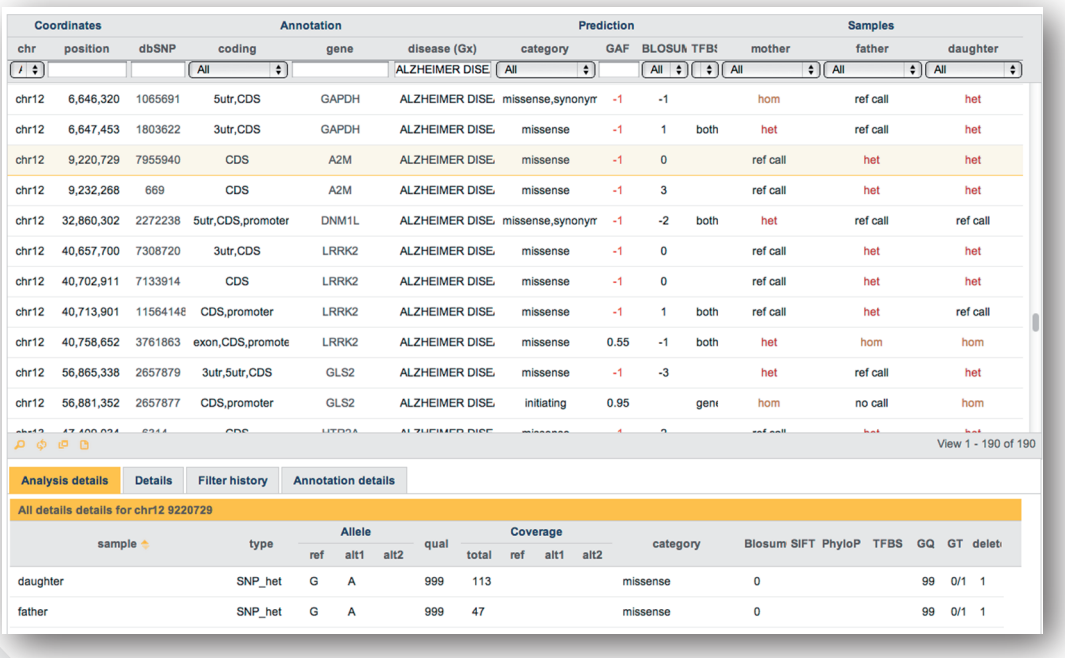
genomatix



GeneGrid: Finding the relevant variants.

Pre-release available now!!

Genomic variants like single nucleotide polymorphisms (SNPs) or small insertions / deletions (InDels) are of major interest to biologists and clinicians alike.

Their impact ranges from determining your eye color to influencing response to medication. They can cause cardiovascular or neurodegenerative diseases, induce cancer or on the other hand trigger resistance to HIV infection. Identifying causal variants is therefore crucial for the understanding of molecular mechanisms and diagnostics of rare or common diseases.

With today's Next Generation Sequencing (NGS) technology it is possible to detect the millions of variants within an individual genome through a single experiment.

One question remains, though:

Which are the relevant ones?

Finding the needle in the haystack.

The Genomatix **GeneGrid** technology enables you to quickly reduce millions of variants to the few or even the single relevant one(s).

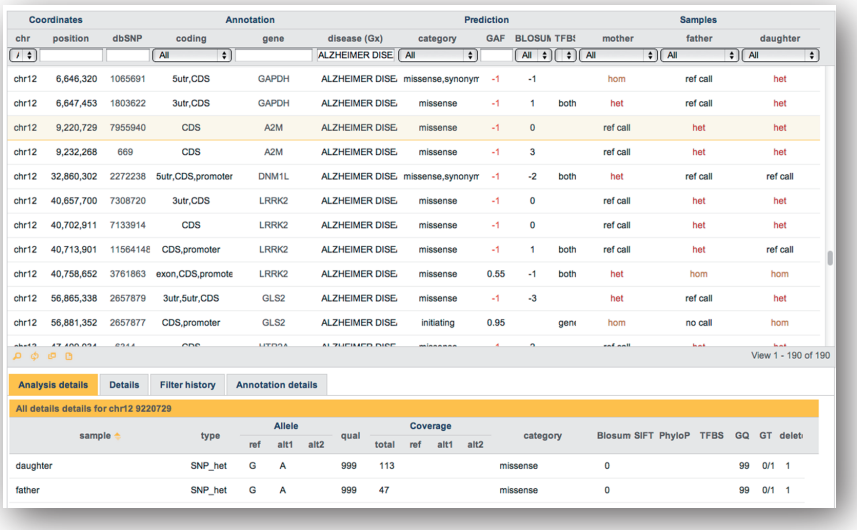
All known & novel SNPs in your results can be annotated using our extensive genome annotation. With the intuitive interface of the software you can filter the list for those variants of interest to you, perform trio analyses, compare case and control sets (using multiple samples) or identify somatic SNPs within minutes.

