GeneticistAssistant™ NGS Interpretative Workbench

Features:

Variant Database

- Historical Database
- Pathogenicity Calling Information
- Pathogenicity Call Supporting Information
- Linkage to External Databases
- Automated Quality Control
- Accessibility
- User Management, Audit Trail, Access Control

Cool Tools

- Custom Report Builder
- Customer Web Portal
- Automated Informatics Pipeline
- Customizable Workflow Builder
- Sample Comparison
- Custom Filtering
- Artifact Flagging
- Process Quality Control
- Positive Control Verification
- Automatic BED file builder with regions of clinical significance

Developed in collaboration with Mayo Clinic



GeneticistAssistant[™]





Efficient...Saves Time & Resources, Controls...Real-time Administration & Reporting, For...Disease Panels and Whole Exome Sequencing data, Compatible...with data from all NGS Systems

Developed in collaboration with the Laboratory Medicine, Information Technology and Health Science Research departments of Mayo Clinic, Geneticist Assistant NGS Interpretative Workbench is a unique tool for the management, control, visualization, functional interpretation and historical knowledge base of next generation sequencing Whole Exome data or Disease Panels targeted at specific genes for the purpose of identifying potentially pathogenic variants associated with specific conditions such as hereditary colon cancer and others.

Geneticist Assistant is compatible with data processed from all leading next generation sequencing platforms including lon Torrent, Illumina and Roche platforms. The program accepts standardized BAM and VCF files, and includes information from the following sources:

Functional Prediction information: SIFT, PolyPhen-2, LRT, MutationTaster, FATHMM, CADD & MutationAssessor

Disease association: ClinVar, OMIM, CIViC & COSMIC*

Conservation scores: phyloP, GERP++, phastCons & SiPhy

Population frequencies: 1000 Genomes, Exome Variant Server, and ExAC

Additionally, information from proprietary databases such as **Alamut** and LOVD (Leiden Open Variation Database) are easily accessible through embedded links. Information from other publicly available databases are easily imported into the workbench.

The new administration function provides a real-time tracking of current statuses; historical information; automated email notifications within a completely customizable workflow built to model your actual activities.

Unique tools include **Custom Filtering**, **Patient Comparison**, i.e. **Trio Comparison**, **CAP Validation Assistance**, **automated BED file builder** which automatically highlights areas of clinical significance, **Positive Control Verification**, and in conjunction with NextGENe software can form a completely **automated informatics pipeline**.

*Requires separate license

Historical Database Development

Geneticist Assistant NGS Interpretative Workbench records variant pathogenicity determination on all found variants, eliminating time consuming duplication of researching the variant, thus speeding diagnosis while reducing costs. As the database is used the number of variants requiring pathogenicity calling is quickly reduced to a few novel variants.

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Historical information on every found variant is recorded and available for instant recall. Additionally prior pathogenicity determination is logged by specific disease panel and globally for all disease panels. The variant review tab provides previously determined variant type, pathogenicity, variant frequency, HGVS Nomenclature, times observed, number of times observed in disease panel and panel group.

Use of the workbench will quickly reduce unnecessary pathogenicity research duplication, speeding diagnoses and reducing costs.

Pathogenicity Calling Information

Geneticist Assistant NGS Interpretative Workbench provides Variant Interpretation, Functional Prediction, Conservation Scores and Disease Associations on each found variant from over 17 sources providing the information in a single view. **Once a call has been** made and confirmed, the research is stored in the database and applied to future recurrences of the variant either in the same disease panel or in any other panel, significantly reducing time and effort on future iterations of the variant in future analyses.



Geneticist Assistant Workbench provides a complete samples which exhibited variant are also detailed.

