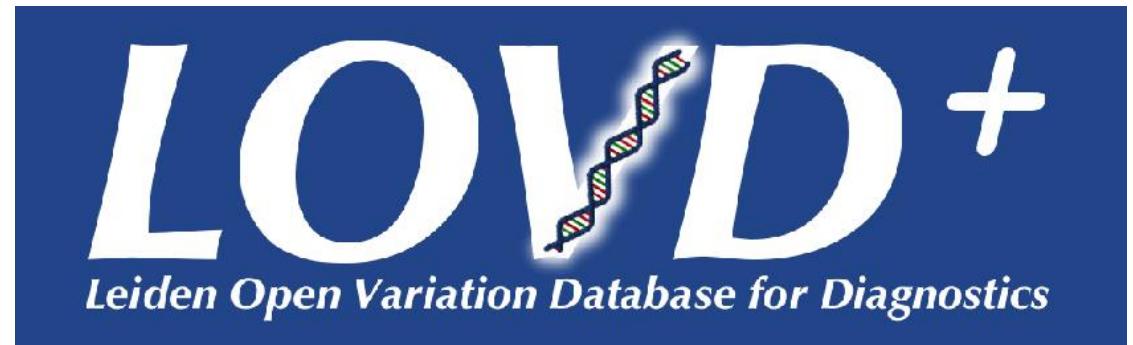


NGS analysis using LOVD⁺



Ivo Fokkema

Claudia Ruivenkamp

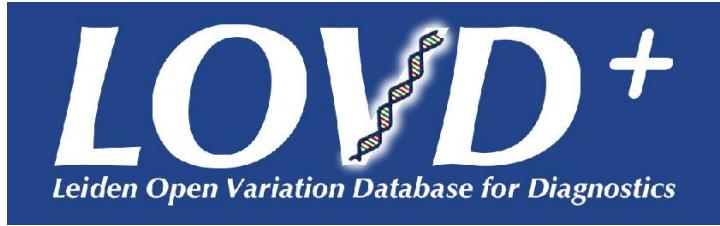
Johan den Dunnen





- Based on LOVD v3 platform
- Load VCF file (genomic positions) into annotation tool, like Ensembl VEP
 - *Maps to selected transcripts*
 - *Adds annotations*
- LOVD+ to filter and analyze results
 - *Used in accredited diagnostic labs*





- Supports trio-analysis
 - *Multi-sample VCF file or genotype of the parents in annotation*
 - *Complex family structures also possible, separate VCF files*
- Pre-set inheritance based filtering
 - *De novo, X-linked, recessive, ...*
- Analysis of whole exome or using gene panels
 - *Create gene panels with full history or define quick custom panels*
- Link to third party software
 - *e.g. Alamut, genome browsers, OMIM, LOVD search*

View samples

View individuals

5825 entries on 233 pages. Showing entries 2276 - 2300.

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Individual ID	ZIS ID	Lab-ID	Gender	Disease	Gene panels	Custom panel	Analysis status	Analysis by	Analysis started	Analysis approved by	Analysis approved
00002322	3487510	Child_12259656	F	ID	GROEI	-	In progress	Quint	2017-09-14	-	Claudia Ruivenkamp 2018-02-02
00002323	6517467	Child_12318677	M	ID	ID	-	Closed	Quint	2017-11-10	-	Mariëtte Hoffer 2017-11-01
00002324	9049502	Child_12218451	F	ID	ID	KMT2D, KDM6A, GLI3	Closed	Quint	2017-10-24	-	Claudia Ruivenkamp 2018-02-02
00002325	6572709	Child_12318280	M	ID	ID	-	Closed	Cindy Giroth	2017-11-07	-	Mariëtte Hoffer 2017-12-18
00002326	9571113	Child_12314511	M	ID	ID	-	Closed	Ivonne van Minderhout	2017-11-21	-	Mariëtte Hoffer 2017-12-13
00002327	2587994	Child_12639773	M	ID	ID	L1CAM	In progress	Ivonne van Minderhout	2017-08-14	-	Claudia Ruivenkamp 2018-02-02
00002328	571588	Child_12318286	F	ID	ID	ARID1B, ARID1A, SMARCE1, SMARCA4, SMARCB1	Closed	Quint	2017-11-09	-	Mariëtte Hoffer 2017-12-18
00002329	3510675	Child_12405546	M	ID	ID	-	In progress	Quint	2017-08-15	-	-
00002330	-	Patient_12639571	-	ID	-	-	Ready for analysis	-	-	-	-
00002331	1506462	Child_12161775	M	ID	ID, OBESE	-	Closed	Quint	2017-11-02	-	Mariëtte Hoffer 2017-12-06
00002332	9552273	Child_12230639	M	HART	HART	-	In progress	Quint	2017-10-26	-	-
00002333	6561695	Child_12228235	F	ID	ID	-	Closed	Quint	2017-10-24	-	Mariëtte Hoffer 2017-11-23
00002334	561699	Child_12229782	M	ID	ID	-	In progress	Ivonne van Minderhout	2017-10-23	-	-
00002334	561699	Child_12229782	M	ID	ID	-	Closed	Ivonne van Minderhout	2017-11-09	-	Mariëtte Hoffer 2017-11-22
00002336	3520294	Patient_12138380	F	SPIER WES	SPIER WES	-	Closed	Quint	2017-09-11	-	Hermine van Duyvenvoorde 2017-10-03
00002337	463662	Child_12229418	F	ID	ID	TBX6, DLL3, MESP2, HES7, LFNNG, SLC35A3, WNT3A, T	Closed	Ivonne van Minderhout	2017-10-09	-	Mariëtte Hoffer 2017-11-13
00002338	5280224	Child_12327535	F	ID	ID	GNAS	Closed	Quint	2017-11-13	-	Claudia Ruivenkamp 2018-01-26
00002339	9587197	Patient_12230856	M	ID	GROEI	FGD1	Closed	Quint	2017-10-27	-	Hermine van Duyvenvoorde 2018-09-03
00002340	8561958	Patient_12318876	F	ID	GROEI, ID, Skeletafwijkingen	-	Closed	Quint	2017-11-13	-	Hermine van Duyvenvoorde 2018-04-10
00002341	7274117	Patient_12318530	M	ID	ID	-	Ready for analysis	-	-	-	-
00002342	6400341	Patient_12229513	F	HART	HART	ADAMTS19, FLNC, GLIS1, HEY2, KLK14, LMCOL1, LTPB2, LZTR1, MAP3K7, PDGFRB, TNS1	In progress	Quint	2017-10-27	-	-
00002343	5134564	Patient_12229515	F	ID	GROEI	-	Closed	Quint	2017-10-27	-	Hermine van Duyvenvoorde 2017-11-07
00002344	1008937	Patient_12317465	M	ID	ID	-	In progress	Quint	2017-11-06	-	-
00002345	2055998	Patient_12318811	F	ONCO XL	ONCO-EL02	TP53	Closed	Quint	2017-11-13	-	Carli Tops 2018-11-01
00002346	-	Patient_12510527	F	MODY	MODY	-	Closed	Maaike Verschuren	2017-08-24	-	Ivonne van Minderhout 2017-09-01

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samples loaded for analysis

View Individual

Miracle ID	100
ZIS ID	-
Lab-ID	Child_00000100
Gender	M
Remarks	-
Custom gene panel	ID
Disease	ID
Total variants imported	13892
Created by	LOVD
Date created	2017-10-06 11:23:47
Last edited by	N/A
Date last edited	N/A

[Edit gene panels](#)

[Options](#)

Log entries

No logs found where event is "AnalysisOpen" or "AnalysisClose"!

