

Making NGS Data Analysis Clinically Practical: Repeatable and Time-Effective Workflows

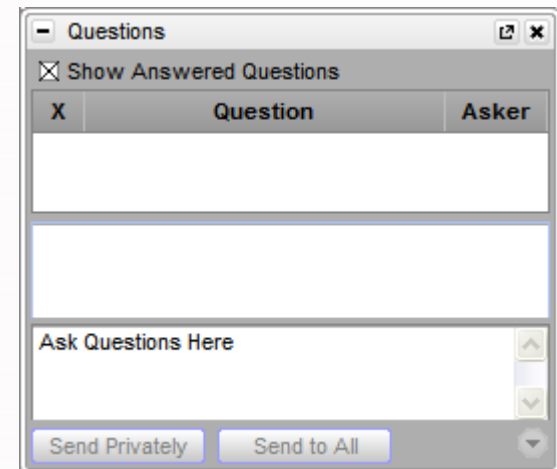


Autumn Laughbaum, Biostatistician
with introduction by Dr. Andreas Scherer,
President & CEO



Questions during the presentation

Use the Questions pane in your GoToWebinar window





Researcher



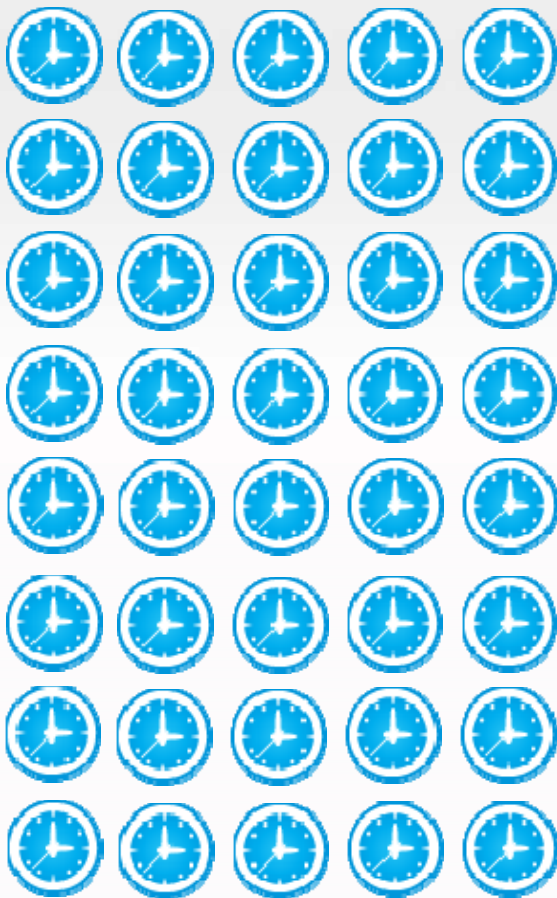
Clinician



Hands-on Time Savings



2 weeks



1 trio using Excel

2 hours



100 trios
using SVS

2 minutes



Unlimited trios
using automated
workflow



1 Status Quo

- Moving from Excel > SVS > Automated Workflows
- General sequencing workflow

2 Example Workflow: Ogden Syndrome

3 Example Workflow: Trio Analysis

4 Discussion

Analyzing Data from a secondary analysis pipeline: Using Excel



example_annotation_report.xlsx - Excel

Autumn Laughbaum

	K	L	M	N	O	P	Q	R	S	T	U	V	W	X	Y	Z	AA	AB
1	NA128911_GQ_91_DP	NA128911_GQ_91_DP	NA128911_GQ_91_DP	NA128911_GQ_91_DP	NA128911_GQ_91_DP	NA128911_GQ_91_DP	NA128911_GQ_91_DP	Classification	Gene(s)	Transcripts(s)	Coding Classification	HGVS Coding	HGVS Protein 1	Name	Function	1kG_Allel	1kG_All Indiv Freq	1kG_EU
2	33,23	71	54	r_r	9,2	21	12	Coding	SAMD11	NM_152486	Nonsyn SNV	c.2029T>C	p.Ser677Pro					
3	0,37	99	37	A_A	0,65	99	65	Coding	NOC2L	NM_015658	Synonymous	c.1843C>T	p.=	rs2272757	coding-synon	A	0.469999999	0.629
4	69,0	99	69	A_r	47,48	99	95	Coding	NOC2L	NM_015658	Nonsyn SNV	c.1528A>C	p.Asn510His	rs72631890	missense	G/T	0	
5	0,50	99	50	A_A	0,75	99	75	Coding	NOC2L	NM_015658	Synonymous	c.1182T>C	p.=	rs3828047	unknown	G	0.930000007	0.949
6	0,56	99	56	A_A	0,70	99	70	Coding	NOC2L	NM_015658	Synonymous	c.918A>G	p.=	rs3748596	coding-synon,missense	C	0.930000007	0.949
7	0,49	99	49	A_A	0,54	99	54	Coding	NOC2L	NM_015658	Nonsyn SNV	c.898A>G	p.Ile300Val	rs3748597	unknown	C	0.930000007	0.949
8	0,112	99	112	A_A	0,13	36	13	Coding	KLHL17	NM_198317	Synonymous	c.609G>C	p.=	rs4970441	coding-synon	C	0.870000005	0.939
9	12,10	99	21	r_r	22,2	5	23	Coding	KLHL17	NM_198317	Nonsyn SNV	c.1157C>G	p.Ala386Gly	rs199823418	missense			
10	15,30	99	43	A_r	21,28	21	47	Coding	KLHL17	NM_198317	Nonsyn SNV	c.1918A>C	p.Thr640Pro	rs188543688	near-gene-5,missense			
11	6,2	15	8	A_r	10,6	99	16	Coding	PLEKHN1	NM_001160184	Nonsyn SNV	c.1121G>C	p.Arg374Pro	rs61732689	missense	C/G	0.01	
12	42,25	99	64	A_r	27,23	99	48	Coding	ISG15	NM_005101	Nonsyn SNV	c.248G>A	p.Ser83Asn	rs1921	missense	A/G	0.340000004	0.400
13	0,66	99	66	A_A	0,56	99	56	Coding	ISG15	NM_005101	Synonymous	c.294A>G	p.=	rs8997	coding-synon	G	0.819999993	0.939
14	10,13	99	22	A_A	0,16	39	16	Coding	AGRN	NM_198576	Synonymous	c.3066A>G	p.=	rs2465128	coding-synon	A/G	0.819999993	0.910
15	57,31	99	84	A_A	0,17	48	17	Coding	AGRN	NM_198576	Synonymous	c.3558T>C	p.=	rs10267	coding-synon	C/T	0.839999974	0.920
16	0,68	99	68	A_A	1,160	99	160	Coding	SDF4	NM_016176	Nonsyn SNV	c.570T>C	p.=	rs6603781	coding-synon	G	0.939999998	0.870
17	39,16	27	54	r_r	26,4	57	29	Coding	SCNN1D	NM_001130413	Stopgain	c.1719C>A	p.Tyr573X					
18	0,9	24	9	A_A	0,13	33	13	Coding	CPSF3L	NM_017871	Synonymous	c.1641A>G	p.=	rs12103	coding-synon,near-gene-3	C	0.389999986	0.810
19	0,105	99	103	A_A	0,179	99	179	Coding	CPSF3L	NM_017871	Synonymous	c.882C>T	p.=	rs12142199	coding-synon	A	0.379999995	0.800
20	1,158	99	158	A_A	0,86	99	86	Coding	CPSF3L	NM_017871	Synonymous	c.264G>C	p.=	rs10907179	coding-synon,intron	G	0.75	0.920
21	0,20	60	20	A_A	0,9	27	9	Coding	GLTPD1	NM_001029885	Synonymous	c.468C>T	p.=	rs307349	coding-synon	T	0.779999971	0.920
22	0,10	30	10	A_A	0,47	99	47	Coding	TAS1R3	NM_152228	Nonsyn SNV	c.2269T>C	p.Cys757Arg	rs307377	unknown	C	0.959999979	0.970
23	7,7	99	14	?_?				Coding	MXRA8	NM_032348	Synonymous	c.735G>C	p.=	rs75904949	coding-synon			
24	6,2	40	8	A_r	3,4	22	7	Coding	CCNL2	NM_001039577	Nonsyn SNV	c.278G>C	p.Arg93Pro	rs200316100	near-gene-5,missense			
25	22,43	99	62	A_r	53,43	99	94	Coding	ATAD3C	NM_001039211	Nonsyn SNV	c.733C>G	p.Arg245Gly					
26	87,0	99	87	A_r	33,38	99	68	Coding	ATAD3B	NM_031921	Nonsyn SNV	c.1253G>A	p.Arg418Gln	rs79849353	missense	A/G	0.0005	
27	68,50	99	113	r_r	58,31	8	85	Coding	ATAD3B	NM_031921	Nonsyn SNV	c.1258C>G	p.Arg420Gly					
28	0,10	24	10	?_?				Coding	MIB2	NM_001170686	Nonsyn SNV	c.214T>C	p.Phe72Leu	rs7418389	ncRNA,missense,untranslat	C	0.509999999	0.709
29	0,14	30	15	?_?				Coding	MIB2	NM_001170686	Nonsyn SNV	c.305T>C	p.Met102Thr	rs12755088	intron,ncRNA,missense	C	0.759999999	0.920
30	26,15	32	39	r_r	1,0	3	1	Coding	MIB2	NM_001170686	Nonsyn SNV	c.1709T>G	p.Val570Gly					
31	67,23	99	86	A_r	77,35	99	107	Coding	SLC35E2B	NM_001110781	Nonsyn SNV	c.934G>A	p.=	rs76114385	unknown	C/T	0.550000012	0.620
32	48,26	99	71	r_r	68,55	99	121	Coding	SLC35E2B	NM_001110781	Synonymous	c.858T>C	p.=	rs74500811	unknown			

