RECOMMENDED SYSTEM STATUS

REVIEWER REPORT

System: Mutalyzer
Proposer: Peter Taschner
Reviewer Report Date: 2013-12-12

Application No: RS002
Date Proposed: 2012-07-05

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: BRYONY THOMPSON

I would definitely recommend approving Mutalyzer for recommended system status. I use Mutalyzer a huge amount to assist my research, whether it be converting to or checking my HGVS nomenclature. I find it easy to use and reliable. Its ability to do batch queries is particularly helpful. Mutalyzer makes my life a lot easier. I think it fulfils all the requirements stipulated.

Recommendation: Approve the application

Bryony Thompson
PhD Candidate
Molecular Cancer Epidemiology Lab
QIMR Berghofer Medical Research Institute

Competing or Conflicts of Interest: None Declared

REVIEWER 2 – JOHN-PAUL PLAZZER

Checks that variants are correctly described according to HGVS guidelines through Name Checker function (It has related functions that are not apprised here). It checks the nomenclature for correctness in relation to coding DNA reference sequence ("c."), and provides the predicted protein alteration ("p."). The program works for variants in potentially all genes and reference sequences (or user-defined sequences), including the Locus Reference Genomic sequences. The program works in intronic as well as exonic regions. The program works with common variant types, and compound variants. Mutalyzer is freely available online, user-friendly, well supported, and updated regularly. There is a web service to allow programmatic access. It can also process batches of variants in a file submission.

Recommendation: Approve the application

John-Paul Plazzer
Database Curator
International Society for Gastrointestinal Hereditary Tumours

Competing or Conflicts of Interest: None Declared

The Human Variome Project International Scientific Advisory Committee resolved to award Recommended System Status to Mutalyzer at their December 2013 meeting, on the proviso that the Mutalyzer development team "explicitly states where the HGVS Nomenclature is followed exactly, where exceptions are and which parts are not yet implemented." It was decided at this meeting that, given the positive nature of the two reports received from reviewers that a recommendation from a third reviewer was not necessary.