

RECOMMENDED SYSTEM STATUS



REVIEWER REPORT

System: HGVS Nomenclature
Proposer: Peter Taschner
Reviewer Report Date: 8 May, 2013

Application No: RS001
Date Proposed: 5/7/2012

BACKGROUND

Recommended System Status is an accreditation process designed to encourage the adoption of HVP Standards and Guidelines and to provide guidance around what systems, procedures and tools are of use to the wider community. The decision to award Recommended System status is made by the International Scientific Advisory Committee of the Human Variome Project. A key component of their assessment process for is the comments and recommendations obtained during peer review by members of the wider human genetics and genomics community.

Applications for Recommended System Status that have made it to the peer-review stage have already been determined to comply with all published HVP Standards and Guidelines by the International Scientific Advisory Committee based on advice provided by the ICO. The peer review stage seeks to provide the International Scientific Advisory Committee with guidance from members of the genomics community on the usability and reliability of the system.

Reviewers are asked to provide an opinion on:

- the compatibility of the system with other Recommended Systems, if any;
- the usability and relevance of the system;
- the reliability and robustness of the system;
- the availability of system documentation and training materials; and
- the extent to which the system is currently being utilised.

REVIEWER 1: DR RAYMOND DALGLEISH

There is absolutely no argument that the HGVS nomenclature system should be adopted an HVP standard. The nomenclature is well developed and supported and there is no competing alternative. Some may find it difficult at first to get to grips with the nomenclature but that's a quite separate education issue.

Perhaps the only issue is that the nomenclature web site could do with a revamp to make it easier to navigate.

Recommendation: Approve the application

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Competing or Conflicts of Interest: None Declared

REVIEWER 2

To me, the most significant problem is that most people still use amino acid changes despite the fact that almost 100% of molecular tests are done on DNA or RNA. We need to work together to enforce description at the nucleotide level. Nomenclature itself is fine, but I guess no matter which system, people need to be educated. All of our professionals need to work together to educate lay doctors and non-doctors. Related to this, I believe that perhaps a bigger pressing problem is that people still use colloquial names, especially gene products, proteins, a lot. For example, ER, PR, and HER2 for breast cancer. Correctly, these are ESR1, PGR, and ERBB2.

I am about to initiate task force within molecular pathology community, so that all pathology and laboratory reports adhere to HUGO (HGNC) gene nomenclature guidelines for describing genes and gene products including proteins. If all lab reports use standardized nomenclature, it will be a big step towards standardized nomenclature for its variation. I would appreciate support from HGVS and Human Variome Project, too.

Recommendation: Approve the application

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Competing or Conflicts of Interest: None Declared