READY AVERAGE: 0.471863909 COUNT: 19629 SUM: 9261.744801 100%

Analyzing Data from a secondary analysis pipeline: Using SVS



The screenshot shows the SVS software interface. On the left is the 'Navigator Window Nodes' tree, which includes a folder for 'SVS_CEPHtrio' containing various tracks like 'Pedigree Data', 'Genotypes', 'dbNSFP v2.0 Matched Variants', and 'Variant Classification'. Overlaid on this is the 'Filter on Variant Frequency Catalog' dialog box. It shows the 'Selected Track' as 'NHLBI_ESP6500SI-V2_Exomes-Variant_Frequencies-2013_03_22_GHI_GRCh_37_Homo_sapiens.idf:1'. The 'Spreadsheet Action' is set to 'Annotate and Filter Variants'. Under 'Filter Criteria', the condition is 'European American MAF is >= 0.01'.

The 'Filter by NS Functional Predictions Track' dialog box is shown. It features the 'Autodetected NS Functional Predictions Track v2.0' and a 'Note' about non-synonymous missense coding variants. The 'Spreadsheet Action' is 'Annotate and Filter Variants', and 'Remove non-annotated variants' is checked. The 'Inactivate rows that pass' dropdown is set to 'All'. Under 'Filter Criteria', several checkboxes are visible, including 'SIFT predicted as... Damaging', 'and PolyPhen2 predicted as... Probably Damaging', 'and MutationTaster predicted as... Disease Causing Known', 'and MutationAssessor predicted as... Predicted Functional (High)', and 'and FATHMM predicted as... Damaging'.

The 'Variant Classification' dialog box is shown with the 'Basic' tab selected. It includes a 'Reference Allele Field' dropdown set to 'Reference', a 'Gene track' selection, and a 'Reference Sequence' selection. Under 'Region Definitions', 'Upstream distance (bp)' and 'Downstream distance (bp)' are both set to 1000. Under 'Output Reports', 'Variant Classification' and 'Coding Variant Classification' are checked. 'Amino Acid Notation' is set to '3 Letter'. At the bottom are 'OK', 'Cancel', and 'Help' buttons.

Analyzing Data from a secondary analysis pipeline: Automated Workflow



SVS Information

This tool will automate a workflow with the following steps:

- 1) Import VCF File(s) including
 - Genotypes
 - Read Depths
 - Genotype Qualities
- 2) Filter to Exon Regions using the following options:
 - *Gene Track=RefSeqGenes-UCSC_GRCh_37_Homo_sapiens.idf:1
 - *Expand regions by 10bp
- 3) Annotate and Filter based on additional VCF fields:
 - *Drop if Read Depth ≤ 10.0
 - *Drop if Genotype Quality ≤ 20.0
- 4) Annotate based on Genomic Super Dups, UCSC
- 5) Annotate and Filter based on dbNSFP_NS_Functional_Predictions-v2.0-2013-03-22-GHI_GRCh_37_Homo_sapiens.idf
- 6) Perform Variant Classification
 - *Activate only variants with the following classifications:
 - [u'Frameshift Sub', u'Stoploss', u'Splicing', u'Init Codon', u'Ins', u'Nonsyn SNV', u'Stopgain', u'Del', u'Frameshift Del', u'Frameshift Ins', u'Sub']
- 7) Filter to de Novo Variants [ENDPOINT 1]
- 8) Annotate and Filter based on NHLBI ESP6500SI-V2 Exomes - Variant Frequencies 2013_03, GHI
 - *Inactivate if European American MAF ≥ 0.01
- 9) Filter to Variants that fit Recessive Inheritance Pattern [ENDPOINT 2]
- 10) Filter to Variants that fit Specific Inheritance Pattern
 - *NA12878 in ['Alt_Ref']
- 11) Score Compound Heterozygous Variants [ENDPOINT 3]

Project Navigator Window

ID	(Linked To)
1	
2	
4	
114	
116	
128	
146	
148	

Node Change Log

Node Id: 181
Tue Sep 3 15:57:22 2013
SVS Win64 7.7.8 (2013-08-15), Autumn Laughbaum
Variant Classification Filter - NA12878 Candidates - S
Subset of Variant Classification Filter - NA12878 Can
----- End of Log Message -----
Node Id: 181
Tue Sep 3 15:57:22 2013
SVS Win64 7.7.8 (2013-08-15), Autumn Laughbaum
Changed node name to de Novo Candidates