Template	DNA
Technique	SEQ-NG
Seq coverage (%)	0.00685041
Derived gender	M
Exome covered	0.998057
Exome covered (father)	0.999157
Exome covered (mother)	0.998131
Reads mapped	0.811067
Reads on target (father)	0.807709
Reads on target (mother)	0.80075
Analysis restricted	1
Trio check: De novo	1
Trio check: Mendelian	0.9704472
Panel coverage	0.965984
Panel coverage (father)	0.971283
Panel coverage (mother)	0.970241
Analysis status	In progress (Close)
Analysis ID	Demo_Account_100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

Screenings

Screening ID: 0000000100 | Panel coverage: 0.965984 | Panel coverage (father): 0.971283 | Panel coverage (mother): 0.970241 | Curation progress: 0 of 0 variants curated | Variants found: 13892 | Analysis status: In progress

[Run analyses](#) [Gene panel analyses](#) [All analyses](#)

De novo (v1)		X-linked recessive (v1)		Recessive (gene panel) (v1)	
Filter	Time Var left	Filter	Time Var left	Filter	Time Var left
apply_selected_gene_panels	0s 625	chromosome_X_ID	0s 594	apply_selected_gene_panels	0s 625
remove_by_quality_lt_10	0s 369	remove_by_quality_lt_100	0s 52	remove_by_quality_lt_100	0s 369
remove_by_indb_count_hc_gt_e_2	0s 263	remove_by_indb_count_hc_gt_e_2	0s 44	remove_by_indb_count_hc_gt_e_5	0s 310
remove_by_indb_count_hc_gt_e_2	0s 232	remove_by_indb_count_hc_gt_e_2	0s 39	remove_by_indb_count_hc_gt_e_2	0s 277
remove_with_any_frequency_gt_2	0s 201	remove_with_any_frequency_gt_2	0s 39	remove_by_indb_count_utr_gt_2	0s 310
remove_with_any_frequency_1000G	0s 186	remove_with_any_frequency_gt_3	0s 38	remove_by_indb_count_utr_gt_2	0s 232
remove_with_any_frequency_dbSNP	0s 87	is_present_father_lt_e_4	0s 13	remove_with_any_frequency_gt_3	0s 214
remove_with_any_frequency_gnGL	0s 83	remove_intronic_distance_gt_8	0s 13	remove_intronic_distance_gt_8	0s 214
remove_with_any_frequency_EVS	0s 80	remove_intronic_distance_gt_2	0s 13	remove_intronic_distance_gt_2	0s 214
remove_intronic_distance_gt_8	0s 10	remove_by_function_utr	0s 4	remove_by_function_utr	0s 122
remove_intronic_distance_gt_2	0s 1	remove_by_function_utr5	0s 3	remove_by_function_utr5	0s 77
is_present_father_lt_e_4	0s 1	remove_by_function_utr_or_intronic	0s 2	remove_by_function_utr_or_intronic	0s 66
is_present_mother_lt_e_1	0s 1	remove_by_function_coding_synonymous	0s 1	remove_by_function_coding_synonymous	0s 46
is_present_father_1	0s 1	remove_by_function_utr_or_intronic_or_synonymous	0s 1	remove_by_function_utr_or_intronic_or_synonymous	0s 42
remove_intronic_distance_gt_8	0s 1			select_homozygous_or_compound_heterozygous	0s 11
remove_by_function_utr3	0s 1				
remove_by_function_utr5	0s 1				
remove_by_function_utr_or_intronic	0s 1				
remove_by_function_coding_synonymous	0s 1				
remove_by_function_utr_or_intronic_or_synonymous	0s 1				

11 entries on 1 page. Showing entries 1 - 11.

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Curation status	Effect	Chr	Allele	DNA change (genomic)	Alamut	PhyloP	#Ind. w/ var.	Var. ind. ratio	#Ind. w/ var & dis.	Var. dis. ind. ratio	Associated diseases	DNA change (cDNA)
-	./.	2	Heterozygous - Maternal (inferred)	g.26486348G>C	Alamut	2.295	100	1.0000	100	1.0000	Metabolic	HADHB:c.209+1G>C
-	./.	2	Heterozygous - Paternal (inferred)	g.26505759T>C	Alamut	4.953	100	1.0000	100	1.0000	Metabolic	HADHB:c.980T>C
-	./.	5	Homozygous	g.60628541_60628546del	Alamut	-0.534	100	1.0000	100	1.0000	ID	ZSWIM6:c.442_447del
-	./.	11	Heterozygous	g.2909505T>G	Alamut	-1.911	100	1.0000	100	1.0000	Growth; Heart; ID; Imprint; Beckwith-Wiedemann syndrome; IMAGE syndrome	CDKN1C:c.2652A>C, CDKN1C:c.2786A>C, CDKN1C:c.2804A>C, CDKN1C:c.2843A>C
-	./.	11	Heterozygous	g.2909513C>G	Alamut	-2.599	100	1.0000	100	1.0000	Growth; Heart; ID; Imprint; Beckwith-Wiedemann syndrome; IMAGE syndrome	CDKN1C:c.2660G>C, CDKN1C:c.2794G>C, SLC22A1AS:c.659G>C
-	./.	12	Homozygous	g.7343109_7343153del	Alamut	-1.444	100	1.0000	100	1.0000	ID; Metabolic	PEX5:c.136_180del, PEX5:c.136_147+3del
-	./.	17	Heterozygous	g.78063998G>A	Alamut	-1.951	100	1.0000	100	1.0000	ID	CDCD40:c.2893G>A
-	./.	17	Heterozygous	g.78064145_78064146insCAC	Alamut	0	100	1.0000	100	1.0000	ID	CDCD40:c.3040_3041insC
-	./.	19	Heterozygous - Paternal (inferred)	g.11314765C>T	Alamut	3.907	100	1.0000	100	1.0000	Heart; ID; Adams-Oliver syndrome	DOCK6:c.2792G>A
-	./.	19	Heterozygous - Maternal (inferred)	g.11358788G>A	Alamut	0.996	100	1.0000	100	1.0000	Heart; ID; Adams-Oliver syndrome	DOCK6:c.760C>T
-	./.	X	Homozygous	g.50376331C>G	Alamut	-0.18	100	1.0000	100	1.0000	ID; Stocco dos Santos X-linked mental retardation syndrome	SHROOM4:c.2742G>C

25 per page [Legend](#)

Analysis individual with ID gene panel (741 genes)

Gene panel design

Create a new gene panel entry

To create a new gene panel entry, please fill out the form below.

