

Diagnostic Variant Database Policy

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Introduction

Coordinated by the NBIC BioAssist program, several groups from Dutch University Medical Centers decided to work together to establish a nation-wide human genomic variant database, the Diagnostic Variant Database (DVD). The purpose is to share all genetic variants detected in the course of next-generation sequencing (NGS) applications between collaborators. This will help medical researchers to distinguish between functionally relevant and benign genetic variants. Therefore, researchers or clinicians can quickly narrow down to those variants that are the potential genetic cause of a particular disease. DVD also allows users to contact the researcher who originally identified a particular genetic mutation. This makes DVD a platform where new research collaboration can be established.

Available Data

Almost all data in DVD is from patient studies and thus may contain sensitive information. Thus, DVD is designed to prevent potential misuse of this information. Access to DVD data is only permitted to registered user groups from Dutch Universities or Medical Centers.

The genomic variants stored in DVD follow the following criteria:

- Only human genomic variants
- Large variants (>200 bps) are not supported
- GRCh37 / hg19 is used as the reference genome. For the mitochondrial reference we use the revised Cambridge version.
- Coverage information is required. (This is important because we do not only need to know which variants have been seen, but also which variants could have been seen. By storing coverage information, we can deduce this.)

Data Access Interface

The DVD interface is simple with just one annotation service. Through its SOAP-based service endpoint, registered users can upload variants in VCF files. DVD will then annotate those variants and send the annotation report back.

Automatic Data Sharing Policy

As a start, four founding member groups (LUMC, UMCN, UMCU, UMCG) kindly contribute a set of variant files to bootstrap DVD. While in operation, the number of stored variants in DVD is growing thanks to an automatic data sharing design:

- After the annotation is performed, all new variant data will be stored in DVD automatically. Thus, future annotation requests can benefit from the growing variant knowledge base of DVD.

Quality Control

To benefit from the growing knowledge base, re-annotation of a previous uploaded VCF file is possible. A built-in script checks the novelty of VCF files and make sure only variants from new files will be added to DVD. Additional database clean-up scripts are also in place to remove wrongly introduced variant set as the result of mistakes in the VCF files.

Data Ownership

Data in DVD is owned collectively by all DVD user groups. Each sample in DVD belongs to the user group who originally uploads it in the form of VCF files. The stored variants are only stored in DVD and are maintained by the DVD administration team. Any further non-maintenance related copying or dissemination of these raw variants in any form is not permitted unless their owner user group explicitly agrees.

Advisory Committee

The founding member groups and their lead PIs form the advisory committee. This committee can provide strategic guidelines in DVD regarding content, interface, membership, hosting.

Current members:

- Johan den Dunnen (LUMC)
- Jeroen Laros (LUMC)
- Joris Veltman (UMCN)
- Morris Swertz (UMCG)
- Edwin Cuppen (Hubrecht/UMCU)
- Victor Guryev (Hubrecht/UMCU)

DVD Administration Team

The DVD administration team consists of main developers of the DVD system and committed members from user groups. This team runs the day to day practicality of DVD including server maintenance, security updates, data backups.

Current members:

- Jeroen Laros (LUMC)
- Christian Gilissen (UMCN)
- Robert Wagner (UMCG)
- Victor Guryev (Hubrecht/UMCU)
- David van Enckevort (NBIC)
- Leon Mei (NBIC)

User Management

Acceptance of new users from Dutch University Medical Centers can be decided by the DVD administration team. New user requests from other organizations are processed by the advisory committee.

Disclaimer

The information in the Diagnostic Variant Database and the database rights are (jointly) owned by the respective partners who provided the information and created the database ('Owners'). Users are not granted any rights other than which are granted below.

Users are allowed to access the database and use the data for non-commercial research. It is expressly prohibited to extract and/or re-utilize the whole of or a substantial part of the contents of the database.

The DVD and its contents is a research tool and not intended as a diagnostic tool, medical advice or treatment method. Users use the information provided at their own risk.

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