Import Sorted VCF Files -- Select Files

Pedigree Spreadsheet:
Pedigree Data - Sheet 1 [Select Sheet]

Base Dataset Name: SVS_CEPHtrio

Input VCF files(s):
CEPHtrio_filteredByChildQC.vcf.gz [Add Files]

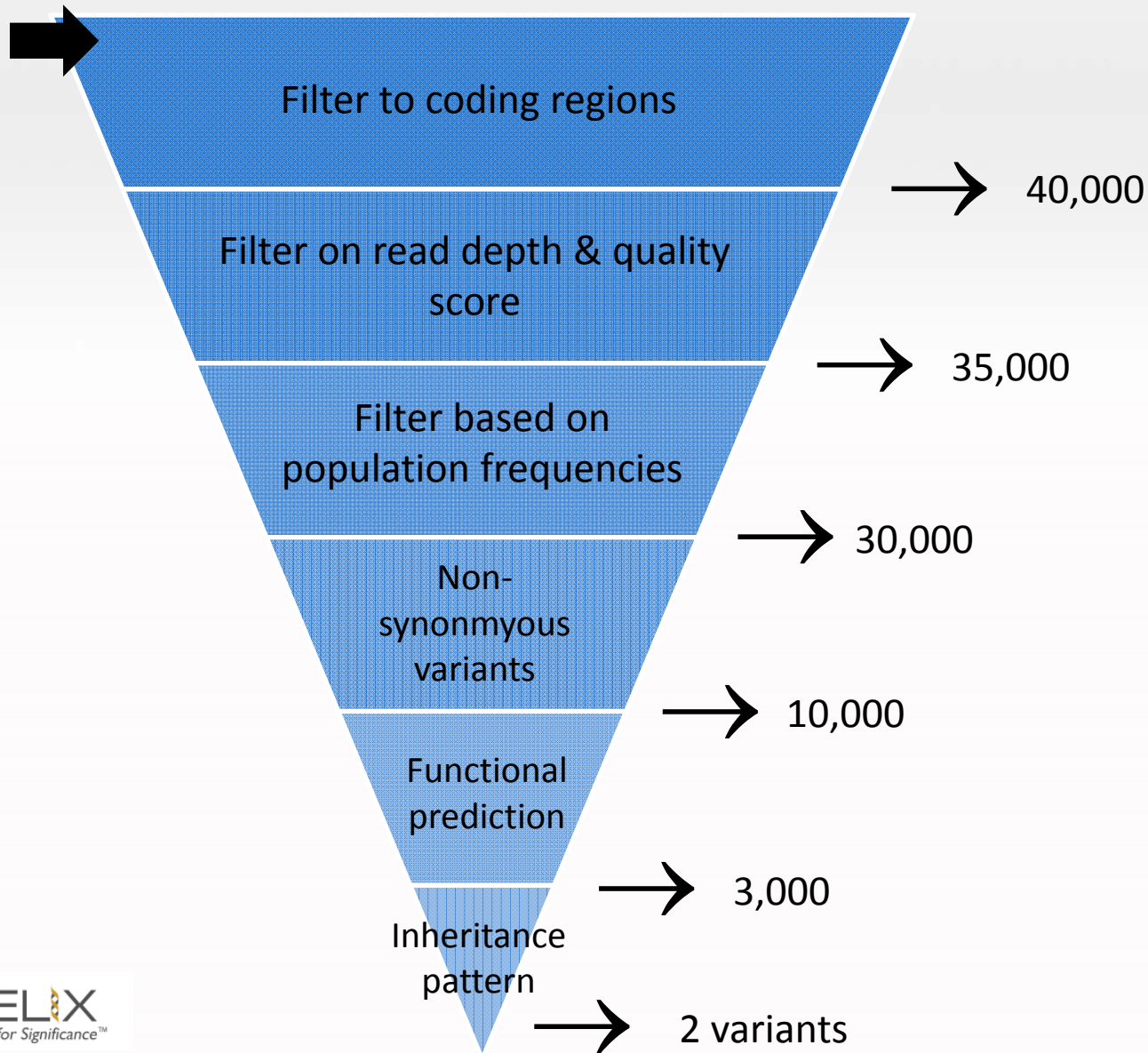
[Add Directory] [Remove]

[Start Workflow] [Cancel] [Help]