General information

Name
Description
Type Gene Panel
Remarks (optional)
Are PMIDs mandatory?

This gene panel has been linked to these diseases Blacklist (Blacklist)
CHA (Cerebrale Hereditaire Angiopathieen)
CRC (inherited colorectal cancer)
FAMMM (Familial Atypical Multiple Mole Melanoma syndrome)
Growth (Groeistoornissen)
HBOC (inherited breast and ovary cancer)
HBP (Hemoglobopathien)
Heart (Congenital heart disease)
ID (Intellectual Disability)
Imprint (Imprinted Genes)
Metabolic (Metabole aandoeningen)
migraine (Migraine (FHM), Hemiplegie, Epilepsie)
MODY (Monogene diabetes)
Motoric (Bewegingsstoornissen)
Muscle (Spierdystrofie)

This gene panel has been linked to these analyses De novo (v1)
Imprinted genes (v1)
Recessive (gene panel) (v1)
X-linked recessive (v1)

*Simple forms for
gene panel design*

NOTE: whether variants are linked to
a gene is defined during
variant annotation.

VEP uses a gene +/- 5000 bp.



Gene panel design

Manage genes for gene panel: ID



The following genes are configured in this LOVD. Click on one to add it to this gene panel.

19858 entries on 1986 pages. Showing entries 1 - 10.

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Symbol	Gene	Chr	Band	Transcripts	Associated with diseases
A1BG	alpha-1-B glycoprotein	19	q13.43	1	-
A1CF	APOBEC1 complementation factor	10	q21.1	6	-
A2M	alpha-2-macroglobulin	12	p13.31	1	-
A2ML1	alpha-2-macroglobulin-like 1	12	p13	2	-
A3GALT2	alpha 1,3-galactosyltransferase 2	1	p35.1	1	-
A4GALT	alpha 1,4-galactosyltransferase	22	q13.2	6	-
A4GNT	alpha-1,4-N-acetylglucosaminyltransferase	3	p14.3	1	-
AAAS	achalasia, adrenocortical insufficiency, alacrimia	12	q13	2	ID, Achalasia-addisonianism-alacrimia syndrome
AACS	acetocetyl-CoA synthetase	12	q24.31	4	-
AADAC	arylacetamide deacetylase	3	q25.1	3	-

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All genes below have been selected for this gene panel.

To remove a gene from this list, click the red cross on the far right of the line.

Symbol	Transcript	Inheritance	PMID	Remarks	
A1BG	-- select --	-- select --			x
AAAS	-- select --	-- select --			x
AARS2	-- select --	-- select --			x
ABCD1	-- select --	-- select --			x
ABCD4	-- select --	-- select --			x
ABHD5	-- select --	-- select --			x
ACAD9	-- select --	-- select --			x
ACO2	-- select --	-- select --			x
ACOX1	-- select --	-- select --			x
ACSF3	-- select --	-- select --			x
ACSL4	-- select --	-- select --			x
ACTB	-- select --	-- select --			x
total

Enter your password for authorization

[Save gene panel](#) [Cancel](#)

LOVD+ provides full gene panel history

NOTE: whether variants are linked to a gene is defined during variant annotation.

VEP uses a gene +/- 5000 bp.



Details NGS analysis

View individual #00000100

Miracle ID	100
ZIS ID	-
Lab-ID	Child_00000100
Gender	M
Remarks	-
Custom gene panel	
Gene panels	ID Edit gene panels
Diseases	ID
Total variants imported	13892
Created by	LOVD
Date created	2017-10-06 11:23:47
Last edited by	N/A
Date last edited	N/A

[Options](#) ▾

details Individual

Template	DNA
Technique	SEQ-NG
SNP mismatch (%)	0.00685041
Derived gender	M
Exome covered	0.998057
Exome covered (father)	0.999157
Exome covered (mother)	0.998131
Reads on target	0.811067
Reads on target (father)	0.807709
Reads on target (mother)	0.80075
Analysis restricted	1
Trio check: De novo	1
Trio check: Mendelian	0.0204472
Panel coverage	0.965984
Panel coverage (father)	0.971283
Panel coverage (mother)	0.970241
Analysis status	In progress (Close)
Analysis by	Demo Account 100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

details NGS

Details NGS

Template	DNA
Technique	SEQ-NG
SNP mismatch (%)	0.00685041
Derived gender	M
Exome covered	0.998057
Exome covered (father)	0.999157
Exome covered (mother)	0.998131
Reads on target	0.811067
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Analysis by	Demo Account 100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

QC
check

Details WES data

Details NGS

Template	DNA
Technique	SEQ-NG
SNP mismatch (%)	0.00685041
Derived gender	M
Exome covered	0.998057
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Analysis by	Demo Account 100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

coverage

Details WES data

Details NGS

Template	DNA
Technique	SEQ-NG
SNP mismatch (%)	0.00685041
Derived gender	M
Exome covered	0.998057
Exome covered (father)	0.999157
Exome covered (mother)	0.998131
Reads on target	0.811067
Reads on target (father)	0.807709
Reads on target (mother)	0.80075
Analysis restricted	1
Trio check: De novo	1
Trio check: Mendelian	0.0204472
Panel coverage	0.965984
Panel coverage (father)	0.971283
Panel coverage (mother)	0.970241
Analysis status	In progress (Close)
Analysis by	Demo Account 100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

Information
and controls

Details WES data

Gene panel analysis

Screenings

Screening ID	Panel coverage	Panel coverage (father)	Panel coverage (mother)	Curation progress	Variants found	Analysis status
0000000100	0.965984	0.971283	0.970241	0 of 0 variants curated	13892	In progress

Run analyses Gene panel analyses All analyses

tabular display of analyses

De novo (v1)		
Filter	Time	Var left
apply_selected_gene_panels	0s	625
ID		
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_2	0s	263
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_2	0s	209
remove_with_any_frequency_1000G	0s	186
remove_with_any_frequency_dbSNP	0s	87
remove_with_any_frequency_goNL	0s	83
remove_with_any_frequency_EVS	0s	80
is_present_father_lte_4	0s	10
is_present_mother_lte_4	0s	1
is_present_father_1	0s	1
is_present_father_1	0s	1
remove_intronic_distance_gt_8	0s	1
remove_intronic_distance_gt_2	0s	1
remove_by_function_utr3	0s	1
remove_by_function_utr5	0s	1
remove_by_function_utr_or_intronic	0s	1
remove_by_function_coding_synonymous	0s	1
remove_by_function_utr_or_intronic_or_synonymous	0s	1

