

Minutes

2025-04-07 HVNC Bi-monthly Regular Meeting

Attendees: Johan T den Dunnen, Alex Wagner, Ivo Fokkema, Jeroen Laros, Matthew Stachowiak, Gwendolyn Bennett, Ros Hastings, Marina DiStefano, Hencher Lee

Absent: Timothy Hefferon, Rachel Taylor

Regrets: Reece Hart

Agenda

1. Opening and Confirm Agenda
2. Confirm minutes from prior meeting
 - a. create anonymized minutes for posting on HUGO website within 1-2 weeks
3. Review action items from previous meeting
 - a. from the next meeting on, we'll only copy the open tasks to the new agenda at the start of the meeting.
 - b. the issue described in #147 will be summarized and forward to the committee members as the draft of SVD-WG011.
 - c. Tim Hefferon will be contacted on the participation in the Google Groups. There may be issues for him to join external initiatives.
4. Announcements and Updates
 - a. update on Parentheses / Variant Observation Status Task Force
 - i. a list of individuals and organizations was presented intended to be invited to the task force. Several committee members will take part, representing the HVNC. Committee members are encouraged to add additional organizations or individuals they think would be appropriate for this effort.
5. Resolutions
 - a. affirm adoption of HVNC Terms of Reference
 - i. some new comments were received. We went through them one by one until all comments were resolved. It was decided that the procedure for how minor and major changes are handled will be moved to a page on the HGVS Nomenclature website. This will allow the TOR to be more immutable.
 - ii. with nine members present, no further review is needed. The Terms of Reference have been approved.
 - b. HVNC co-chair selection
Alex Wagner was the only candidate; no other members stepped forward. Alex was approved as co-chair
6. Discussions
 - a. a proposal for the nomenclature of positions upstream and downstream of transcripts was introduced.
 - i. earlier, we had agreed to allow transcript-based positions outside of transcripts (e.g., the promotor region). However, the syntax was never discussed. Jeroen shows his suggestions for describing those positions (five in total).
 - ii. option 4 (c.u10 and c.d10) has issues in case the UTR lengths of transcript reference sequences are updated without a change of the version number. Using, e.g., c.-100-u10 would allow for a more "recoverable" notation when that happens.

Whether reference sequences are still changed this way without a version change is unknown.

iii. a formal proposal to be discussed in the next meeting will be made.

b. list of invalid DNA descriptions extracted from the HGVS nomenclature website.

i. IF built a tool that recognizes HGVS-compliant variant descriptions, also without a reference sequence present. An analysis of the variant descriptions given on the HGVS nomenclature website revealed 89 errors. Ivo highlighted a few common errors.

1. repeat nomenclature in introns break triplet rule. Decision: the triplet rule is for coding regions only; intronic repeats can be of any length, also on coding reference sequences. The documentation will need to be updated. Ivo will draft these changes.

2. variant positions using “cen” are given, e.g.,

NC_000011.10:g.15825273_cen_qter. Cen should be removed, this will be corrected.

Side point: The ISCN pages are out of date and need updating. ISCN will send a Word doc with updates and the required changes to the website will be made.

3. for other variants that Ivo is unsure about, he will first email JdD. Whenever he thinks that Johan's reply warrants a broader agreement within the committee, he will forward the issue to the committee.

c. Introduction resolution to issue #147 (POSTPONED to next meeting)

d. (copied from the previous meeting's agenda) Can we resolve the various interpretations of NC(NM) reference sequences? (related discussion, another one, another related discussion)

i. the reasoning of the issue and why it needs to be resolved is explained. HVNC members have been working on a proposal; it's nearly done. It will be send around when done.

e. do we need to schedule HGVS releases, or agree on how long changes need to wait for a new release?

i. decided to release after agreement in the committee meetings. We can prepare a link for everybody to check the changes since the last release on GitHub. Reviewing the changes since the last release will become a fixed agenda point.

f. updating ISCN on HGVS website

7. AOB

Regarding rejected proposal SVD-WG010; is there a new proposal being formed? Yes, see Action item #7.