First Steps with GeneTalk

What is GeneTalk?

GeneTalk combines a web-based tool for filtering and analyzing human sequence variants and a professional online network for geneticists worldwide.
Clinicians and scientists looking for information about specific sequence variants can find other users with expertise for a particular variant.

Filter and prioritize sequence variants with highly effective algorithms that speed up your analysis

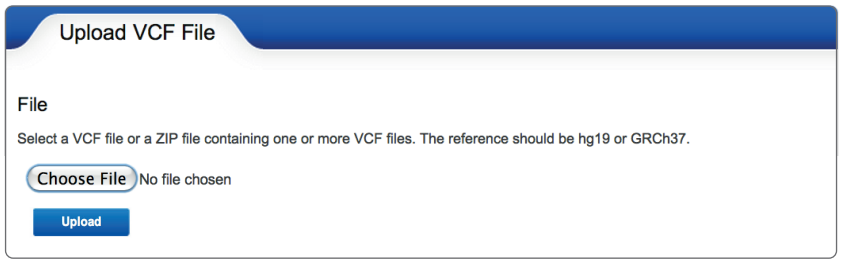
Annotate sequence variants and find out what other GeneTalk users say about specific mutations

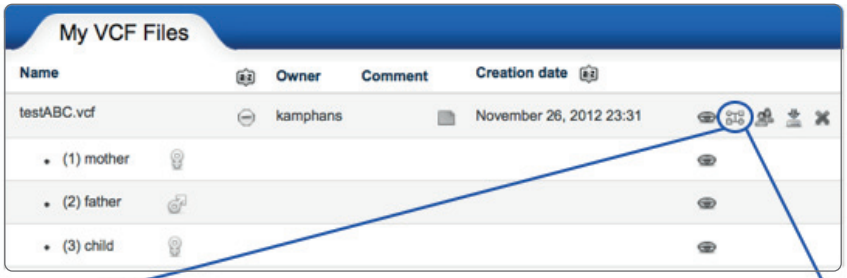
Join the large professional online network for geneticists worldwide and discuss unclassified variants

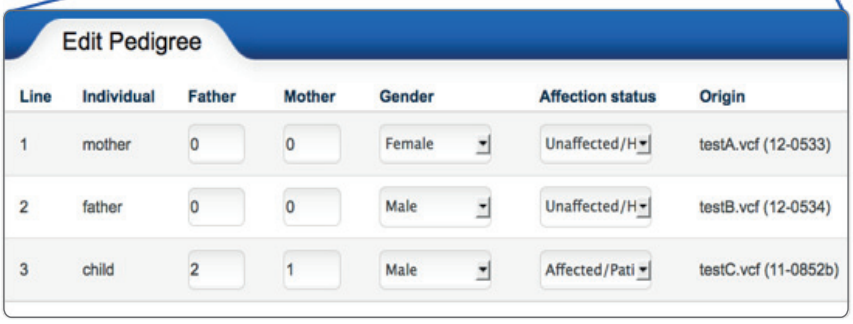
How to use Genetalk

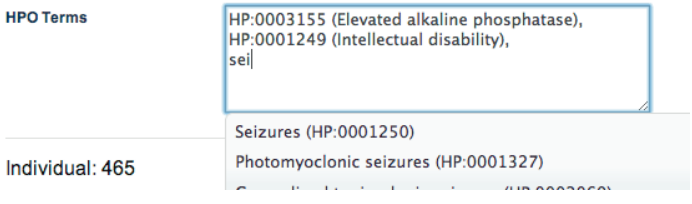
① **Upload your sequence variants** in VCF format of single or multiple samples (*Manage Files→Upload VCF*). Make sure the hg19 or GRCh37 reference is used.
Uploaded files are preprocessed and listed in the file manager (*Manage Files* → *List*). Individuals in a multiple VCF file are shown after clicking the ⊕ icon. You can view the file 👁 and share it with other users 👥

② **Edit the pedigree and phenotype information** of the individuals in your VCF file by clicking the icon. Phenotypic features can be specified by terms of the *Human Phenotype Ontology* (HPO).
These information allow for powerful filtering of variants, for example,, by mode of inheritance (dominant, recessive homozygous, recessive compound heterozygous).









