

Minutes

2025-09-29 HVNC Bi-monthly Regular Meeting

Attendees: Alex Wagner, Ivo Fokkema, Jeroen Laros Matthew Stachowiak, Gwendolyn Bennett, Timothy Hefferon, Marina DiStefano, Hencher Lee

Regrets: Johan den Dunnen

Absent: Ros Hastings

Agenda

1. Opening and Confirm Agenda
 - a. discussion about recording; where does this end up? (likely, with HUGO)
2. Confirm minutes from prior meeting
 - a. confirmed
3. Review action items from previous meeting
 - a. more time was requested to review the proposal on variant distance (agenda item 5f); one week was added(October 6th)
4. Announcements
 - a. anonymized minutes from July meeting
 - i. skipped to next meeting
 - b. update on Parentheses Task Force
 - i. a proposal was created, which was reviewed and discussed in a first meeting. There is progress, and we'll meet again this Friday
 - c. update in ISCN committee and ISCN 2024 on HGVS website.
 - i. skipped to next meeting
5. Discussions
 - a. Review any open or merged changes to the site since the last meeting
 - i. Merged: PR #231, PR #233
 - ii. Open: PR #151. We will return to this next meeting
 - b. Advancing proposals Proposal Creation and Voting Procedure
 - i. We reviewed the document in detail, making improvements directly within it. We also added a few action points in the comments.
 - ii. There were no further objections to accept this document.
 - c. Proposal to resolve the various interpretations of NC(NM) reference sequences (v2).
 - i. We went through the updated proposal, after which some questions came up and were discussed.
 - ii. Since we ran out of time, please add your comments to this discussion. Please note that the discussion highlights the issue, but doesn't include the current updated proposal.
 - iii. During the next meeting, we will discuss this proposal again.

(end of meeting)

Not discussed

 - e. Nomenclature of positions upstream and downstream of transcripts.
 - i. Proposal for using n.- and n.* instead of c.u and c.d, respectively.
 - f. Combining variants (revision of SVD-WG010).
 - g. HGVS Validation Test Suite

- h. Paper on using Unicode characters in the HGVS Nomenclature.
 - i. Raymond Dagleish really wants to respond and is preparing a response.
- i. Email from David Hernandez (Sophia Genetics) on the insertion/fs syntax on the protein level.
- j. Introduction to his proposal to better standardize the repeat syntax:
<https://gist.github.com/jfjlaros/5580d355a948a57a867ee6b152c39bdd>

6. AOB