Sources included:	Variant Interpretation: dbSNP Exome Variant Server
	Functional Prediction: SIFT
	PolyPhen-2 LRT

LRT MutationTaster MutationAssessor FATHMM CADD



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Geneticist Assistant Workbench provides a complete overview of information regarding variant pathogenicity in one detailed view. Prior

Conservation Scores: phyloP phastCons GERP++ SiPhy Population frequencies: 1000 Genomes Exome Variant Server ExAC

Disease Association: COSMIC* ClinVar & OMIM CIViC Alamut* LOVD (Leiden Open Variation Database) And others

*Requires separate license

Pathogenicity Call Supporting Information

Supporting information for a pathogenicity call is easily added to the database by a right mouse click in the variant tab. Data from any source such as dbSNP can be added for future recall.

Geneticist Assistant NGS Interpretative Workbench also includes a "mini web browser" which allows a user to search and link scientific information from any web source such as NCBI in support of the pathogenicity call which can be recalled at any time by authorized users. PubMed abstracts can be automatically downloaded into the workbench.

Add Supporting Info	
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O The Development of the second of the secon	L. Pice for. Mik Age 12:710/refw184; dog 10.575/rescal_aves.094516, doklars.m. 2015.
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A simple right mouse click enters information	n and PubMed abstracts can be automatically downloaded into

Linkage to External Databases

pathogenicity call into Geneticist Assistant

Interpretative Workbench.

Retrieving further information from external proprietary tools such as Alamut, UCSC Genome Browser, or the LOVD database is a simple click away. (Alamut requires a license)

ID	Chr : ChrPos *	Rs	Pathogenicity	Pathogenicit	y Status	Gene		HGVS Coding
28	3:37056045	rs182733777	Unassigned		Variant De	tails		90+10A>G
29	3:37081751	rs267607840	Unassigned		Show Vari	ants Filte	ered by Panel	.633A>G
6	3:37083740	rs9876116	Benign		Edit Variar	nt		.668-19A>G
13	14:75505016	rs175075	Benign		External		,	Minu In Alamut
14	14:75513883	rs175081	Benign		Evnort			View In Addition
31	14:75514489	rs28756986	Unassigned		Reports			View in OCSC
15	17:7579472	rs1042522	Deletenous		Logit Cal			View PubMed Abstract
1443	17:7579669	rs17878362	Unassigned		LOAD COIL	imns		6+32_96+47delGGGCTGGGGA
18	17:63554591	rs2240308	Likely Deleterious	Confirmed		AXIN2	NM_004655.3:4	:.148C>T
19	18:48584856	rs386387676	Likely Benign			SMAD4	NM_005359.5:	.904+45_904+46insTT

Alamut licensees can quickly retrieve information without error prone and tedious retyping by simply selecting variant of interest and clicking on the drop down menu.

LOVD	Data:									
Symbol	ID	Position mRNA	Position Genomic	Variant DNA	Variant DBID	Times Reported	Chromosome	Allele	Affects Function (Reported)	Affects Function (Concluded)
IVD	16587	NM_002225.3:c.1276_1278	chr15:40710457_40710459	c.(1276_1278del)	IVD_000013	1	N/A	Unknown	Effect unknown	Effect unknown

Retrieving information from the LOVD database is a simple linked operation.

Automated Quality Control

Geneticist Assistant NGS Interpretative Workbench automatically monitors coverage depth, flagging regions to the base level that do not meet your pre-set requirements. The software will track over time the amplicon or regions' performance, providing feedback on the sequence performance, which may alert you to areas that require performance improvement.

Directories Quality Control	Pathopericity Calers	Sab Preferences	Variant Preferences Alama
Minimum Coverlage:			50
Average Coverage:			800
Percent Cavered(%)			100.00

