Connecting diagnostic labs: Cafe Variome and DNA sequencing software

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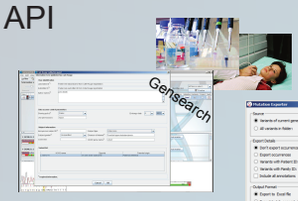
There is a considerable need and desire for diagnostic labs to be able to check each other's databases for the presence of mutations they observe in their own patients. This is countered by the understandable reluctance and impracticability of sharing the content of each group's database with other labs, or indeed the world.

Cafe Variome core concept

Public API

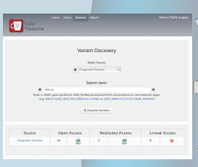


Laboratory personnel can use "Cafe Variome-enabled" mutation interpretation software to push variant data to Cafe Variome

Other sources of variants are advertised and made discoverable via Cafe Variome

Cafe Variome

Open discovery of the existence of data through Cafe Variome Portal



End users can seek access to data under one of three models:

openAccess

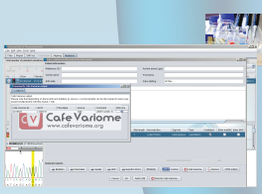
Unrestricted access for anyone

restrictedAccess

Either pre-approved access for specific groups or access requested via automated email to the data owner

linkedAccess

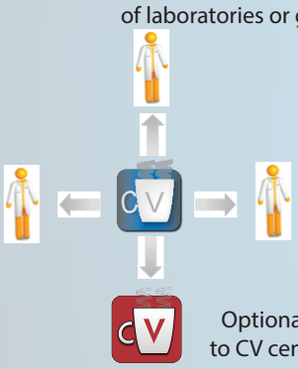
Linking to source of variant to obtain full record



Lab personel can use "Cafe Variome-enabled" software to query directly Cafe Variome for variants detected by the sofware

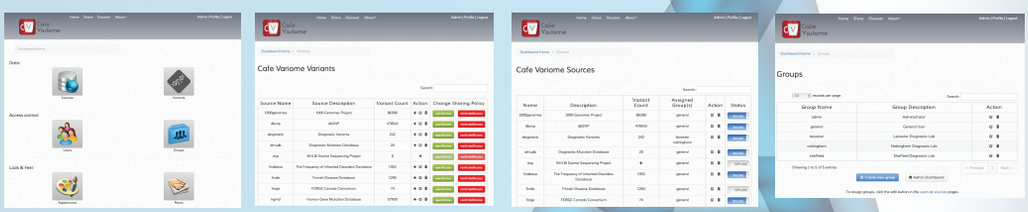
Cafe Variome for Rare Disease Networks

Use Cafe Variome to share variants between private networks of laboratories or groups.



Optional: connect your private CV to CV central to advertise your data.

Intuitive admin interface and sharing dashboard gives you fine grained access control over variants, sources, users and groups

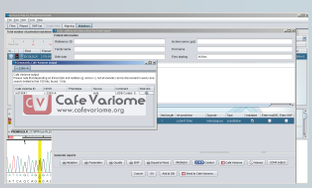


Installs can be hosted by the Cafe Variome team or on your own servers.

API use case examples

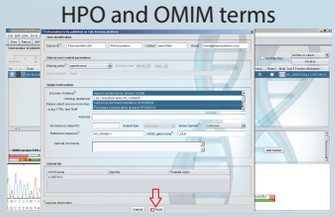
Capillary DNA sequencing

Checking a Cafe Variome instance for information on variants detected in a patient, using Gensearch.



Query simultaneously all sources available in the public instance (dbSNP, HGMD, LSDBs,...) through federation, query own private data as well.

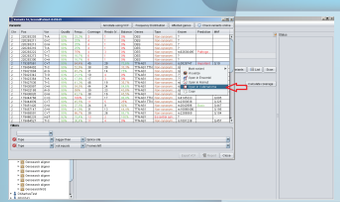
Publish the variants on your local Cafe Variome instance or on the public instance. Annotate using HPO and OMIM terms



You control access rights as well as amount of details to be published. For fine grained access control use the CV control panels.

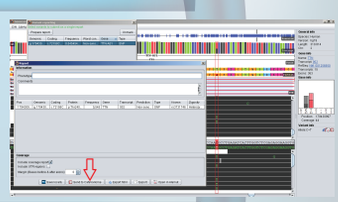
NGS sequencing

Looking up a Cafe Variome instance for information on variants detected in a patient, using GensearchNGS.



Opens the Cafe Variome web page on the genomic position selected, presenting all available sources.

Publish the variants on your local Cafe Variome instance or on the public instance.



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www.CafeVariome.org

PhenoSystems SA
www.phenosystems.com

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