Analyzing Sequencing Data



Data from secondary analysis pipeline (VCF) – **2 million variants**

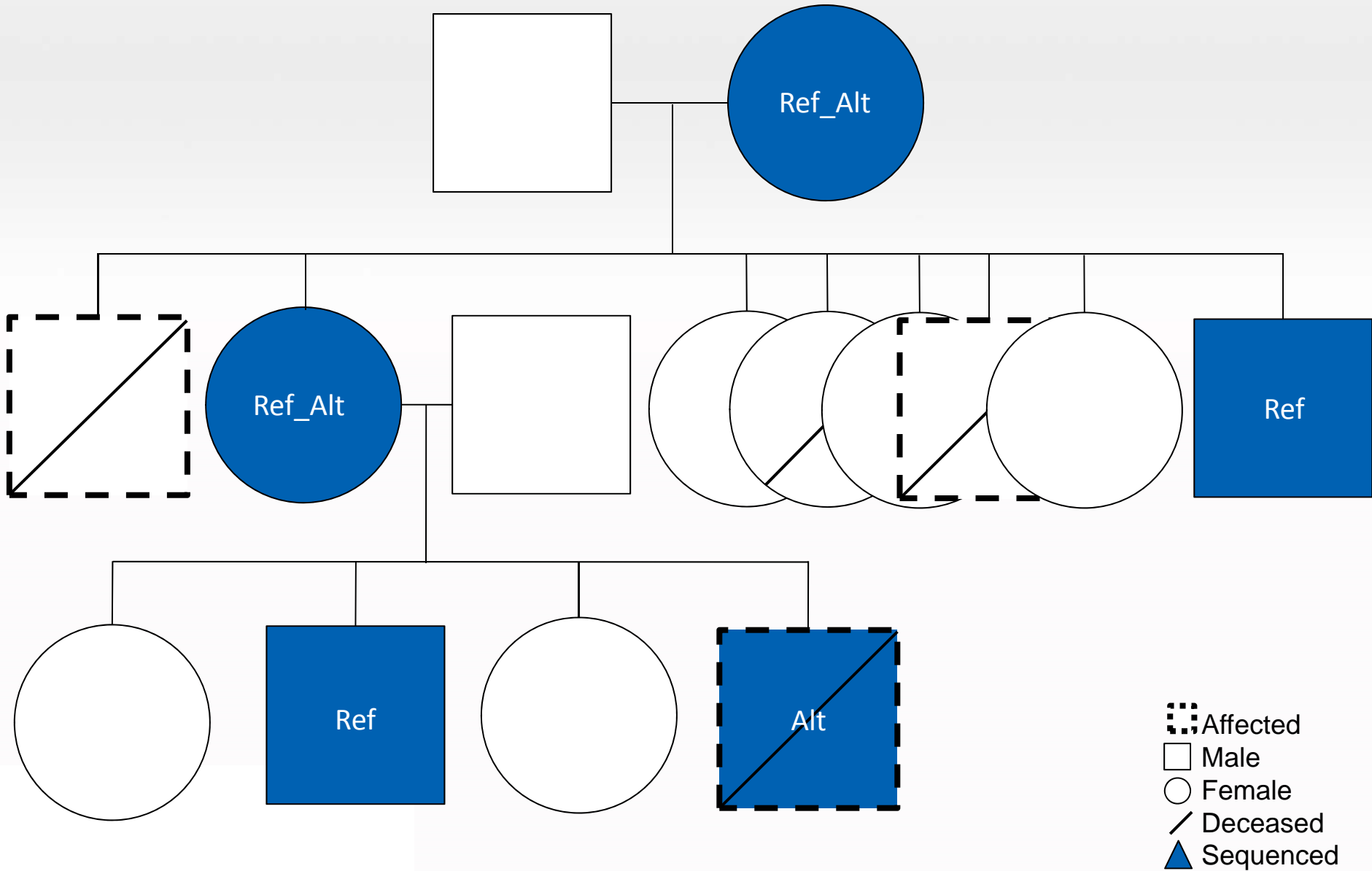




Workflow Example – Ogden Syndrome

WORKFLOW EXAMPLE – TRIO ANALYSIS

Pedigree

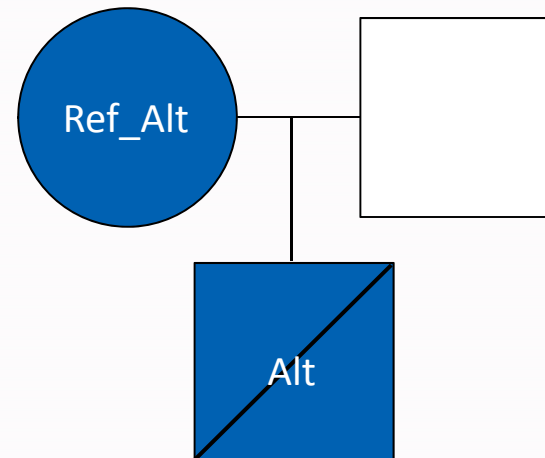
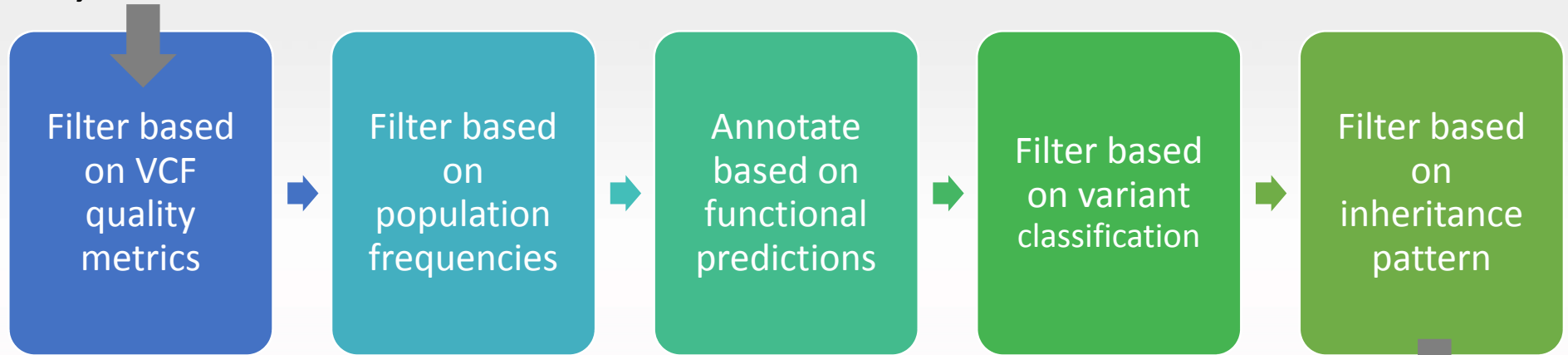




