Resource Summary Report

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cafe variome

RRID:SCR_006162
Type: Tool

Proper Citation

cafe variome (RRID:SCR_006162)

Resource Information

URL: http://www.cafevariome.org/

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Description: Clearinghouse and exchange portal for gene variant (mutation) data produced by diagnostics laboratories, offering users a portal through which to announce, discover and acquire a comprehensive listing of observed neutral and disease-causing gene variants in patients and unaffected individuals. Cafe Variome is not a ""database"" for the hosting/display/release of data, but a shop window for finding data. As such, it holds only core info for each record, and uses this merely to enable holistic searching across resources. Diagnostics laboratories routinely assess DNA samples from patients with various inherited disorders, and so produce a great wealth of data on the genetic basis of disease. Unfortunately, those data are not usually shared with others. To address this gross deficiency, a novel system has been developed that aims to facilitate the automated transfer of diagnostic laboratory data to the wider community, via an internet based Cafe for routinely exchanging genetic variation data. The flow of research data concerning the genetic basis of health and disease is critical to understanding and developing treatments for a range of genetic diseases. Overall, the project aims to lower the barriers and provide incentives for a willing community to share data, and thereby facilitate the broader exploitation of diagnostic laboratory data. Cafe Variome aims to address the above data flow problems by: # Minimizing the effort required to publish variant data # Ensuring attribution for data creators working in diagnostic laboratories Key elements of the project strategy are: * Data publication will be automated by endowing standard analysis tools used by laboratories with an online data submission function. Submissions will be received by a central Internet depot, which will serve as a place where published datasets are advertised, and subsequently discovered by diverse 3rd parties. * Each dataset will be unambiguously linked with the data submitter""s identity, and systems devised to facilitate citation of published variant datasets so they can be cited in the literature. Data creators will thus be credited for their contributions. Data submitters can use Cafe Variome to simply announce or publicize their data to the world. To

enable this, only core, non-identifiable data is submitted to the central repository, enabling users to search and discover records of interest in the source repository. The data are not automatically handed on to the user (unless intended by the submitters). Hence, the concept is used to deal with the challenge of maximally sharing data whilst fully respecting ethico-legal considerations.

Abbreviations: Cafe Variome

Resource Type: service resource, data set, data or information resource, storage service resource, data repository

Keywords: phenotype, gene variant, mutation, gene, normal, disease

Related Condition: Diseased, Healthy

Funding: European Union FP7/2007-2013- the GEN2PHEN project

Availability: Open access, Restricted access and Linked access

Resource Name: cafe variome

Resource ID: SCR_006162

Alternate IDs: nlx_151664

Record Creation Time: 20220129T080234+0000

Record Last Update: 20250519T203433+0000

Ratings and Alerts

No rating or validation information has been found for cafe variome.

No alerts have been found for cafe variome.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 11 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

Doolan BJ, et al. (2022) A clinician's guide to omics resources in dermatology. Clinical and experimental dermatology, 47(5), 858.

Pereira A, et al. (2022) Semantic Data Visualisation for Biomedical Database Catalogues. Healthcare (Basel, Switzerland), 10(11).

Townend GS, et al. (2018) MECP2 variation in Rett syndrome-An overview of current coverage of genetic and phenotype data within existing databases. Human mutation, 39(7), 914.

Viennas E, et al. (2017) Expanded national database collection and data coverage in the FINDbase worldwide database for clinically relevant genomic variation allele frequencies. Nucleic acids research, 45(D1), D846.

Lelieveld SH, et al. (2016) Novel bioinformatic developments for exome sequencing. Human genetics, 135(6), 603.

Smith TD, et al. (2015) Standard development at the Human Variome Project. Database : the journal of biological databases and curation, 2015.

Vihinen M, et al. (2014) Variation Ontology for annotation of variation effects and mechanisms. Genome research, 24(2), 356.

Porter LF, et al. (2014) Personalized ophthalmology. Clinical genetics, 86(1), 1.

Papadopoulos P, et al. (2014) Developments in FINDbase worldwide database for clinically relevant genomic variation allele frequencies. Nucleic acids research, 42(Database issue), D1020.

Sahajpal R, et al. (2014) HGV&TB: a comprehensive online resource on human genes and genetic variants associated with tuberculosis. Database : the journal of biological databases and curation, 2014, bau112.

van Schaik TA, et al. (2014) The need to redefine genomic data sharing: A focus on data accessibility. Applied & translational genomics, 3(4), 100.