Report Name	Chron Start - End - To Covered	Average Correrage	Minimum Conerage	Salat	Avwage % Covered	Average Average Coverage	It in age Minimum Converage	Facund	Persed Percent	Palet.	7,04
M942.NM_000251	2:47630301 - 47630571 200%	403.03	138	Pessed+	200%	353	131	10	90,9091%	1	11
MSH2:NM,000251	2:47635500-47635724-100%	772.87	354	Passed+	100%	516	229	10	00-9090 %	1	11
MSH2:NM_000251	2:41637303-43637543 300%	674.03	369	Passed *	200%	525	143	10	90,0001%	4	11
MSH2.NM_000251	2 47639523 - 47639729 100%	777.8	393	Pessed+	100%	579	312	10	#2006.00	1	11
MSH0:HM.000251	2: 47641778 - 47641587 100%	£36.61	3.27	Passed+	100%	450	164	10	90.9090%	1	53
MEH2-NM_000251	2:47643405 - 47643508 100%	867.31	485	Passed+	100%	662	367	10	90,9091%	1	11
MEH2.NM. 500251	2: 47656851 - 47657110 100%	894.67	431	Passed+	100%	757	347	10	90.9092%	1	11
MEH2:NIM_000251	2:47672657 - 47672625 100%	560.26	343	Passed+	100%	436	284	10	90.3090 %	1	11
MSH2 NM4 000251	2: 47690540 - 47690323 500%	718.4	458	Passed*	100%	495	75.3	10	90.3091%	1	11
MEH2-NM 000252	2: 47693767 - 47693977 100%	565.8	13	faint .	100%	463	81 I	4	54.5655%	5	22
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Quality data is presented for both the current sample and a complete history of analysis of all samples for a disease panel. Metrics provided include Minimum Coverage, Average Coverage, % Coverage Across Region and Pass/Fail Status of current run. Historical data includes average coverage of all runs, average percent coverage, absolute Pass/Fail counts, total samples for the region and passed percentage. Sequencing that often fails is easily reviewed, allowing user to determine and correct cause of sequencing failures.

10-	Name	Run Date Time	Add Date Time	Run	Panel	PanelGroup	Reference	# Regione	# Regions Passed	Patient External ID	Status	Missed Clinical Variants
8	800466.variants.filter	5/14/2014 11:05:33 AM	5/14/2014 11:18:38 AM	Demo	DLMP	default	Human 37	154	151	XV2709	Complete *	Yes
9	800402.variants.filter	5/14/2014 11:05:33 AM	5/14/2014 11:20:03 AM	Demo	DUMP	default	Human 37	154	150	A9C123	New *	Ves
20	800451.variants.filter	5/14/2014 11:05:33 AM	5/14/2014 11:21:26 AM	Demo	DLMP.	default	Human 37	154	152	8C-13-15487	OC.Passed +	Ves
11	800474.variants.filter	5/14/2014 11:05:33 AM	5/14/2014 11:22:46 AM	Demo	DUMP	default	Human 37	154	159	BC-13-20683	Reviewed *	Ves
12	272305 variants filter	5/14/2014 11:05:33 AM	5/14/2014 11:24:08 AM	Demo	DLMP	default	Human 37	154	4	BC-13-20476	New -	Yes

Importantly, Geneticist Assistant NGS Interpretative Workbench, monitors areas of clinical significance providing a quick review of missed clinical variants as determined by the ClinVar database information.

Accessibility

Geneticist Assistant NGS Interpretative Workbench is comprised of a local installed database, either Linux or Windows[®], and a client Windows program which provides the easy-to-use, graphical user interface. All data is stored locally, accessible only to authorized users. Off-site collaborators or sister facilities can securely (HTTPS security protocol) access the database via the internet.



Database and Client may reside on single computer
 Geneticist Assistant can be accessed by any computer having client within institution network
 Off-site collaborators or sister facilities can securely (HTTPS security protocol) access the database via the internet.



Quality control requirements are easily set in the Quality Control tab, the software will then monitor the sequence performance to the base level, indicating regions of non-performance.





User Management, Audit Trail, Access Control

Geneticist Assistant NGS Interpretative database employs a customizable password system (such as an 8 character alpha-numeric password) to protect data integrity. Database records all log-in and log-off and all user-activity by user, which can be recalled by administrative personnel. Access to various information contained in the database can be granted or limited by individuals, and groups. Geneticist Assistant NGS Interpretative Workbench records and tracks all changes and comments for future recall.