SVS Demo



5 Samples
107,000 variants



1 damaging variant

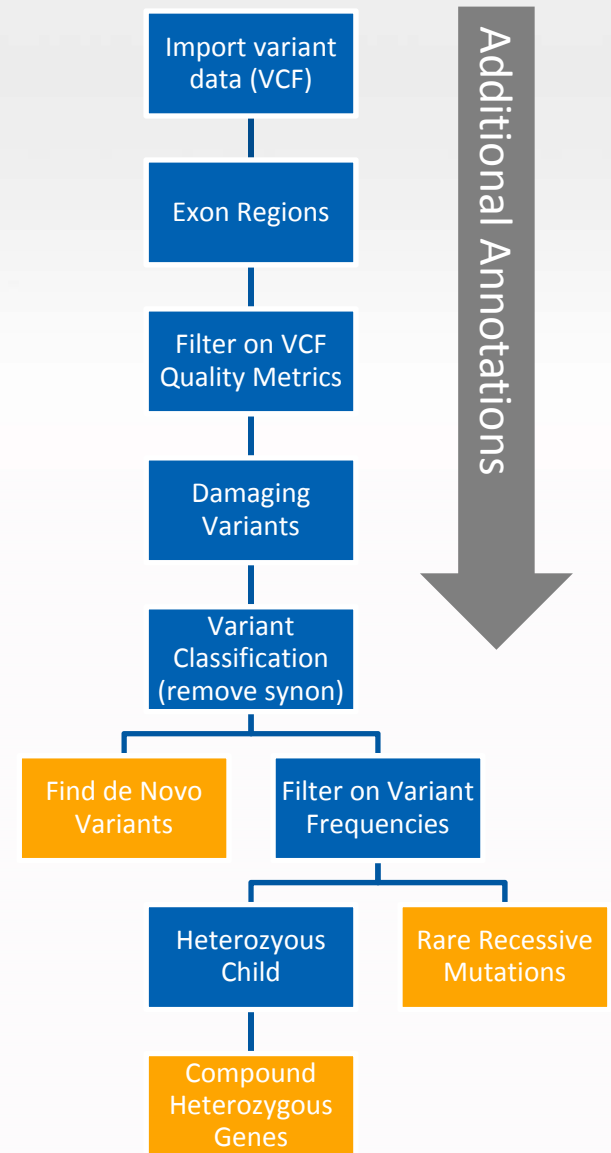


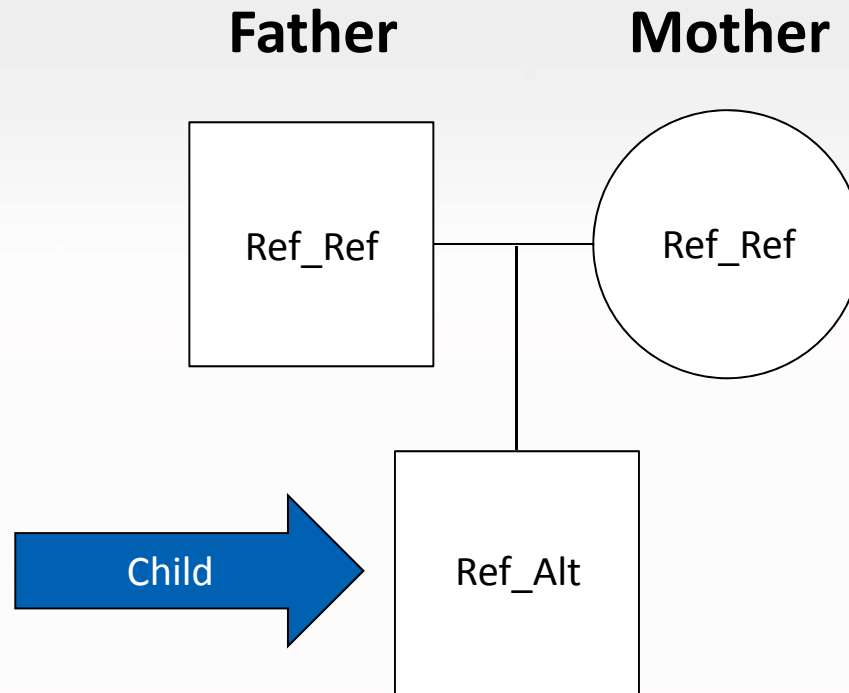
Workflow Example – Trio Analysis

WORKFLOW EXAMPLE – TRIO
ANALYSIS

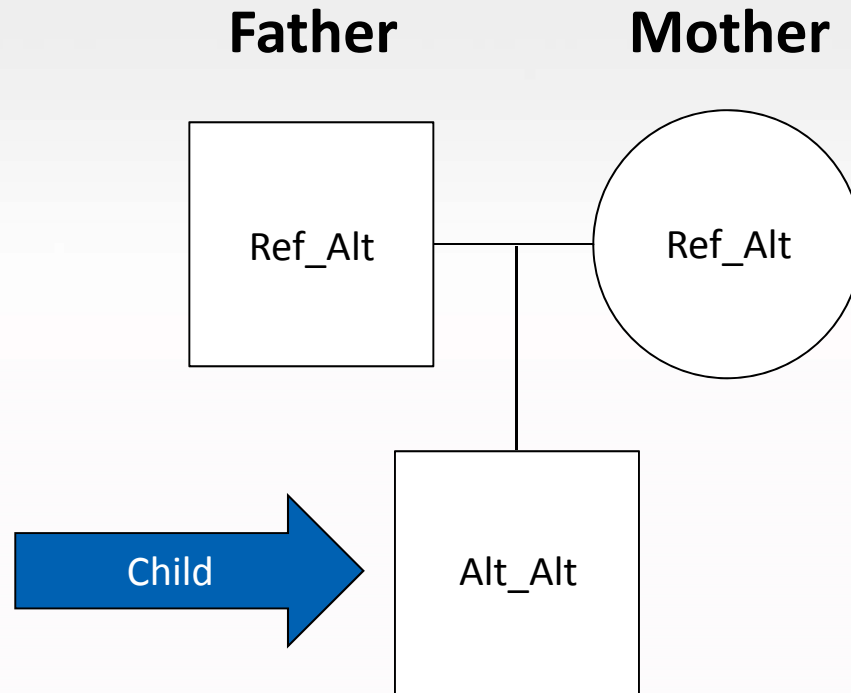


- de Novo Mutations
- Rare Recessive Mutations
- Compound Heterozygous Mutations

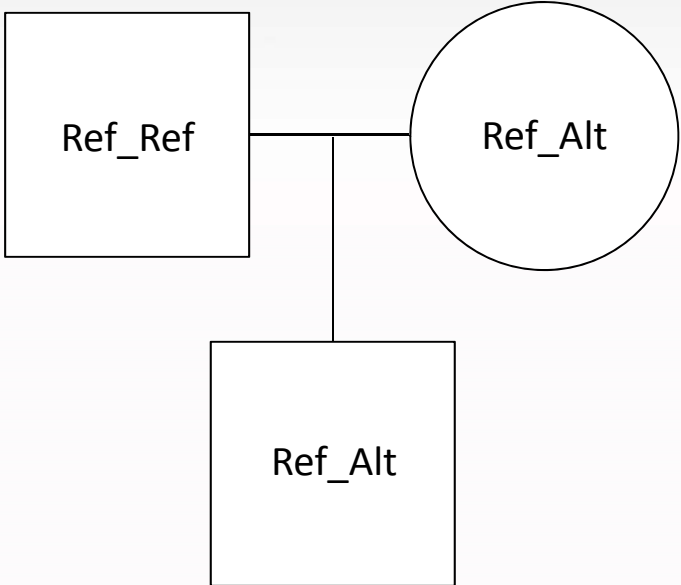
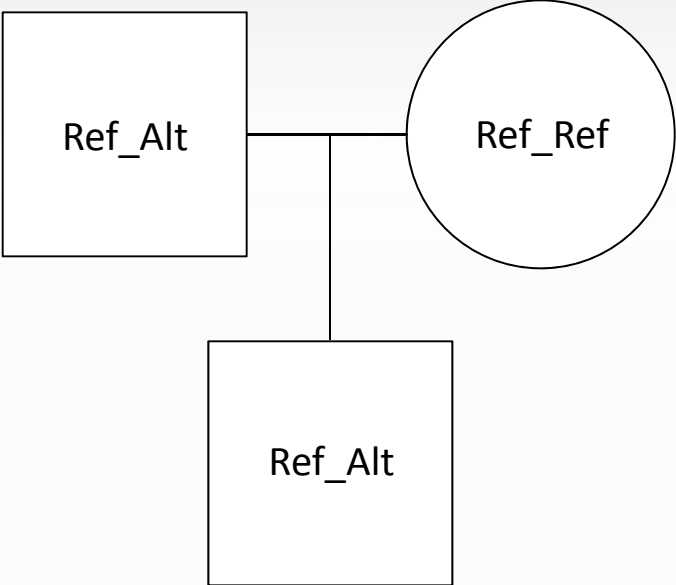




Rare Recessive Mutation



Compound Heterozygous Mutation




Re

Trio Analysis – Automated Workflow



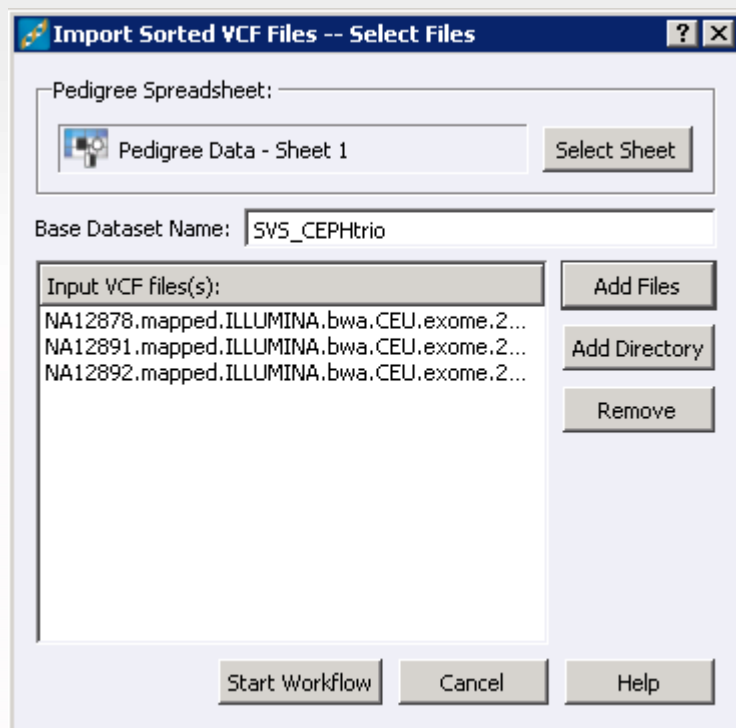
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 - *NA12878 in ['Alt_Ref']
- 11) Score Compound Heterozygous Variants [ENDPOINT 3]

