

Coordinates			Annotation			Prediction			Samples			
chr	position	dbSNP	coding	gene	disease (Gx)	category	GAF	BLOSUM	TFBS	mother	father	daughter
chr12	6,646,320	1065691	5utr,CDS	GAPDH	ALZHEIMER DISE.	missense,synonymr	-1	-1		hom	ref call	het
chr12	6,647,453	1803622	3utr,CDS	GAPDH	ALZHEIMER DISE.	missense	-1	1	both	het	ref call	het
chr12	9,220,729	7955940	CDS	A2M	ALZHEIMER DISE.	missense	-1	0		ref call	het	het
chr12	9,232,268	669	CDS	A2M	ALZHEIMER DISE.	missense	-1	3		ref call	het	het
chr12	32,860,302	2272238	5utr,CDS,promoter	DNM1L	ALZHEIMER DISE.	missense,synonymr	-1	-2	both	het	ref call	ref call
chr12	40,657,700	7308720	3utr,CDS	LRRK2	ALZHEIMER DISE.	missense	-1	0		ref call	het	het
chr12	40,702,911	7133914	CDS	LRRK2	ALZHEIMER DISE.	missense	-1	0		ref call	het	het
chr12	40,713,901	11564148	CDS,promoter	LRRK2	ALZHEIMER DISE.	missense	-1	1	both	ref call	het	ref call
chr12	40,758,652	3761863	exon,CDS,promote	LRRK2	ALZHEIMER DISE.	missense	0.55	-1	both	het	hom	hom
chr12	56,865,338	2657879	3utr,5utr,CDS	GLS2	ALZHEIMER DISE.	missense	-1	-3		het	ref call	het
chr12	56,881,352	2657877	CDS,promoter	GLS2	ALZHEIMER DISE.	initiating	0.95		gen	hom	no call	hom

All details details for chr12 9220729																
sample	type	Allele			qual	Coverage			category	Biosum	SIFT	PhyloP	TFBS	GQ	GT	delet
		ref	alt1	alt2		total	ref	alt1								
daughter	SNP_het	G	A		999	113			missense	0				99	0/1	1
father	SNP_het	G	A		999	47			missense	0				99	0/1	1

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:

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chr	position	dbSNP	coding	gene	disease (Gx)	category	GAF	BLOSUM TFBt	mother	father	daughter
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chr12	9,232,268	669	CDS	A2M	ALZHEIMER DISE	missense	-1	3	ref call	het	het
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All details details for chr12 9220729													
sample	type	Allele	qual	Coverage		category	Blosum SIFT	PhyloP	TFBS	GQ	GT	delet	
				ref	alt1								alt2
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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).

Filter by annotation: **disease (Gx)** Show only deleterious: Show only differential: Compared 3 samples (case: 3) with 62315 variants

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chr12	40,702,911	7133914	CDS	LR	ALZHEIMER DISE	missense	-1	0	ref call	het	het
chr12	40,713,901	11564146	CDS,promoter	LRRK2	ALZHEIMER DISE	missense	-1	1	both	ref call	het
chr12	40,758,652	3761863	exon,CDS,promote	LRRK2	ALZHEIMER DISE	missense	0.55	-1	both	het	hom
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<http://linkedin.com/company/genomatix>

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

The screenshot displays a genomic alignment view with a detailed gene information box for GAPDH. The gene info box includes:

- General information for GAPDH:** Functional information, Protein Name, Synonyms, and Protein Name.
- Extracellular:** A diagram showing the extracellular domain of GAPDH with residues ASP1, ASN1, PHE1, and SER1.
- Cytosol:** A diagram showing the cytosolic domain of GAPDH with residues THR1, THR2, THR3, THR4, THR5, THR6, THR7, THR8, THR9, THR10, THR11, THR12, THR13, THR14, THR15, THR16, THR17, THR18, THR19, THR20, THR21, THR22, THR23, THR24, THR25, THR26, THR27, THR28, THR29, THR30, THR31, THR32, THR33, THR34, THR35, THR36, THR37, THR38, THR39, THR40, THR41, THR42, THR43, THR44, THR45, THR46, THR47, THR48, THR49, THR50, THR51, THR52, THR53, THR54, THR55, THR56, THR57, THR58, THR59, THR60, THR61, THR62, THR63, THR64, THR65, THR66, THR67, THR68, THR69, THR70, THR71, THR72, THR73, THR74, THR75, THR76, THR77, THR78, THR79, THR80, THR81, THR82, THR83, THR84, THR85, THR86, THR87, THR88, THR89, THR90, THR91, THR92, THR93, THR94, THR95, THR96, THR97, THR98, THR99, THR100.
- Nucleus:** A diagram showing the nuclear domain of GAPDH with residues THR1, THR2, THR3, THR4, THR5, THR6, THR7, THR8, THR9, THR10, THR11, THR12, THR13, THR14, THR15, THR16, THR17, THR18, THR19, THR20, THR21, THR22, THR23, THR24, THR25, THR26, THR27, THR28, THR29, THR30, THR31, THR32, THR33, THR34, THR35, THR36, THR37, THR38, THR39, THR40, THR41, THR42, THR43, THR44, THR45, THR46, THR47, THR48, THR49, THR50, THR51, THR52, THR53, THR54, THR55, THR56, THR57, THR58, THR59, THR60, THR61, THR62, THR63, THR64, THR65, THR66, THR67, THR68, THR69, THR70, THR71, THR72, THR73, THR74, THR75, THR76, THR77, THR78, THR79, THR80, THR81, THR82, THR83, THR84, THR85, THR86, THR87, THR88, THR89, THR90, THR91, THR92, THR93, THR94, THR95, THR96, THR97, THR98, THR99, THR100.
- Measurements:** Measurements (e.g., no measurement data available).
- DNA Sequence Based Information:** Biological Knowledge Mining, Small Molecules and Drugs.

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.