

RECOMMENDED SYSTEM STATUS



REVIEWER REPORT

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

Application No: RS003
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.

REIEWER 1

the compatibility of the system with other Recommended Systems, if any;

Since there are no other recommended systems this is not answerable.

the usability and relevance of the system;

It is very relevant to the HVP. It also has the flexibility to grow as recommendations are determined.

the reliability and robustness of the system;

We have been running an LOVD database for several years now and have experienced very few problems. It is flexible enough to meet the different needs of the LSDBs as well as the preferences of their curators, while providing a uniform look and feel to the users.

the availability of system documentation and training materials; and

There is sufficient documentation that comes with the code, and is easily accessible.

the extent to which the system is currently being utilised.

In searching LSDBs for the PhenCode project and contacting the curators of those databases, I have seen LOVD used frequently for new databases as well as an upgrade for some existing databases.

Recommendation: Approve the application

Competing or Conflicts of Interest: None Declared

REVIEWER 2

Disclaimer

The following comments are mainly related to LOVD version 2. These comments are based on a 3-years experience as an administrator of LOVD installations and on a 7-years experience of LSDBs curation process.

Review

LOVD is a web-based tool designed to store, manage and analyse genetic data. The main usage concerns the gathering of genetic variants related to one or several genes. Therefore LOVD is considered as a system to build Locus-Specific DataBases. In that field, LOVD is clearly the most widely used system.

LOVD has two characteristics which are in-line with the goals and objectives of the Human Variome Project : it is free and open-source. Its characteristics (PHP/MySQL) make it compatible with any common computer and installation and configuration requires only basic informatics knowledge. Moreover, the LOVD team provides hosting facilities. The database schema is robust and clever. Setting up a new gene database is also fast and easy and achieved in a few minutes.

LOVD provides several user levels, which are appropriate for the various tasks required by the system. The configurations and settings tabs are clear and provide sufficient functionalities. Several functions such as « Find and replace » are implemented to facilitate data management and have proven useful, but should be used with extreme caution. The possibility to empty and completely refill the database with new/modified data is excellent. There are also various possibilities of customization. Creating new columns, new custom link types, etc. is appreciable. The storage and management parts of the software are really impressive. The logging system directly available through the web interface is also complete.

However, some work could be done to improve the analysis part of the project. Indeed, if LOVD is really useful to find existing variants, meta-analysis must be done separately (i.e. you need to download the data and work on them using your own methods). True statistical methods could be implemented. However, this would be relevant only for harmonised data. That is why a deeper integration of mutalyzer is important. This point is being addressed in LOVD3.

Concerning the content of the native fields, automated integration of fields such as protein domains (using e.g. UNIPROT) would enrich the dataset. Improvements could be done concerning the web interface. Two main points could be mentioned. First, querying the database. Native implemented queries are poor. If you can query by simply typing on search fields, and by combining several fields you can achieve quite complex queries, demonstrating that the tool is powerful, this is not really suitable for every potential user. Therefore enrichment of the native possibilities such as, "How many pathogenic missense in domain X" (with access to the details) would be of great use. Specific web pages should be implemented.

The second point is related to the interface design. LOVD is not yet web 2.0. Visually, it looks like an Excel spread sheet. Clever usage of new web technologies could modernise a little bit the web design and dynamise the experience. For example, a page for user comments for each installation could be useful in some cases.

Finally, documentation is complete and clear. But using images, or videos, could make it more attractive.

Recommendation: Approve the application

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Competing or Conflicts of Interest: None Declared