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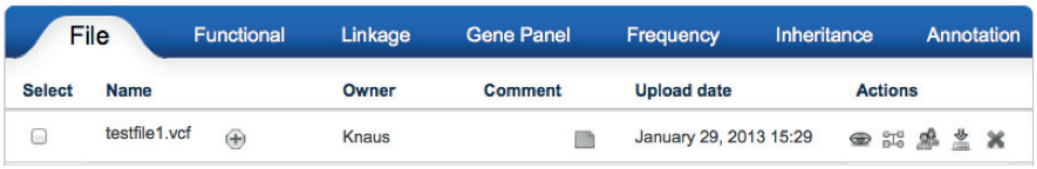
③ Filter Sequence Variants

Click on *Filter VCF* in the main menu.

The filter settings menu is organized using tabs in the top row. Choose one or more VCF files for filtering with the checkboxes in the *File* tab.

The other tabs provide several options for reducing the number of variants in your VCF file(s).

Select your filter settings and click the *Filter* button.



Functional Filter: Reduce to variants that have an effect on the protein level.

Linkage Filter and Gene Panel Filter: Restrict the variant list to a set of genes or to a linkage interval.

Frequency Filter: Remove all variants with genotypes that occur more often in the background population than an adjustable cutoff. (Data from the 1000 genomes and 5000 exome sequencing project are used as reference.)

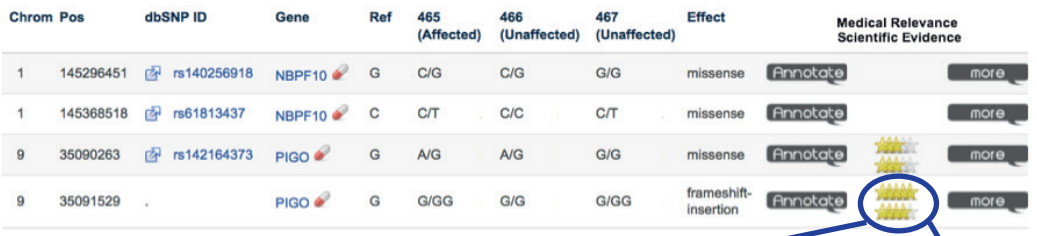
Inheritance Filter: Restrict analysis to genotypes that fit to a certain mode of inheritance.

Annotation Filter: Filter out variants based on ratings for medical relevance and scientific evidence.

④ View Sequence Variants

The filtered VCF file contains significantly fewer sequence variants.

Click the *Annotate* button to provide an annotation for a variant. The *More* button displays a popup with detailed variant information.

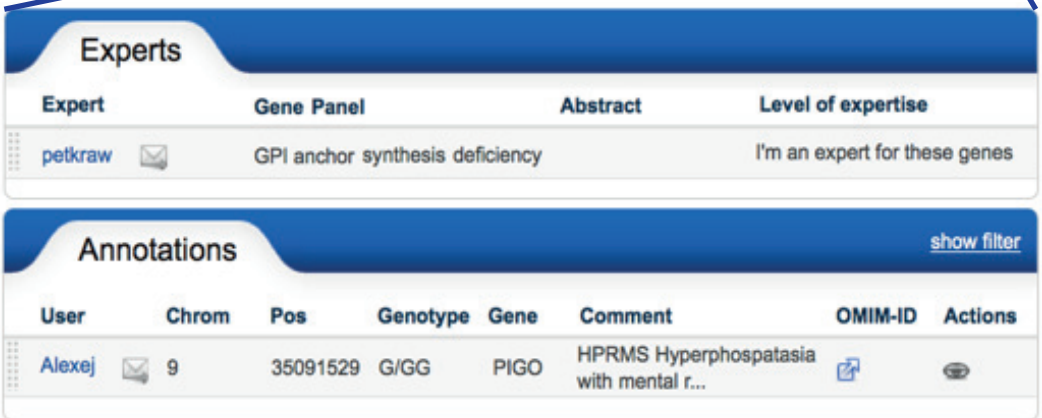


⑤ Find Annotations, Experts and Patients

The annotations column indicates if for the position exists at least one annotation (with the shown votings for *medical relevance* and *scientific evidence*), an expert, or another patient. Click the stars to open the list of annotations, experts, and patients.


Expert


Patient



For position 35091529 in chromosome 9, there is an expert, *petkraw*, in GPI anchor deficiency. You can contacted this user for further discussion. Additionally, the user *Alexej* has left an annotation for this position in PIGO, rating an insertion as disease causing.

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