X-linked recessive (v1)		
Filter	Time	Var left
chromosome_X	0s	354
apply_selected_gene_panels	0s	88
ID		
remove_by_quality_lte_100	0s	52
remove_by_indb_count_hc_gte_5	0s	310
remove_by_indb_count_ug_gte_5	0s	277
remove_by_indb_count_hc_gte_2	0s	250
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_3	0s	38
is_present_father_lte_4	0s	13
remove_intronic_distance_gt_8	0s	13
remove_intronic_distance_gt_2	0s	13
remove_by_function_utr3	0s	4
remove_by_function_utr5	0s	3
remove_by_function_utr_or_intronic	0s	2
remove_by_function_coding_synonymous	0s	1
remove_by_function_utr_or_intronic_or_synonymous	0s	1

Recessive (gene panel) (v1)		
Filter	Time	Var left
apply_selected_gene_panels	0s	625
ID		
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_5	0s	310
remove_by_indb_count_ug_gte_5	0s	277
remove_by_indb_count_hc_gte_2	0s	250
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_3	0s	38
remove_intronic_distance_gt_8	0s	214
remove_intronic_distance_gt_2	0s	214
remove_by_function_utr3	0s	122
remove_by_function_utr5	0s	77
remove_by_function_utr_or_intronic	0s	66
remove_by_function_coding_synonymous	0s	46
remove_by_function_utr_or_intronic_or_synonymous	0s	42
select_homozygous_or_compound_heterozygous	0s	11



pane selected

11 entries on 1 page. Showing entries 1 - 11.

25 per page	Legend	Curation status	Effect	Chr	Allele	DNA change (genomic)	Alamut	PhyloP	#Ind. w/ var.	Var. ind. ratio	#Ind. w/ var & dis.	Var. dis. ind. ratio	Associated diseases	DNA change (cDNA)
		-	./.	2	Heterozygous - Maternal (inferred)	g.26486348G>C	Alamut	2.295	100	1.0000	100	1.0000	Metabolic	HADHB:c.209+1G>C
		-	./.	2	Heterozygous - Paternal (inferred)	g.26505759T>C	Alamut	4.953	100	1.0000	100	1.0000	Metabolic	HADHB:c.980T>C
		-	./.	5	Homozygous	g.60628541_60628546del	Alamut	-0.534	100	1.0000	100	1.0000	ID	ZSWIM6:c.442_447del
		-	./.	11	Heterozygous	g.2909505T>G	Alamut	-1.911	100	1.0000	100	1.0000	Growth; Heart; ID; Imprint; Beckwith-Wiedemann syndrome; IMAGE syndrome	CDKN1C:c.-2652A>C, CDKN1C:c.2786A>C, SLC22A18AS:c.667A>C
		-	./.	11	Heterozygous	g.2909513C>G	Alamut	-2.599	100	1.0000	100	1.0000	Growth; Heart; ID; Imprint; Beckwith-Wiedemann syndrome; IMAGE syndrome	CDKN1C:c.2660G>C, CDKN1C:c.2794G>C, SLC22A18AS:c.659G>C
		-	./.	12	Homozygous	g.7343109_7343153del	Alamut	-1.444	100	1.0000	100	1.0000	ID; Metabolic	PEX5:c.136_180del, PEX5:c.136_147+33del

Gene panel analysis: 741 ID genes

- 3 standard inheritance patterns (de novo, X-linked, recessive)
- Stringent variant filtering

Imprinted genes

Imprinted genes		
Filter	Time	Var left
remove_not_in_gene_panel	2s	1508
remove_by_quality_lte_100	0s	550
remove_not_imprinted	0s	18
remove_by_indb_count_hc_gte_2	0s	14
remove_by_indb_count_ug_gte_2	0s	11
remove_with_any_frequency_gt_2	0s	11
remove_with_any_frequency_1000G	0s	11
remove_with_any_frequency_dbSNP	0s	3
remove_with_any_frequency_goNL	0s	3
remove_with_any_frequency_EVS	0s	3
remove_intronic_distance_gt_8	0s	2
remove_intronic_distance_gt_2	0s	2
remove_by_function_utr3	0s	2
remove_by_function_utr5	0s	1
remove_by_function_utr_or_intronic	0s	1
remove_by_function_coding_synonymous	0s	0
remove_by_function_utr_or_intronic_or_synonymous	0s	0

*separate analysis for variants in
imprinted genes*

Whole exome analysis

Run analyses Gene panel analyses All analyses

De novo (v1)		
Filter	Time	Var left
remove_not_in_gene_panel	2s	1372
Apply selected gene panels	-	-
Remove variants with sequencing quality <= 100	0s	541
Remove with inhouse DB count >= 2 (hc)	0s	456
Remove with inhouse DB count >= 2 (ug)	0s	392
Remove variants with frequency > 2 %	0s	365
Remove variants in 1000G	0s	347
Remove variants in dbSNP	0s	163
Remove variants in GoNL	0s	163
Remove variants in EVS	0s	158
Remove variants probably in mother	0s	32
Remove variants probably in father	0s	8
Remove variants found in mother	0s	0
Remove variants found in father	0s	0
Remove variants with distance > 8 bp	-	-
Remove intronic with distance > 2 bp	0s	1
Remove variants only in 3' UTR	0s	0
Remove variants only in 5' UTR	0s	0
Remove variants only in UTR or intron	0s	0
Remove synonymous variants	0s	0
Remove variants only UTR, intron or synonymous	-	-

X-linked recessive (v1)		
Filter	Time	Var left
Select X chromosome	2s	701
remove_not_in_gene_panel	0s	168
Apply selected gene panels	-	-
Remove variants with sequencing quality <= 100	0s	47
Remove with inhouse DB count >= 2 (hc)	0s	40
Remove with inhouse DB count >= 2 (ug)	0s	36
Remove variants with frequency > 3 %	0s	35
Remove variants probably in father	0s	15
Remove intronic with distance > 8 bp	-	-
Remove intronic with distance > 2 bp	0s	1
Remove variants only in 3' UTR	0s	0
Remove variants only in 5' UTR	0s	0
Remove variants only in UTR or intron	0s	0
Remove synonymous variants	0s	0
Remove variants only UTR, intron or synonymous	-	-

Recessive (gene panel) (v1)		
Filter	Time	Var left
remove_not_in_gene_panel	1s	1372
Apply selected gene panels	-	-
Remove variants with sequencing quality <= 100	0s	541
Remove with inhouse DB count >= 5 (hc)	0s	526
Remove with inhouse DB count >= 5 (ug)	0s	504
Remove with inhouse DB count >= 2 (hc)	0s	447
Remove with inhouse DB count >= 2 (ug)	0s	392
Remove variants with frequency > 3 %	0s	368
Remove variants with frequency > 3 %	0s	368
Remove intronic with distance > 8 bp	-	-
Remove intronic with distance > 2 bp	0s	42
Remove variants only in 3' UTR	0s	32
Remove variants only in 5' UTR	0s	25
Remove variants only in UTR or intron	0s	20
Remove synonymous variants	0s	13
Remove variants only UTR, intron or synonymous	-	-
Select homozygous or possibly compound heterozygous	0s	2

