUGO website.

2. Scope of the HVNC

- 2.1. Define a standard for the unambiguous description of all sequence variants at the DNA, RNA, and protein level, the so-called HGVS nomenclature standard; and
- 2.2. Ensure the recommendations are usable by both humans and computers; and
- 2.3. Publish the current sequence variant description recommendations on the HGVS nomenclature website (hgvs-nomenclature.org); and
- 2.4. Promote the use of the HGVS nomenclature using all available means; and
- 2.5. Report annually on the activities of the HVNC to the HUGO Nomenclature Standards committee and the HUGO board; and
- 2.6. Provide advice to the HUGO Nomenclature Standards committee and the HUGO board about any organisational issues hampering the committee and how to maintain the committee and the HGVS nomenclature standard into the future.

3. Membership of the HVNC

- 3.1. Members of the HVNC are appointed by the HUGO board.
 - 3.1.1. The HVNC aims to have 7-11 voting members.
 - 3.1.2. Members will be appointed on a bi-annual (once every two years) basis.
 - 3.1.2.1. Application for membership of the HVNC will be organised every two years (every even year) by the HUGO board.
 - 3.1.2.2. The application process and candidate requirements will be established by the current membership.
 - 3.1.2.3. Any member of the global professional genomics community will be eligible to apply to become a member of the HVNC. Appointed members should together represent interested communities, including gene/disease specific database curators, central repositories, clinical geneticists, diagnostic laboratories, bioinformaticians, scientific journals, etc.
 - 3.1.2.4. Applications for membership will be evaluated and recommendations for new members from among the applicants will be assembled by the HVNC. The

committee Chairs will propose recommendations for new members to the HUGO board based upon the consensus of the HVNC.

3.1.2.5. Based on the proposal of the HVNC, at their annual meeting, the HUGO board will appoint new members of the HVNC.

3.1.2.6. One member of the ISCN standing committee, preferably the Chair, will be an appointed member of the HVNC, representing the cytogenomics community.

3.1.2.7. As appropriate, standing members from other professional nomenclature committees may be invited by current members of the HVNC.

3.1.3. The term of membership for each member is four years.

3.1.3.1. Members may not be re-appointed beyond two consecutive terms. Only a maximum of 50% of the committee membership can be re-appointed at any one time.

3.1.3.2. Members reaching their limit on consecutive terms must wait four years before reapplying for the HVNC.

3.1.4. Each member should attend a minimum of 50% of meetings over an 18-month period; otherwise, they will forfeit their position.

3.2. Co-Chairs of the HVNC are appointed by the HUGO board.

3.2.1. The HVNC will have two co-Chairs.

3.2.2. The HVNC co-Chairs are together referred to as “the Chairs”. Use of “a Chair” throughout this document refers to either co-Chair.

3.2.3. Nominations for Chair positions will have deadlines the same as those organized by the HUGO committee for membership applications (Section 3.1).

3.2.4. The term for appointment as Chair is four years.

3.2.4.1. A Chair may be re-appointed beyond two consecutive member terms.

3.2.5. Nominations for appointment as a Chair should include a motivation letter and a CV.

3.2.5.1. Self-nominations are allowed.

3.2.5.2. Nominations should be e-mailed to the HVNC by the nomination deadline.

3.2.6. The Chairs (like HVNC members) must be nominated for re-appointment to position at the conclusion of each term as a Chair.

3.2.7. Nominations for appointment as a Chair will be submitted by the HVNC to the HUGO board for consideration.

3.2.7.1. If there are only enough nominations to fill vacant Chair positions, the HVNC will be notified and these nominations will be advanced to HUGO by the current Chairs without further discussion.

3.2.7.2. If there are more nominations than vacant Chair positions, the HVNC will discuss all nominees, and a consensus recommendation of the HVNC for the vacant chair positions will be proposed to HUGO by the current Chairs.

3.2.8. The Chairs must be selected from the active HVNC membership.

3.2.9. The Chairs are expected to equally share responsibility for leading the activities of the HVNC.

- 3.2.10. The Chairs will represent the committee at meetings of the HUGO Nomenclature Standards Committee.
- 3.3. Non-voting advisors may serve in the committee at the request of the HVNC.
 - 3.3.1. A Chair may propose to appoint an advisor to the HVNC.
 - 3.3.2. Advisors must be approved by the HVNC.
 - 3.3.3. Advisor appointments are renewable on a bi-annual basis.
- 3.4. Interim appointments will be considered on an as-needed basis at the discretion of the Chairs.
 - 3.4.1. Interim appointments may be proposed by a Chair when an HVNC member, Chair, or advisor ends their term prematurely.
 - 3.4.2. Interim appointments for HVNC members or Chairs must be approved by the HVNC and the HUGO board. Interim appointments for HVNC advisors must be approved by the HVNC.
 - 3.4.3. An interim appointment lasts until the conclusion of the term the interim member was invited to complete.
 - 3.4.4. Interim appointments do not count against the term limits for HVNC members.

4 Organisation

- 4.1. No business should be transacted at a meeting of the committee, whether in person or online, unless at least one of the HVNC Chairs is present, along with a quoracy of >50% attendance.
- 4.2. At the request of a Chair, individuals who are not members of the committee may be invited to attend meetings to provide advice and assistance where necessary. Invitees will have no voting rights and may be requested to leave the meeting at any time by a Chair.
- 4.3. Business should be transacted following the agenda distributed in advance of the meeting.
 - 4.3.1. The HVNC Chairs are responsible for distributing a preliminary agenda to the committee within two weeks following the prior meeting.
 - 4.3.2. Meeting agenda proposals may be submitted by committee members at least three weeks in advance of the next scheduled meeting.
 - 4.3.3. The committee Chairs are responsible for distributing a finalized meeting agenda to the committee at least two weeks in advance of the next scheduled meeting.
 - 4.3.4. Planned agenda items not discussed should carry over to the subsequent meeting and should take precedence over new agenda items.
- 4.4. The HGVS Nomenclature should undergo consultation with the committee and the community for any substantial changes to the recommendations.
- 4.5. Decisions in the committee are made on the basis of majority votes.
 - 4.5.1. The committee Chairs may motion for a formal decision of the committee during the course of a meeting.
 - 4.5.2. Any motion for decision may proceed with unanimous consent if no request for a vote on the motion is requested by a committee member.

- 4.5.2.1. If a request for a vote on the motion is raised by a committee member and the request is seconded by another committee member, the motion proceeds to a full vote of the committee.
- 4.5.2.2. If no request for a vote on the motion is raised or no member seconds the request for a vote, then the committee is considered to have unanimously approved the motion.
- 4.5.2.3. Votes will be taken by show of hands during the meeting.
- 4.5.2.4. If a vote on the motion is tied, the Chairs will cast the deciding votes.
- 4.5.2.5. If the Chairs also are a tied vote, the motion fails.
- 4.5.3. If a quorum cannot be achieved on a vote, a committee discussion should identify solutions (e.g. designated alternates, extended voting period) to reach a quorum.
- 4.5.4. Aggregate (anonymous) outcomes of votes will be provided to the general public as part of the meeting minutes.
- 4.6. To promote transparency and community involvement, anonymised minutes of meetings of the HVNC will, after approval by its members, be published online by HUGO.