Activity Proposal

Sequence variant description committee

Need

Stimulated by the HGVS, standards have been developed regarding the description of sequence variants, the so called *HGVS recommendations for the description of sequence variants* ("Mutation nomenclature"). Over time, these recommendations have developed into a standard that is used world-wide, esp. in the field of human genetics.
Questions regarding the current recommendations and requests for modifications and extensions directed at the HGVS are currently collected, answered and discussed by Johan den Dunnen, using the HGVS website (www.HGVS.org/mutnomen). Final decisions on specific topics are made a bit ad-hoc, based on community opinions, gathered through the website and discussed at meetings of the HGVS and HVP. What is highly desired is a committee of experts that meets regularly to discuss the topics raised and together make a more official decision.

Scope

The "***Sequence variant description committee***" should cover at least;

- the description of sequence variant(s) on DNA, RNA and protein level
- the curation of the HGVS website covering the *HGVS recommendations for the description of sequence variants* (www.HGVS.org/mutnomen)
- answer questions and clarify issues regarding the existing standards
- make proposals, ensure community discussion and finally decide on requests to extend the current recommendations where necessary/desired
- promote the use of the standards, a.o. by developing a web-based educational package

Plan of action

Assign a group of experts (5-7), covering all areas where sequence variant detection and description are used (genetics, biology). Include at least 1 (bio)informatics expert to ensure that the standards developed can be implemented in software.

Resources required

Time. Administrative support from HVP. A room to meet & discuss regularly, preferably connected to HGVS/HVP and/or ASHG/eSHG meetings. I consider development of a web-based educational package as essential to further promote the use of the existing standards. Hiring somebody to achieve this within a reasonable time-frame seems appropriate (estimated cost USD 5-10,000).

Expected deliverables

A document describing the current recommendations to be published in a peer-reviewed scientific journal. This paper should replace the last publication from 2000 in Human Mutation). A draft is ready for a final check by the committee, the paper will be published, on behalf of the members of HGVS.

Recommendation

People that are knowledgeable on the subject and responsive when asked, so obvious candidates for the Committee, include;

-Raymond Dalgleish (Leicester, UK) - gene variant database expert, active LSDB curator

-Peter Taschner (Leiden, NL) - gene variant database expert, (bio)informatics (Mutalyzer)

-Jean McGown-Jordan (Ottawa, Canada) - member ISN committee, asked to participate like JdD is member of the ISCN

Representatives from other stakeholders;

-representative from US commercial labs (Heidi Rehm?)

-representative from central repositories (NCBI/EBI - Donna Maglott or Fiona Cunningham)

-Johannes Zschocke, on a regular basis positively criticizing current recommendations