Recessive (whole exome) (v1)		
Filter	Time	Var left
Remove variants with sequencing quality <= 100	11s	12600
Select called by both UG and HC	2s	5426
Remove with inhouse DB count >= 5 (hc)	1s	5237
Remove with inhouse DB count >= 5 (ug)	1s	4958
Remove with inhouse DB count >= 2 (hc)	1s	4306
Remove with inhouse DB count >= 2 (ug)	0s	3684
Remove variants with frequency > 3 %	0s	3432
Remove intronic with distance > 8 bp	-	-
Remove intronic with distance > 2 bp	0s	740
Remove variants only in 3' UTR	0s	647
Remove variants only in 5' UTR	0s	586
Remove variants only in UTR or intron	0s	561
Remove synonymous variants	0s	437
Remove variants only UTR, intron or synonymous	-	-
Remove missense with low PhyloP	0s	284
Select homozygous or possibly compound heterozygous	0s	22
Apply gene blacklist	0s	19

Imprinted genes (v1)		
Filter	Time	
remove_not_in_gene_panel	2s	
Apply selected gene panels	-	
Remove variants with sequencing quality <= 100	0s	
Remove variants not in imprinted genes	0s	
Remove with inhouse DB count >= 2 (hc)	0s	
Remove with inhouse DB count >= 2 (ug)	0s	
Remove variants with frequency > 2 %	0s	
Remove variants in 1000G	0s	
Remove variants in dbSNP	0s	
Remove variants in GoNL	0s	
Remove variants in EVS	0s	
Remove variants only in 3' UTR	0s	
Remove intronic with distance > 2 bp	0s	
Remove variants only in 5' UTR	0s	
Remove synonymous variants	0s	
Remove variants only UTR, intron or synonymous	0s	

19 entries on 1 page. Showing entries 1 - 19.

25 per page ▾ Legend

Effect	Chr	Allele	DNA change (genomic)	Alamut	PhyloP	#Ind. w/ var.	Var. ind. ratio	#Ind. w/ var & dis.	Var. dis. ind. ratio	Associated diseases	DNA change (cDNA)	Protein	GVS function	OMIM
-?.	2	Homozygous	g.20867122..20867123insGGC...	Alamut	0.064	361	0.0625	311	0.1480	-	GDF7:c.123_124insGGC...	p.?	coding	GDF7
-?.	(-?) 2	Heterozygous	g.70524451C>T	Alamut	1.673	1099	0.1903	853	0.4060	-	FAM136A:c.387G>A	p.(Met129Ile)	missense	
-?.	(-?) 2	Heterozygous	g.70524477G>C	Alamut	3.466	717	0.1241	550	0.2618	-	FAM136A:c.361C>T	p.(Leu121Val)	missense	
-?.	3	Homozygous	g.42251580..42251581insGGA...	Alamut	-0	855	0.1480	635	0.3022	-	TRA1K:c.1892_1893insGGA..., TRA1K:c.1844_1845insGGA..., TRA1K:c.1963+103..1963+104insGGA..., TRA1K:c.2066_2067insGGA...	p.(?), p.?	intron, codingComplex	TRA1K
-?.	(-?) 3	Homozygous	g.133969437..133969438insG	Alamut	0.144	2114	0.3660	1476	0.7025	-	RYK:c.59_60insC	p.?	frameshift	RYK
-?.	(-?) 3	Homozygous	g.133969487..133969488insC	Alamut	-0.231	1055	0.1827	759	0.3613	-	RYK:c.9_10insG	p.?	frameshift	RYK
-?.	(-?) 5	Heterozygous	g.60628181..60628182insCCG	Alamut	1.218	291	0.0504	240	0.1142	ID	ZSWIM6:c.82_83insCGC	p.?	coding	
-?.	(-?) 5	Homozygous	g.60628575..60628589del	Alamut	0.196	287	0.0497	284	0.1352	ID	ZSWIM6:c.476_490del	p.?	coding	
??.	9	Homozygous	g.129854097..129854098del	Alamut	6.025	2	0.0003	2	0.0010	-	ANGPTL2:c.113..113del	p.?	frameshift	ANGPTL2
-?.	(-?) 12	Heterozygous	g.424969690T>C	Alamut	3.696	1814	0.3141	1477	0.7030	-	GXYLT1:c.701A>G, GXYLT1:c.794A>G	p.(Tyr234Cys), p.(Tyr265Cys)	missense	GXYLT1
-?.	(-?) 12	Heterozygous	g.424969692A>T	Alamut	0.406	1812	0.3137	1473	0.7011	-	GXYLT1:c.697T>A, GXYLT1:c.792T>A	p.?	stop-gained	GXYLT1
-?.	(-?) 12	Heterozygous	g.424969694A>T	Alamut	5.264	1808	0.3130	1471	0.7001	-	GXYLT1:c.697T>A, GXYLT1:c.790T>A	p.(Tyr233Asn), p.(Tyr264Asn)	missense	GXYLT1
-?.	(-?) 12	Heterozygous	g.42499701C>A	Alamut	-0.623	1842	0.1819	1492	0.7101	-	GXYLT1:c.690G>T, GXYLT1:c.783G>T	p.(Arg230Ser), p.(Arg261Ser)	missense	GXYLT1
-?.	(-?) 12	Heterozygous	g.42499711C>A	Alamut	4.733	1859	0.3218	1507	0.7173	-	GXYLT1:c.680G>T, GXYLT1:c.773G>T	p.(Arg227Leu), p.(Arg258Leu)	missense	GXYLT1
-?.	(-?) 12	Heterozygous	g.42499738T>C	Alamut	3.696	1887	0.3267	1534	0.7301	-	GXYLT1:c.653A>G, GXYLT1:c.746A>G	p.(Glu218Gly), p.(Glu249Gly)	missense	GXYLT1
-?.	(-?) 12	Heterozygous	g.42499739C>T	Alamut	6.375	1905	0.3298	1547	0.7363	-	GXYLT1:c.652G>A, GXYLT1:c.745G>A	p.(Glu218Lys), p.(Glu249Lys)	missense	GXYLT1
-?.	(-?) 12	Homozygous	g.132313098..132313099insGCT...	Alamut	-0.53	2453	0.4247	1607	0.7649	-	MMP17:c.59_60insGCT...	p.?	coding	MMP17
-?.	(-?) 15	Homozygous	g.23086365..23086367del	Alamut	0.391	2007	0.3475	810	0.3855	BEWEG	NIPA1:c.45_47del, NIPA1:c.-48..-453..-48..-455del	p.?, p.(=)	coding, intron	NIPA1
-?.	17	Homozygous	g.44408795G>A	Alamut	-0.405	1854	0.3210	1493	0.7106	-	ARL17B:c.259+21391C>T, LRRK37A:c.4152G>A	p.?	intron, coding-synonymous	-

Only (!) with informed consent
(de novo, X-linked, recessive)

Variant coloring

19 entries on 1 page. Showing entries 1 - 19.