Server-	https://localhost	
iemame:	Administrator	
assword:		

Geneticist Assistant Workbench employs a customizable password system (such as an 8 character alpha-numeric password) to gain access to the database.

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Access to various information within Geneticist Assistant can be granted by individual and groups.

Geneticist Assistant Workbench records and tracks all changes and comments made to the database by users for future recall.

ID	14	Coverage	344	Protein	NP 001035197.1	Times Observed Per Panel	11	Ref 1
Chromosome	34	Fathogenicity	Bengn	Coding Base	24%	Times Observed Per Panel Group	11	
Chromosome Position	75513683	Fathogenicity Status	315 (J	Codon Position	1 2	Samples Per Panel	14	
Chr: ChePos	14:75515883	Variant Frequency	1	AA Position	826	Samples Per Panel Group	13	
88	117091	Typesity	hana	HG/S Genomic	0.75353883T>C	Times Observed Per Patient Per Panel	0	
Ref	T	Read Balance	0	HGVS Coding	6.2406A3G	Samples Per Patient Per Panel	0	
Ref M	Ann	Gene	NOO .	HGVS Protein	p.Amil.36Ap	Patient Variant Frequency	NA.	
Alt	0	Gene Strand		Variant Comment		Trans	5	
ALAA	Asp	Exas Number	2	Times Observed Per Run	12	GHAF		
Type	NUMBER	Transcript	NH_001040108.1	Fand	DUMP	AR	¢.	
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Type	Value	All and the second seco	Dete 1	Corment				
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Cool Tools

Custom Report Builder

Geneticist Assistant's Report Designer allows users to create highly customizable report templates for the quick and easy creation of standardized reports for each sample/patient. Using the Report Designer users can select the content to be included in the report and define formatting for the report such as report headers, page headers, as well as the inclusion of a lab logo image.

Custom tables can be created to pull data such as variant information and patient details directly from the Geneticist Assistant database. Custom text fields can also be added to include descriptions such as methods, clinical information and/or a disclaimer. Any custom section can be added when creating a report template through the Report Designer. The report template can then be saved for later use in saving reports. Multiple report templates can also be created for different report types. Templates can also be saved for individual sections within a report to allow the quick implementation of the same content when creating a new report template.



Customer Web Portal

Geneticist Assistant offer access to a customer we for tracking and managing tests ordered from refer web portal is directly linked to the Geneticist Assis information regarding patients and sample submis Geneticist Assistant and included in reports.

The web portal provides:

- Customizable interface
- Production and recognition of sample barcodes
- Secure encryption of patient information
- Patients and sample submissions linked directly with Geneticist Assistant
- · Printing of packing slips for sample tracking

Patient Tracking

Patient information, including a patient ID, DOB, gender, relationships, and phenotype, can be imported to the Geneticist Assistant database. Each imported sample can then be assigned to a patient