OK

Trio Analysis – Automated Workflow





Project Navigator Window

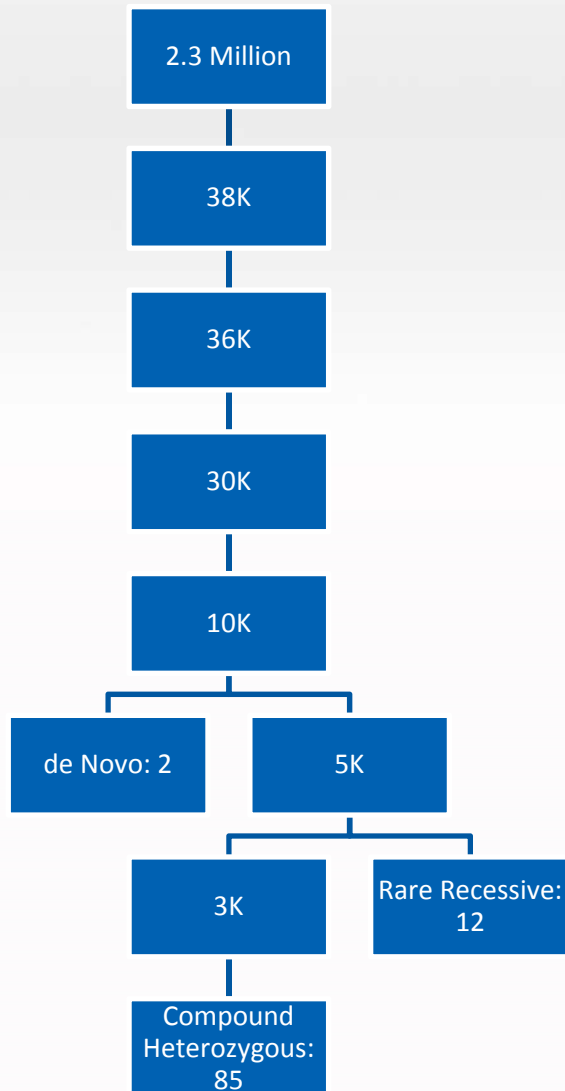
Navigator Window Nodes	ID	(Linked To)
▼ Pedigree Data	2	
▼ Pedigree Data - Sheet 1	4	
▼ SVS_CEPHtrio - Genotypes	5	
▼ SVS_CEPHtrio - Genotypes - Sheet 1	7	
▼ Exon Region Filter	19	
▼ Exon Region Filter - Genotypes filtered to No-call	37	
▼ All missing columns removed	39	
▼ Genomic Super Dups, UCSC Annotations	42	
▼ All missing columns removed - Sheet 2	43	
▼ dbNSFP v2.0 Matched Variants	46	
▼ dbNSFP Filter Results	48	
▼ Variant Classification	51	
▼ Coding Variant Classification	55	
▼ Invalid Transcripts	59	
▼ dbNSFP Filter Results - Sheet 2	60	
▼ Variant Classification Filter	62	
▼ Variant Classification Filter - NA12878 Candidates	64	
▼ de Novo Candidates	69	
▼ De Novo Candidate Variants	67	
▼ Variant Classification Filter - Sheet 2	97	
▼ NHLBI ESP650051-V2 Exomes - Variant Frequencies 2013_03, GHI Variant Matches and Filters	100	
▼ Variant Frequency Filter - NHLBI	102	
▼ Recessive Inheritance Filter	104	
▼ Variant Frequency Filter - NHLBI - Sheet 2	132	
▼ Compound Het Inheritance Filter	134	
▼ Score Compound Heterozygous Genes	137	
▼ Score Compound Heterozygous Variants	141	
▼ Compound Het Inheritance Filter - Sheet 2	143	
▼ Compound Heterozygous Variants	145	
▼ SVS_CEPHtrio - Genotypes Recoded AA/Ar/r	22	
▼ SVS_CEPHtrio - Read Depths (DP)	9	
▼ SVS_CEPHtrio - Read Depths (DP) - Sheet 1	11	
▼ SVS_CEPHtrio - Genotype Qualities (GQ)	12	
▼ SVS_CEPHtrio - Genotype Qualities (GQ) - Sheet 1	14	
▼ SVS_CEPHtrio - 0/1 Genotypes (GT)	15	
▼ SVS_CEPHtrio - 0/1 Genotypes (GT) - Sheet 1	17	
▼ SVS_CEPHtrio - Sample Collated Spreadsheet	32	
▼ SVS_CEPHtrio - Sample Collated Spreadsheet - Sheet 1	34	
▼ Master Annotation Report - de Novo Variants - Mapped Sheet 1	93	
▼ Master Annotation Report - de Novo Variants	95	
▼ Master Annotation Report - Mapped Sheet 1	128	
▼ Master Annotation Report - Rare Recessive Variants	130	
▼ Master Annotation Report - Mapped Sheet 1	173	
▼ Master Annotation Report - Compound Het Variants	175	
▼ Detailed Report	177	

Node Change Log

Node Id: 145
 Wed Sep 4 16:51:13 2013
 SVS Win64 7.7.8 (2013-08-15), Christophe Lambert
 Compound Het Inheritance Filter - Subset created fr
 Subset of Compound Het Inheritance Filter - Sheet 2
 ----- End of Log Message -----
 Node Id: 145
 Wed Sep 4 16:51:13 2013
 SVS Win64 7.7.8 (2013-08-15), Christophe Lambert
 Changed node name to Compound Heterozygous Va

User Notes

Trio Analysis – Automated Workflow



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 - *NA12878 in ['Alt_Ref']
- 11) Score Compound Heterozygous Variants [ENDPOINT 3]

OK

Golden Helix SVS Workflow Automation

Results from automated workflow "NGS Trio Complete Workflow" in project SVS_CEPHtrio at 2013-09-04 15:51:57

Final deNovo Variant Count: 2

- *4:144801662-SNV at 4:144801662, Ref/Alt=C/G,T,T, gene=GYPE, transcript=NM_002102, NM_198682, p.Gly13Ala
- *17:18647625-SNV at 17:18647625, Ref/Alt=T/A, gene=FBXW10, transcript=NM_031456, p.Ile23Asn