	Effect	Chr	Allele	DNA change (genomic)	Alamut	PhyloP	#Ind. w/ var.	Var. ind. ratio	#Ind. w/ var & dis.	Var. dis. ind. ratio	Associated diseases	DNA change (cDNA)	Protein	GVS function	OMIM	
	-?.	2	Homozygous	g.20867122_20867123insGGC...	Alamut	0.064	361	0.0625	311	0.1480	-	GDF7c.c.123_124insGGC...	p.?	coding	GDF	
	-?.	(?)	2	Heterozygous	g.70524451C>T	Alamut	1.673	1099	0.1903	853	0.4060	-	FAM136A:c.387G>A	p.(Met129Ile)	missense	-
	-?.	(?)	2	Heterozygous	g.70524477G>C	Alamut	3.466	717	0.1241	550	0.2618	-	FAM136A:c.361C>G	p.(Leu121Val)	missense	-
	-?.	3	Homozygous	g.42251580_42251581insGGA...	Alamut	-0	855	0.1480	635	0.3022	-	TRAK1:c.1892_1893insGGA...	p.(=), p.?	intron, codingComplex	TRAK	
	-?.	(?)	3	Homozygous	g.133969437_133969438insG	Alamut	0.144	2114	0.3660	1476	0.7025	-	RYK:c.59_60insC	p.?	frameshift	RYK
	-?.	(?)	3	Homozygous	g.133969487_133969488insC	Alamut	-0.231	1055	0.1827	759	0.3613	-	RYK:c.9_10insG	p.?	frameshift	RYK
	-?.	(?)	5	Heterozygous	g.60628181_60628182insCG	Alamut	1.218	291	0.0504	240	0.1142	ID	ZSWIM6:c.82_83insGCG	p.?	coding	-
	-?.	(?)	5	Homozygous	g.60628575_60628589del	Alamut	0.196	287	0.0497	284	0.1352	ID	ZSWIM6:c.476_490del	p.?	coding	-
	??.	9	Homozygous	g.129854097_129854098del	Alamut	6.025	2	0.0003	2	0.0010	-	ANGPTL2:c.1133_1134del	p.?	frameshift	ANGPTL2	
	-?.	(?)	12	Heterozygous	g.424996907>C	Alamut	3.696	1814	0.3141	1477	0.7030	-	GXYLT1:c.701A>G, GXYLT1:c.794A>G	p.(Tyr234Cys), p.(Tyr265Cys)	missense	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499692A>T	Alamut	0.406	1812	0.3137	1473	0.7011	-	GXYLT1:c.699T>A, GXYLT1:c.792T>A	p.?	stop-gained	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499694A>T	Alamut	5.264	1808	0.3130	1471	0.7001	-	GXYLT1:c.697T>A, GXYLT1:c.790T>A	p.(Tyr233Asn), p.(Tyr264Asn)	missense	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499701C>A	Alamut	-0.623	1842	0.3189	1492	0.7101	-	GXYLT1:c.690G>T, GXYLT1:c.783G>T	p.(Arg230Ser), p.(Arg261Ser)	missense	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499711C>A	Alamut	4.733	1859	0.3218	1507	0.7173	-	GXYLT1:c.680G>T, GXYLT1:c.773G>T	p.(Arg227Leu), p.(Arg258Leu)	missense	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499738T>C	Alamut	3.696	1887	0.3267	1534	0.7301	-	GXYLT1:c.653A>G, GXYLT1:c.746A>G	p.(Glu218Gly), p.(Glu249Gly)	missense	GXYLT1
	-?.	(?)	12	Heterozygous	g.42499739C>T	Alamut	6.375	1905	0.3298	1547	0.7363	-	GXYLT1:c.652G>A, GXYLT1:c.745G>A	p.(Glu218Lys), p.(Glu249Lys)	missense	GXYLT1
	-?.	(?)	12	Homozygous	g.132313098_132313099insGCT...	Alamut	-0.53	2453	0.4247	1607	0.7649	-	MMP17:c.59_60insGCT...	p.?	coding	MMP17
	-?.	(?)	15	Homozygous	g.23086365_23086367del	Alamut	0.391	2007	0.3475	810	0.3855	BEWEG	NIPA1:c.45_47del, NIPA1:c.-48+453_48+455del	p.?, p.(=)	coding, intron	NIPA1
	-?.	17	Homozygous	g.44408795G>A	Alamut	-0.405	1854	0.3210	1493	0.7106	-	ARLL7B:c.259+21391C>T, LRRK37A:c.4152G>A	p.(=)	intron, coding-synonymous	-	

25 per page ▾ Legend

- variants colored based on predicted effect

- red = nonsense, frame shift, splicing*
- orange = missense, in-frame coding*
- green = silent, in UTR, intronic*
- blue = more than 1 predicted effect*

Linking to other software



1 entry on 1 page. Showing entry 1.

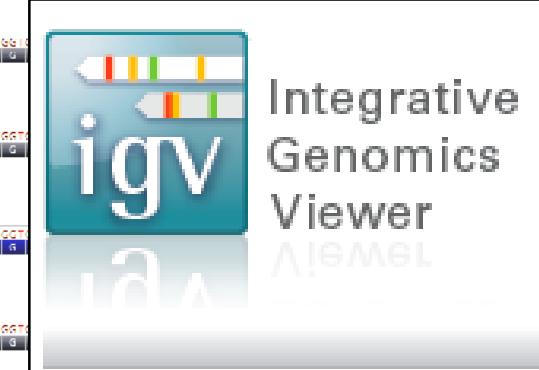
Curation status	Effect	Chr	Allele	DNA change (genomic)	Alamut	PhyloP	#Ind. w/ var.	Var. ind. ratio	#Ind. w/ var & dis.	Var. dis. ind. ratio	Associated diseases	DNA change (cDNA)	Protein	GVS function
Requires Confirmation	+/?	12	Heterozygous	g.49579770C>T	Alamut	5.702	1	0.0002	1	0.0005	HART, ID	TUBA1A:c.379G>A	p.(Asp127Asn)	missense