Compare Samples



Farmily	Compari	son of 4	Samples:														
D.M	CO.	QW	C Cm	APR APR	E Car	0. AL	O Day	1D-	Ov I ChrPee	Ra	Gene	Ref AA	At A&	Type	Colverage	Pathogenicity	HGVS Coding
0.494	106	0.433	119	3.000	121	0.554	1.27	1640	1:114443899	817464525	APHEL	Gly	Gy	synonymous	121	Unassigned	NM_001253852.1:c.576C>T
0.327	93 .	0.448	51	1.000	56	0.984	64	1745	1:108307727	137528153	VAV3	Thr	Sec	missense	56	Unassigned	NM_006113.4:c.892A>T
0.409	22	0.315	10	1.000	2	1.000	11	3167	2.206911228	152200111	2N080D	Ala	Val	missense	7	Deservation	NM_017799.4:c.1070C>T
0.636	11	0.667	24	0.773	22	8.300	10	3306	2111304496	1571231856	RCPD6	Val	Val	synonymous	22	Renign	NM_001123363.3:c.1560G>A
0.471	121	0.500	15	1,000	.97	1.000	79	3382	2:210557380	100720652	MAP2	His	Hu	synonymous	97	Unessigned	NM_002374.3:c.486C>T
0.485	130	0.426	122	1.000	135	0.992	138	3390	2.210557542	11741007	MAP2	Thr.	Thr	synonymous	135	Unassigned	NM_002314.3:c.6485+A
6.455	101	0.469	113	6.987	76	0.959	74	3396	2-210558162	n741006	MAP2	Arg	Lys	missense	76	Likely Eenign	NM_002374.3:±12686>A
0.374	187	0.379	145	1.000	169	1.000	130	3400	2,210559960	1:2739672	MAP2	Val	Val	synonymous	169	Likely Benign	NM_002314.3xt.3066G>T
0.425	80	0.554	64	0.982	35	1.000	59	3428	2.211456637	11047883	CPSL	Thr	Ala	missense	55	Beruge	NM_001122633.2 c.1048A>G
0.436	78	0.597	52	0.982	.57	1.000	59	3432	2.211456639	m2229589	CPSI	Thr	Thr	synonymous	57	Unassigned	NM_001122633.2xc.1050C+T
0.413	134	0.500	182	0.961	257	1.000	126	3440	2.211481257	12287599	CP51	Gly	Gh	synonymous	157	Unassigned	NM_001122633.2+c.2697C>G
0.625	72	0.319	79	1.000	.85	1.000	82	3594	2139663616	1510497199	DAPL	Ate	Thr	missense	86	Likely Deleterious	NM_001017920.2+196G>A

eb portal that can be used
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ssions can be viewed in

- Patient Ordering Provide First Name Last Name Phone Email Street City State Zip External Id Mother Father Gender Sue Smith 1234567890 800426 800418 800402 Female sue@hospital.org 2 Main St Specimen City PA 2017-01-23 **Date Collected** Units Sample Type Blood Genetic Counselor Tests Requested First Name Last Name John Jones 3456789012 Phone Email Reason For Testing John@counselor.org Indication **Clinical Diagnosis** Referring Hospital Or Laboratory Organiz Oncology Lat Phone Street City 6789012345 5 Main St B Addition Paramet Althest, Sections dentifie (Scolumbic Scole Refiniences Antering Salar Starting at 12 12 Attiles therefore Terra

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Create a comparison of multiple samples to view differences in variant calls and/or variant frequencies. Output from different pipelines can be compared by importing VCF files from each pipeline and comparisons of **family members**, such as a **trio comparison**, can be created.

1	Autosomal Re	cessive			1		
	Compound heteropygous						
panel: (Build37_CCD5	Exons Her	eOverlaps		-		
Sangle Name	Patient External ID	Relationship	Phenotype	e Zygosty			
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Sanule Name Mutation_Report1_Filtered Mutation_Report1_Filtered	Patient External ID UDP3168 UDP3165	Relationship Father • Hother •	Phenotype Unaffected Unaffected	e Zygosty • Heterozygour • Heterozygour	E E		
Sample Name Mutation_Report1_Filtered Mutation_Report1_Filtered Mutation_Report1_Filtered	Patient External ID UDP3168 UDP3165 UDP2753	Relationship Father • Hother • Son •	Phenotype Unaffected Unaffected Affected	 Zygosiły Heterozygouł Heterozygouł Homozygouł 			

For family comparisons, specify the relationships and phenotypes for each patient to create a comparison based on a selected inheritance pattern.

Comparison results show variant coverage and allele frequency values for each patient to quickly identify differences and shared variants.

Custom Filtering Options

Variants lists, as well as any other data tables in Geneticist Assistant, can be filtered based on a combination of any data fields.



Drag and drop any data field to use for filtering. Multiple filters can be combined and the combined filter can be saved for later use.

Customizable Workflow Builder

Geneticist Assistant NGS Interpretative Workbench now includes a completely customizable workflow builder that enables you to model your physical NGS workflow. A workflow can then be designated for any cases entered in Geneticist Assistant.