Final Rare Recessive Variant Count: 12

- *1:52306064-Del at 1:52306064, Ref/Alt=TCT/-, gene=NRD1, transcript=NM_001101662, NM_001242361, NM_002525, p.Glu22del
- *1:145302704-SNV at 1:145302704, Ref/Alt=A/G, gene=NBPF10, transcript=NM_001039703, p.Lys381Arg
- *3:190106072-Del at 3:190106072, Ref/Alt=G/-, gene=CLDN16, transcript=NM_006580, p.Ala56fs
- *3:190106074-SNV at 3:190106074, Ref/Alt=G/C, gene=CLDN16, transcript=NM_006580, p.Ala56Pro
- *5:139931630-Ins at 5:139931630, Ref/Alt=-/G, gene=SRA1, transcript=NM_001035235, p.Val110fs
- *5:140531592-SNV at 5:140531592, Ref/Alt=C/T, gene=PCDHB6, transcript=NM_018939, p.Thr585Ile
- *5:140563579-SNV at 5:140563579, Ref/Alt=C/T, gene=PCDHB16, transcript=NM_020957, p.Thr482Ile
- *5:140564088-SNV at 5:140564088, Ref/Alt=C/T, gene=PCDHB16, transcript=NM_020957, p.Arg652Cys
- *5:140581220-SNV at 5:140581220, Ref/Alt=C/A, gene=PCDHB11, transcript=NM_018931, p.Arg625Ser
- *12:10573094-SNV at 12:10573094, Ref/Alt=C/G, gene=KLRC3, transcript=NM_002261, NM_007333, p.Trp19Ser
- *15:43925134-SNV at 15:43925134, Ref/Alt=T/A, gene=CATSPER2, transcript=NM_172095, ?
- *17:18565350-SNV at 17:18565350, Ref/Alt=G/C, gene=ZNF286B, transcript=NM_001145045, p.Thr490Ser

Final Compound Het Variant Count: 85

- *1:1423281-SNV at 1:1423281, Ref/Alt=G/A, gene=ATAD3B, transcript=NM_031921, p.Arg418Gln
- *1:1423286-SNV at 1:1423286, Ref/Alt=C/G, gene=ATAD3B, transcript=NM_031921, p.Arg420Gly
- *1:65146995-SNV at 1:65146995, Ref/Alt=A/C, gene=CACHD1, transcript=NM_020925, p.Asp1103Ala
- *1:65157120-SNV at 1:65157120, Ref/Alt=C/G, gene=CACHD1, transcript=NM_020925, p.Pro1183Arg
- *1:70460304-SNV at 1:70460304, Ref/Alt=A/G, gene=LRRC7, transcript=NM_020794, p.Asn293Ser
- *1:70504932-SNV at 1:70504932, Ref/Alt=C/A, gene=LRRC7, transcript=NM_020794, p.Ala1104Glu

Automated Workflow Results



Master Annotation Report - de Novo Variants [95]

File Edit Select Quality Assurance Analysis Plot Scripts Help

All: 2 x 26
Active: 2 x 26

Unsort		G 1	I 2	R 3	G 4	I 5	R 6
Map	Variant	NA12878_GT	NA12878_DP	NA12878_GQ	NA12891_GT	NA12891_DP	NA12891_GQ
1	4:144801662-SNV	?_?	?	?	?_?	?	
2	17:18647625-SNV	A_r	244	40.25	A_A	247	68.5100021

Master Annotation

Master Annotation Report - Rare Recessive Variants [130]

File Edit Select Quality Assurance Analysis Plot Scripts Help

All: 12 x 28
Active: 12 x 28

Unsort		G 1	I 2	R 3	G 4	I 5	R 6
Map	Variant	NA12878_GT	NA12878_DP	NA12878_GQ	NA12891_GT	NA12891_DP	NA12891_GQ
1	1:52306064-Del	A_A	63	99	A_r	76	99
2	1:145302704-SNV	A_A	111	48.1300010681152	A_r	117	99
3	3:190106072-Del	A_A	95	99	A_r	105	99
4	3:190106074-SNV	A_A	95	99	A_r	103	99
5	5:139931630-Ins	A_A	38	99	A_r	50	99
6	5:140531592-SNV	A_A	81	36.0800018310547	A_r	102	99
7	5:140563579-SNV	A_A	120	99	A_r	125	99
8	5:140564088-SNV	A_A	29	42.1100006103516	A_r	46	99
9	5:140581220-SNV	A_A	75	42.0699996948242	A_r	67	64.8099975585938
10	12:10573094-SNV						
11	15:43925134-SNV						
12	17:18565350-SNV						

Master Annotation

Master Annotation Report - Compound Het Variants [175]

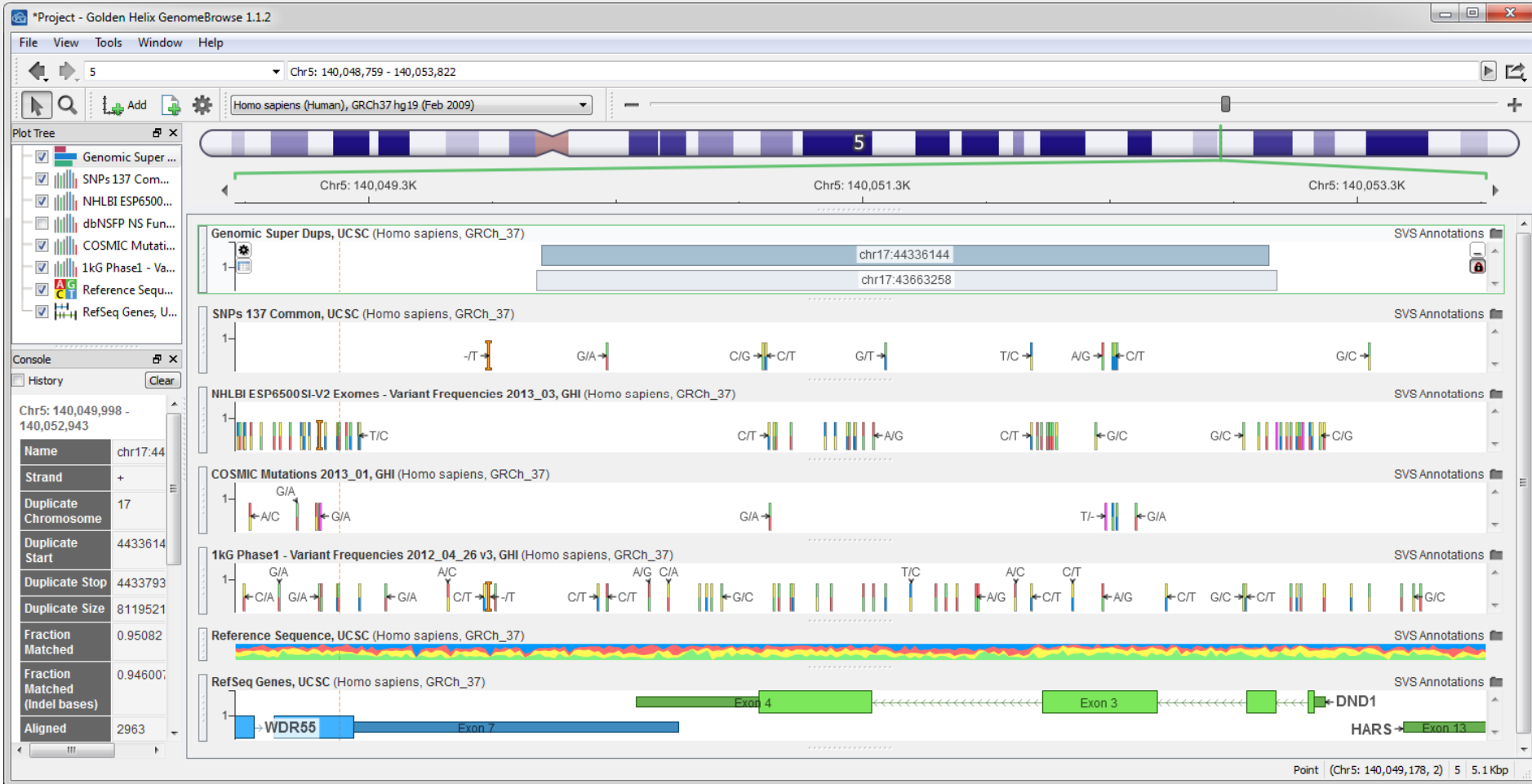
File Edit Select Quality Assurance Analysis Plot Scripts Help