25 per page

Legend



gene



BAM file

Variant details

Individual ID	00000003 (Non public)
Chromosome	16
Allele	Heterozygous - Maternal (inferred)
Affects function (reported)	Not curated
Affects function (concluded)	Not curated
Curation status	-
Confirmation status	-
DNA change (genomic) (Relative to hg19 / GRCh37)	g.69362995C>T
Reference	-
DB-ID	chr16_000370 See all 100 reported entries
dbSNP ID	-
Sequencing quality	2118.16
Genotype quality	99
Sequencing filter	pass
Read depth	104
Read depth Ref	51
Read depth Alt	53
Read depth Alt (fraction)	0.50962
GATKcaller	UG,HC
Variant present in father	1
Genotype of father	0/0
Genotype quality of father	99
Read depth father	91
Read depth father Alt (fraction)	0
Variant present in mother	6
Genotype of mother	0/1
Genotype quality of mother	99
Read depth mother	61
Read depth mother Alt (fraction)	0.60656
GoNL AF	-
GoNL AF (old)	-
EVS AF	0
1000G AF	-
ExAC AF	-
Alamut link	-
HGMD reference	-
HGMD association	-
Phast conservation	1
PhyloP conservation	5.242
CADD Phred	35
CADD Raw	5.53228
INDB Count UG	-
INDB Count HC	-
INDB Global Samples	-
INDB Count Out Of Panel Hom	-
INDB Count In Panel Het	-
INDB Count Out Of Panel Het	-
INDB Count In Panel Hom	-
INDB In Panel Samples	-
INDB Count Global Hom	-
INDB Count Global Het	-
INDB Out Of Panel Samples	-
Variant remarks	-
Automatic mapping	Off
Average frequency (large NGS studies)	Variant not found in online data sets
Owner	LOVD
Variant data status	Non public
Created by	LOVD
Date created	2017-10-06 10:32:50
Last edited by	N/A
Date last edited	N/A

Options ▾

Summary annotations



Annotations that may be applicable to any instance of a particular variant can be stored in a summary annotation record. Click here to create a summary annotation record for this variant.

Classification	As reported classification	As final classification
Not curated	96	96
VUS	0	0
Pathogenic	0	0
Likely pathogenic	0	0
Likely benign	2	4
Benign	2	0

Observation Counts

There is no existing Observation Counts data | [Generate Data](#)

Upload attachments

Upload a file Choose File No file chosen
 File type -- select --

Variant on transcripts

Gene	Transcript	DNA change (cDNA)	Position	RNA change	Protein	GVS function
VPS4A	NM_013245.2	c.*4783C>T	6097	r(=)	p.(=)	utr-3
PDF	NM_022341.1	c.662G>A	662	r(?)	p.(Gly221Asp)	missense
COG8	NM_032382.4	c.*114G>A	1953	r(=)	p.(=)	utr-3

Change filters

Screenings

Screening ID	Panel coverage	Panel coverage (father)	Panel coverage (mother)	Curation progress	Variants found	Analysis status
0000000100	0.965984	0.971283	0.970241	0 of 0 variants curated	13892	In progress

Run analyses

Gene panel analyses

All analyses

De novo (v1)	Time	Var left
Filter		
apply_selected_gene_panels	0s	625
ID		
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_2	0s	263
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_2	0s	209
remove_with_any_frequency_1000G	0s	186
remove_with_any_frequency_dbSNP	0s	87
remove_with_any_frequency_goNL	0s	83
remove_with_any_frequency_EVS	0s	80
is_present_mother_lte_4	0s	10
is_present_father_lte_4	0s	1
is_present_mother_1	0s	1
is_present_father_1	0s	1
remove_intronic_distance_gt_8	0s	1
remove_intronic_distance_gt_2	0s	1
remove_by_function_utr3	0s	1
remove_by_function_utr5	0s	1
remove_by_function_utr_or_intronic	0s	1
remove_by_function_coding_synonymous	0s	1
remove_by_function_utr_or_intronic_or_synonymous	0s	1

Recessive (gene panel) (v1)	Time	Var left
Filter		
apply_selected_gene_panels	0s	625
ID		
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_5	0s	310
remove_by_indb_count_ug_gte_5	0s	277
remove_by_indb_count_hc_gte_2	0s	250
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_3	0s	214
remove_intronic_distance_gt_8	0s	214
remove_intronic_distance_gt_2	0s	214
remove_by_function_utr3	0s	122
remove_by_function_utr5	0s	77
remove_by_function_utr_or_intronic	0s	66
remove_by_function_coding_synonymous	0s	46
remove_by_function_utr_or_intronic_or_synonymous	0s	42
select_homozygous_or_compound_heterozygous	0s	11

De novo (v1 modified)	Time	Var left
Filter		
apply_selected_gene_panels	0s	625
ID		
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_2	0s	263
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_2	0s	209
remove_with_any_frequency_1000G	-	-
remove_with_any_frequency_dbSNP	-	-
remove_with_any_frequency_goNL	-	-
remove_with_any_frequency_EVS	-	-
is_present_mother_lte_4	0s	43
is_present_father_lte_4	0s	1
is_present_mother_1	0s	1
is_present_father_1	0s	1
remove_intronic_distance_gt_8	0s	1
remove_intronic_distance_gt_2	0s	1
remove_by_function_utr3	0s	1
remove_by_function_utr5	0s	1
remove_by_function_utr_or_intronic	0s	1
remove_by_function_coding_synonymous	0s	1
remove_by_function_utr_or_intronic_or_synonymous	0s	1

4 filters disabled

Variant classification

Screenings

Screening ID	Template	Technique	Trio check: De novo	Trio check: Mendelian	Panel coverage	Panel coverage (father)	Panel coverage (mother)
0000000526	DNA	SEQ-NG	-	-	0.468468	-	-
De novo			X / ?	X / ?	X / ?	X / ?	X / ?
Filter						Time	Var left
remove_not_in_gene_panel						0s	228
remove_by_quality_lte_100						0s	88
remove_by_indb_count_hc_gte_2						0s	51
remove_by_indb_count_ug_gte_2						0s	40
remove_with_any_frequency_gt_2						0s	35
remove_with_any_frequency_1000G						0s	30
remove_with_any_frequency_dbSNP						0s	10
remove_with_any_frequency_goNL						0s	10
remove_with_any_frequency_EVS						0s	6
is_present_mother_lte_4						0s	0
is_present_father_lte_4						0s	0
is_present_mother_1						0s	0
is_present_father_1						0s	0
remove_intronic_distance_gt_8						0s	0
remove_intronic_distance_gt_2						0s	0
remove_by_function_utr3						0s	0
remove_by_function_utr5						0s	0
remove_by_function_utr_or_intronic						0s	0
remove_by_function_coding_synonymous						0s	0
remove_by_function_utr_or_intronic_or_synonymous						0s	0
remove_by_function_utr_or_intronic_or_synonymous						0s	0
Filter						Time	Var left
chromosome_X						0s	98
remove_not_in_gene_panel						0s	13
remove_by_quality_lte_100						0s	5
remove_by_indb_count_hc_gte_2						0s	2
remove_by_indb_count_ug_gte_2						0s	2
remove_with_any_frequency_gt_3						0s	2
is_present_father_lte_4						0s	0
remove_intronic_distance_gt_8						0s	0
remove_intronic_distance_gt_2						0s	0
remove_by_function_utr3						0s	0
remove_by_function_utr5						0s	0
remove_by_function_utr_or_intronic						0s	0
remove_by_function_coding_synonymous						0s	0
remove_by_function_utr_or_intronic_or_synonymous						0s	0
select_homozygous_or_compound_heterozygous						0s	4
Filter						Time	Var left
remove_not_in_gene_panel						0s	228
remove_by_quality_lte_100						0s	88
remove_by_indb_count_hc_gte_5						0s	60
remove_by_indb_count_ug_gte_5						0s	48
remove_by_indb_count_hc_gte_2						0s	42
remove_by_indb_count_ug_gte_2						0s	40
remove_with_any_frequency_gt_3						0s	37
remove_intronic_distance_gt_8						0s	37
remove_intronic_distance_gt_2						0s	37
remove_by_function_utr3						0s	29
remove_by_function_utr5						0s	19
remove_by_function_utr_or_intronic						0s	17
remove_by_function_coding_synonymous						0s	14
remove_by_function_utr_or_intronic_or_synonymous						0s	13
select_homozygous_or_compound_heterozygous						0s	4

1 entry on 1 page. Showing entry 1.