Geneticist Assistant NGS Interpretative Workbench features a completely customizable workflow builder that enables users to model physical workflow in "silico".

Process Quality Control

Control Charting for real time and historic evaluation

Track run-to-run variability of control samples. Data is tracked for each individual target region. The data can be used to determine drift in the analytical quality both globally as well as for specific genes and target regions. In addition, the data can be used to easily determine changes between manufacturer reagent lots. The tabular format can easily be exported in csv format to create control charts and graphs.

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7 Contrain	298.0	362.0	254.0	275,657542857	42.5900090728	0.5-	8.5	43	45	6.0
*	289.0	316.7	299.0	250	43.7207044774	45.	0.5	43	85	4.0
	204.2	334.0	338.2	325.4	27.4365354438	1.5	33	45	45	-84
*	ins.	204.0	1915	1824	11.3630779073	43	- 03	43	65	80
	\$22.0	998.0	628.2	696.575	105458029644	0.5	1.0	1.0	1825	0.242961450138
1	344.0	523.0	487.9	430.2	72.420941403	0.5	10	45	07	8,254568(74208
,	315.0	494.0	362.0	3634	44.4675062038	4.5	- 8.8	83	03	80
3	104.5	4412	403.8	386.4	53.680891094	45	15	43	45	44
	1510	346.0	118.0	III1679	40.0300074689	8.5	18	875	\$75	623
11	011	6143	483	NALISIBBERG,	76.5LDelimonta	4.5	18	1.0	0.9000000000	0.29(347)(0)(0)
30	234.0	504.0	367.5	342.2	58-3071233952	0.5	3.0	1.0	0.85	6.229138784798
10	3463	586.0	456.2	462.4	71.3290642908	0.5	18	45	87	0.244948674278
11	204.2	587.2	422.8	432.63636068	72.5407368689	0.5	10	10	0.000000000000	E2N04THOR
30	256.3	407.0	30.5	129.5	13.1337963207	3.8	18	1.0	3.8	80
11	301.0	594.0	811.5	285.805000007	79.0675052621	18	10	1.0	1.0	50
12	747	MILL	978	18/5	42 201000 2011	18	10	18	16	88

Geneticist Assistant records variants in control samples allowing instant review and long term monitoring of process.

or energy frequency of 100-101-14	runs. Ann 🖸				
CONTRACTOR OF BRIDE	Detter	DOM: N	OLD DATE 211	COLL OWING 1	Orte
Notes of Samples hereight	report Schwarders	IN A RIGE COMPARING	According to Commission Pro-	ALMONG: COLUMNAL PRODUCTION	Contraction of the
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	100.0	114.80	1047	101.00	111206048
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H	200.0	14713	101.08	210.86	ALC SOCIES IN
18.	9812	DED .	20136	254.02	The Overstanding
18	2412	152.90	20000	DOM	200,00000000
18	3812	386.05	396-41	207.58	25,85066
DK.	386.2	17275	311.54	209.38	121.36383834
18	2002	190.02	194	MO.HE	210.3000/000
	346.0	196.02	434.56	176.9	3H-JATUTELI
4	3812	28.87	6151	128.5	128.0720204
8	MUD	2544	LIBAS	2644	277.36
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8	386.0	245.44	474.26	156.43	246.040540405
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0	- MED	246,05	407.05	102.77	127101
4	388.2	316.05	401.52	364.87	tif//hooiti
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a	and a second sec	1000	1011 (M	ALC: NO.	ADL/S
	2012		807.015	100.00	400-2179/5/25

Control Sample Coverage is automatically captured by Geneticist Assistant on each run providing real time review of process while developing a historical overview to highlight any changes in the process over time.

Positive Control Verification

Many users opt to incorporate a positive control, such as NIST Genome in a bottle, with each sequencing run. Geneticist Assistant captures the positive control data, permitting a quick review of the run's efficacy and captures time-based data so that negative trends can be quickly observed and remedied.