You can filter by:

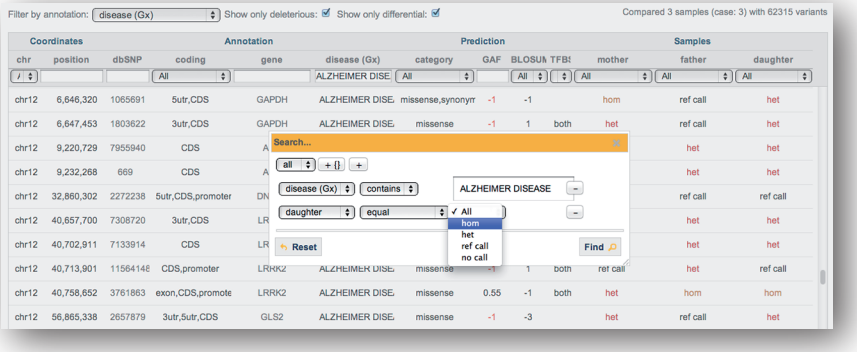
- effects on the protein (mis-,non-sense, etc.)
- amino acid substitution, conservation and the 1000 Genome Project's global allele frequency scores
- dbSNP identifiers
- associations with diseases (e.g. from the COSMIC cancer database), tissues, pathways, transcription factor binding sites and small molecules

Subsequently you can export the lists and associations. You can also switch to the Genomatix Pathway System to look at networks of the affected genes or view the genomic location of a SNP in our Genome Browser.

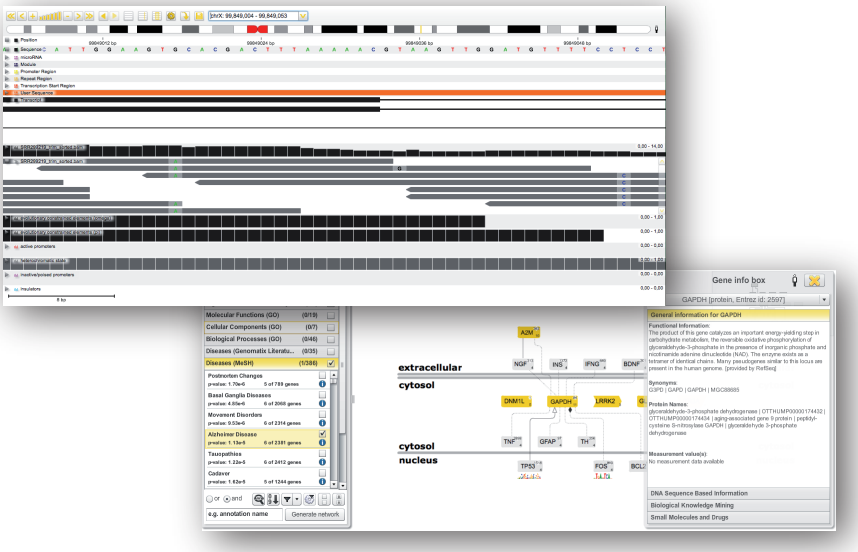
Here are some sample screenshots of the GeneGrid interface:



A trio analysis based on 1000 Genome Project data from a family of father, mother and daughter. The upper part contains the list of filtered deleterious and differential SNPs together with genome allele frequency (GAF) and amino acid substitution (BLOSUM62) scores and annotation. The GAF score has been set to a maximum of 0.01 to identify rare variants and using the disease filter only results associated with Alzheimer's disease are displayed. This leaves only 132 entries from an original list of more than 60,000. The lower part shows the analysis details for the line selected in the list, including the alleles, depth of coverage and amino acid substitution / conservation scores (BLOSUM62, SIFT and PhyloP).



Quick and simple filtering of variants is available, along with an advanced query interface for building more sophisticated filters using Boolean logic.



Direct integration with our Genome Browser allows for the visualization of a variant in genomic context as well as a detailed alignment view down to a single nucleotide. Genes associated with variants can instantly be used in the Genomatix Pathway System (GePS) e.g. for generation of networks, association with pathways or identification of regulators or targets.

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