

## Minutes

2025-02-03 HVNC Bi-monthly Regular Meeting

Attendees: Johan T den Dunnen, Alex Wagner, Ivo Fokkema, Jeroen Laros, Matthew Stachowiak, Gwendolyn Bennett, Marina DiStefano, Hencher Lee, Rachel Taylor  
Absent: Timothy Hefferon  
Regrets: Ros Hastings

## Agenda

1. Opening and Confirm Agenda

2. Confirm minutes from prior meeting

3. Review action items from previous meeting

4. Announcements

a. Second vote results on SVD WG010

- i. Prior vote was split 4-4, with 3 abstaining
- ii. Background and Response\_Proposal\_SVD-WG010.pdf
- iii. Discussion in 10/7 minutes, item 7
- iv. Voting (without abstain) requested again: HVNC SVD-WG010

1. 10 votes, 60% against adopting the proposal, 40% suggest adopting. The proposal was hereby rejected. The website should be updated.

2. The question now is how to proceed. This proposal has been rejected, but the issue that it was trying to solve is still present. Will somebody else create a new proposal? It was proposed to isolate the main issue (the difference between variant descriptions on c. and g. levels because of the codon-exception) into a proposal. Making smaller proposals may help the decision-making process, as larger proposals are more easily rejected because one disagrees with just one part of the proposal.

b. Updated Terms of Reference now available

- i. HVNC Terms of Reference
- ii. Open for review and further comment
- iii. Need for further discussion - co-Chair

1. Following HUGO resolution: HUGO Board Resolution\_031624.pdf

2. Revisions to document made to accommodate 2 chairs

a. No objections from the committee to have two chairs. [...] offers to take up the position, there are no objections.

b. Chairs should have equal rights; if, for a certain vote, the committee is equally divided, the chair used to decide. With two chairs, when the chairs disagree on a vote, the motion fails.

c. Give everybody two weeks to go through the Terms of Reference again and suggest/request changes. When there are no substantial changes or requests, we can finalize it in the next meeting and submit it to HUGO.

d. It's probably best to have the terms on the HUGO website, and we can refer to it from the HGVS nomenclature website. We can do the same for the minutes; have anonymous minutes on the HUGO website, while we have the full minutes on the Google Drive.

e. We discussed the process of how chairs are nominated. Chairs should self-nominate, after which we will discuss the applications during a meeting. When only two people apply, perhaps things can be simplified and, e.g., a motivation letter would not be needed. Otherwise, we may want to have each applicant explain what they want to focus on and in what

direction they want to move. The committee can then discuss and nominate two chairs to HUGO. The names of other applicants will also be shared.

## 5. Discussions

### a. Request to develop solution to issue

i. Issue: Use of Parentheses for Variant Observation Status in HGVS

ii. It is suggested to create a task force to handle the issue involving using or not using parentheses in the HGVS nomenclature. It's a strongly debated issue, and it's been ongoing for a long time. Perhaps a task force focused on this issue to come up with a proposal to submit to the committee. The task force would also be responsible for the community involvement.

iii. Voting: 6 agree to set up the task force, 1 against, 2 abstaining. Result: Accept the proposal to create the task force.

iv. Is there a proposal already? Heidi Rehm's proposal isn't a full proposal that we can vote for to adopt it or not, so should we ask her to create and submit a proposal? Or should the task force come up with a proposal?

v. Would it be possible to have progress already before the next meeting? Setting up the committee itself, including a call for the community to join, may take some time.

vi. Do we have a formal process on how to create a proposal? What is the difference between opening a discussion or creating an SVD-WG proposal?

### b. Introduction of a resolution to issue #147

i. Delayed until next time, the proposal is nearly done.

c. (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. We will make a proposal to solve this issue.

(end of time) - the chair ends the meeting

Not discussed (first on next meetings' agenda).

d. Introduction to a proposal for the nomenclature of positions upstream and downstream of transcripts

e. List of invalid DNA descriptions extracted from the HGVS nomenclature website.

## 6. AOB