C:/Users/soft/Des	ctop/GA	/referenc	es/Huma	n 37/NISTInte	gratedCal	s_14datasets_	131103_
Chromosome	17	DPSum	494	PLILLWG	393,42,0	TrancheSSEmin2	0
Chromosome Position	63533789	HRun	2	PLIIPCRFree	1628,129,0	YesPLtot	10
ID		HapNoVar	0	PLIONEX	170,21,0	allalts	с
Ref	т	NoPLTot	0	PLPlatGen	6514,520,0	datasetcalls	11
Alt	С	PL454WG	369,39,0	PLXII	897,72,0	geno	3
Qual	15292	PLCG	671,78,0	PLminsum	1295	genoMapGood	10
Filter	PASS	PLHSWEX	67,6,0	PLminsumOverDP	2.62	platformbias	none
HGVS Genomic		PLHSWG	918,93,0	TrancheABQDmin2	0	platformnames	ill,454,ion,cg
HGVS Coding		PLILL250	650,60,0	TrancheAlignmin2	0	platforms	4
HGVS Protein		PLILLCLIA	3015,235,0	TrancheMapmin2	0	varType	SNP



Geneticist Assistant captures positive control data which is very useful in determining efficacy of sequencing run and for determining quality trending.

Import Existing Knowledge Base

For variants with previously determined pathogenicity, a VCF file can be imported to automatically update the pathogenicity for these variants in the Geneticist Assistant database.

Import Variants		10 - 23
Sample File(*.vcf):	1	
Reference:	Human 37	•
Panel:	DUMP	
Panel Group: Set Variant Pathogenicit	default y by	
Import from VCF's I	NFO fields	
Select manually		
Pathogenicity an	nd Pathogenicity Status t	
OK Car	zel	

Recommended Hardware Requirements

Server: 2 cores 4 GB RAM 100 GB hard drive space available (solid state drive recommended)

Client: 2 cores 8 GB RAM 250 GB hard drive 64bit Windows Vista, 7, 8, Server 2003 through Server 2012 R2

Flag Artifacts

Geneticist Assistant NGS Interpretative Workbench allows users to flag variants that have been identified as artifacts and indicate the type of artifact, for example due to errors caused by chemistry or alignment. The variant can then automatically be flagged as an artifact when found in subsequent samples and can be easily filtered.

ID I	Or DePort	80	Fattopericity -	Participancie Status	HOISI	Colling	YESPUMP	Gew III	Tor	Coverage	Artifict Type
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3	3.17056045	014070007	Unsalgeet		M married	Abre				229	None
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8	3.5786755	6201006	Unassigned		N DE DOOR	1			1	40	Note
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11	14/25815826	14220023	Erroge		N					426	None
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п.,	14/75314489	0.2025000	Unusigned 1		Newson		Contraction of the local data	1996	100	472	Neva
18	\$2,757M72.	11342512	1000		NAL 000346.3 x 223	0.6	NP.000537.3:p.Pm72Atg	1951 - 19	1000	503	None

For more information or to arrange a free webinar or trial of Geneticist Assistant NGS Interpretative Workbench please visit www.softgenetics.com or email: info@softgenetics.com

Automatic BED file builder

Geneticist Assistant includes the BED File Builder Tool which can be used to create custom BED files for any panel. Simply enter the name of each gene to be included, or load a text file with multiple genes, choose the desired transcript, indicate the type of regions to be included and optionally choose to include a set number of bases at either end of each region.



Complete Analysis Pipeline

In conjunction with NextGENe® software

Geneticist Assistant can be used in conjunction with NextGENe's AutoRun Tool to provide a seamless pipeline for analysis, review and database submission. NextGENe can be configured to automatically access and begin processing data from the sequencing platform, and to then export results to the Geneticist Assistant database. Geneticist Assistant can also be configured to automatically import data from other analysis packages through a simple script.





64bit Linux (Ubuntu 12.04 or higher is recommended) or Windows Vista, 7, 8, 10 or Server 2003 through Server 2012 R2



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For Clinical Research