All: 85 x 30
Active: 85 x 30

Unsort		G 1	I 2	R 3	G 4	I 5	R 6
Map	Variant	NA12878_GT	NA12878_DP	NA12878_GQ	NA12891_GT	NA12891_DP	NA12891_GQ
1	1:1423281-SNV	A_r	64	99	r_r	?	
2	1:1423286-SNV	A_r	84	99	A_r	118	
3	1:65146995-SNV	A_r	123	99	A_r	110	
4	1:65157120-SNV	A_r	84	99	r_r	?	
5	1:70460304-SNV	A_r	112	99	r_r	?	
6	1:70504932-SNV	A_r	87	99	A_r	100	89.62000274658
7	1:145293510-SNV	A_r	250	99	r_r	?	
8	1:145365316-SNV	A_r	191	99	A_r	213	
9	1:145365372-SNV	A_r	155	99	A_r	174	
10	1:208252715-SNV	A_r	18	99	A_r	17	
11	1:208272311-SNV	A_r	20	69.120002746582	r_r	?	
12	2:28824803-SNV	A_r	45	99	A_r	106	
13	2:28852004-SNV	A_r	38	70.870002746582	r_r	?	

Master Annotation Report - Compound Het Variants

Additional Annotations





Example in GenomeBrowse

WORKFLOW EXAMPLE – TRIO
ANALYSIS



Discussion

WORKFLOW EXAMPLE – TRIO
ANALYSIS



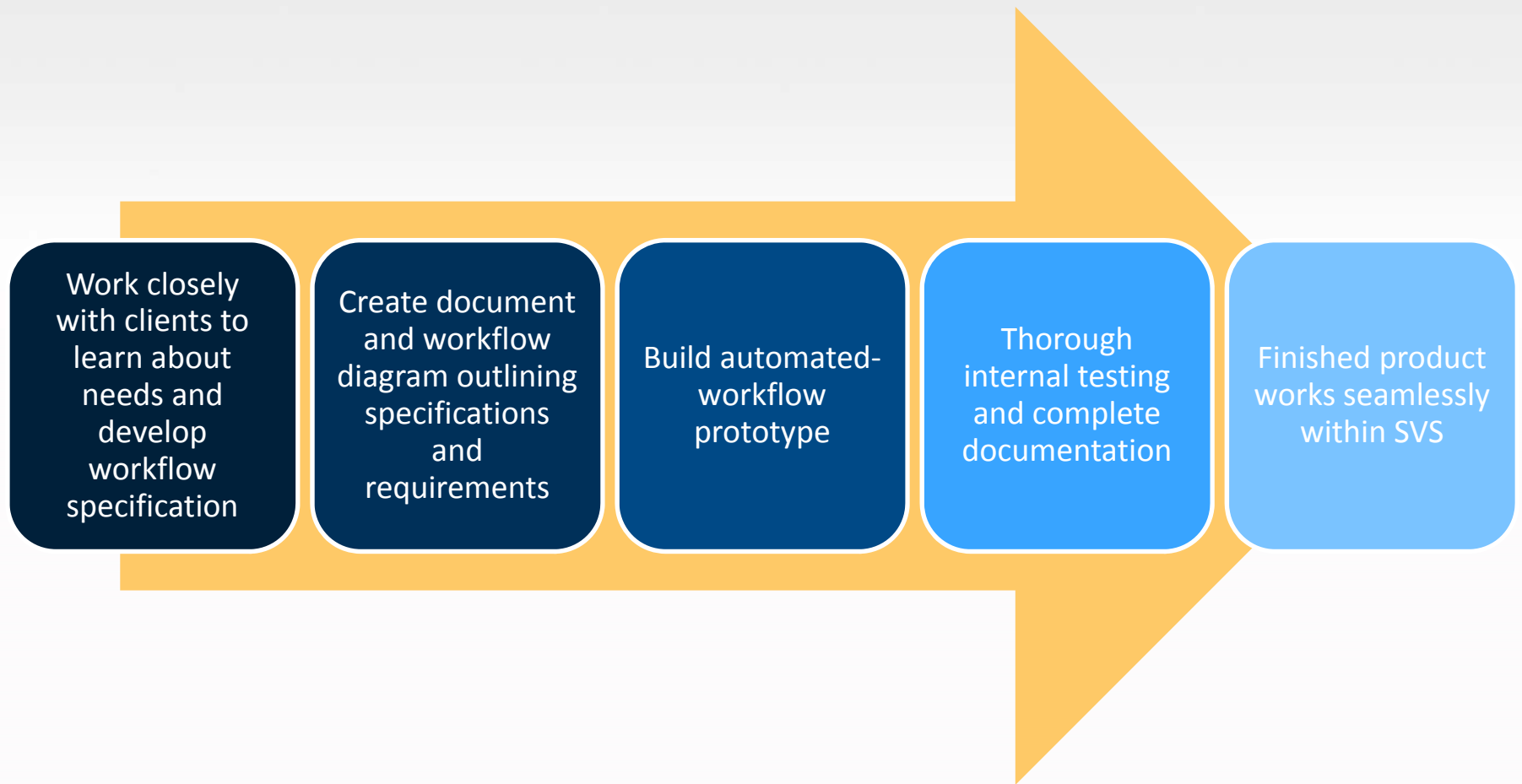
Clinicians

- Running well-defined workflow on additional samples
- Minimum user-interface knowledge
- Small learning curve
- Limited hands-on time

Researchers

- Building and testing workflows
- More complex but intuitive interface
- Larger learning curve
- Power to investigate and manipulate data

Conclusion – The GHI Approach





Questions during the presentation

Use the Questions pane in your GoToWebinar window