25 pages ▾ Legend

- Select all 2 entries
- Unselect all
- Set variant effect to "Not curated"
- Set variant effect to "VUS"
- Set variant effect to "Pathogenic"
- Set variant effect to "Likely pathogenic"
- Set variant effect to "Likely benign"
- Set variant effect to "Benign"
- Set curation status
- Set confirmation status

y	Read depth Alt (fraction)	PhyloP conservation	Alamut link	HGMD association
2.77	0.47945	5.702	Alamut	Lissencephaly with cerebellar hypoplasia

5 classes

Variant reporting

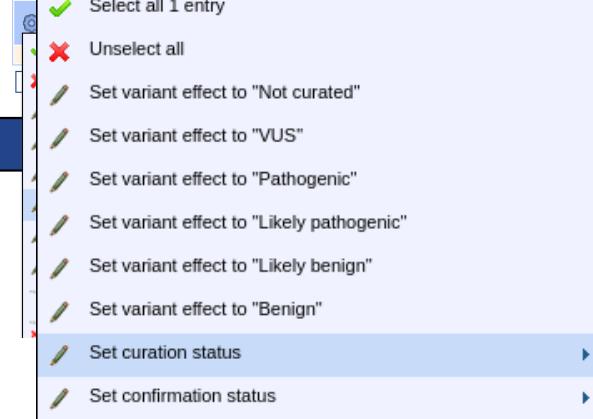
Screenings

Screening ID	Template	Technique	Trio check: De novo	Trio check: Mendelian	Panel coverage	Panel coverage (father)	Panel coverage (mother)																																																														
0000000526	DNA	SEQ-NG	-	-	-	0.468468	-																																																														
De novo			X / ?	X-linked recessive	X / ?	Recessive (gene panel)	X / ?																																																														
<table border="1"> <thead> <tr> <th>Filter</th> <th>Time</th> <th>Var left</th> </tr> </thead> <tbody> <tr><td>remove_not_in_gene_panel</td><td>0s</td><td>228</td></tr> <tr><td>remove_by_quality_lte_100</td><td>0s</td><td>88</td></tr> <tr><td>remove_by_indb_count_hc_gte_2</td><td>0s</td><td>51</td></tr> <tr><td>remove_by_indb_count_ug_gte_2</td><td>0s</td><td>40</td></tr> <tr><td>remove_with_any_frequency_gt_2</td><td>0s</td><td>35</td></tr> <tr><td>remove_with_any_frequency_1000G</td><td>0s</td><td>30</td></tr> <tr><td>remove_with_any_frequency_dbSNP</td><td>0s</td><td>10</td></tr> <tr><td>remove_with_any_frequency_goNL</td><td>0s</td><td>10</td></tr> <tr><td>remove_with_any_frequency_EVS</td><td>0s</td><td>6</td></tr> <tr><td>is_present_mother_lte_4</td><td>0s</td><td>0</td></tr> <tr><td>is_present_father_lte_4</td><td>0s</td><td>0</td></tr> <tr><td>is_present_mother_1</td><td>0s</td><td>0</td></tr> <tr><td>is_present_father_1</td><td>0s</td><td>0</td></tr> <tr><td>remove_intronic_distance_gt_8</td><td>0s</td><td>0</td></tr> <tr><td>remove_intronic_distance_gt_2</td><td>0s</td><td>0</td></tr> <tr><td>remove_by_function_utr3</td><td>0s</td><td>0</td></tr> <tr><td>remove_by_function_utr5</td><td>0s</td><td>0</td></tr> <tr><td>remove_by_function_utr_or_intronic</td><td>0s</td><td>0</td></tr> <tr><td>remove_by_function_coding_synonymous</td><td>0s</td><td>0</td></tr> <tr><td>remove_by_function_utr_or_intronic_or_synonymous</td><td>0s</td><td>0</td></tr> </tbody> </table>							Filter	Time	Var left	remove_not_in_gene_panel	0s	228	remove_by_quality_lte_100	0s	88	remove_by_indb_count_hc_gte_2	0s	51	remove_by_indb_count_ug_gte_2	0s	40	remove_with_any_frequency_gt_2	0s	35	remove_with_any_frequency_1000G	0s	30	remove_with_any_frequency_dbSNP	0s	10	remove_with_any_frequency_goNL	0s	10	remove_with_any_frequency_EVS	0s	6	is_present_mother_lte_4	0s	0	is_present_father_lte_4	0s	0	is_present_mother_1	0s	0	is_present_father_1	0s	0	remove_intronic_distance_gt_8	0s	0	remove_intronic_distance_gt_2	0s	0	remove_by_function_utr3	0s	0	remove_by_function_utr5	0s	0	remove_by_function_utr_or_intronic	0s	0	remove_by_function_coding_synonymous	0s	0	remove_by_function_utr_or_intronic_or_synonymous	0s	0
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1 entry on 1 page. Showing entry 1.

25 per page

Legend



Read depth Alt (fraction)	PhyloP conservation	Alamut link	HGMD association
77	0.47945	5.702 Alamut	Lissencephaly with cerebellar hypoplasia

- Variant of Interest
- Requires Confirmation
- Confirmed
- Artefact
- Clear curation status**

4 classes, flexible

- Different user levels (multi-user analysis)

- *Read only account*
- *Analyzer*
- *Manager*
- *Administrator*

Create a new summary annotation record

To create a new summary annotation record, please fill out the form below.

Affects function	Likely benign
Remarks (optional)	<input type="text"/>
Interpretation (optional)	<input type="text"/> IF: High frequency.
Enter your password for authorization	
<input type="button" value="Create summary annotation record"/>	

- Variant annotation record

- *Variant-specific information, independent from observation*
- *Fields can be easily customized*

- Easy gene panel definition

- *Can be combined with quick custom gene panels per individual*

- Open source software
 - See: www.lovd.nl/plus/
 - *Preparing new release*
 - *Preparing documentation*
- Support
 - *May be required for*
 - Installation, customization or handling different file formats
 - Training
 - Adding / modifying analyses
 - *Offered by:*

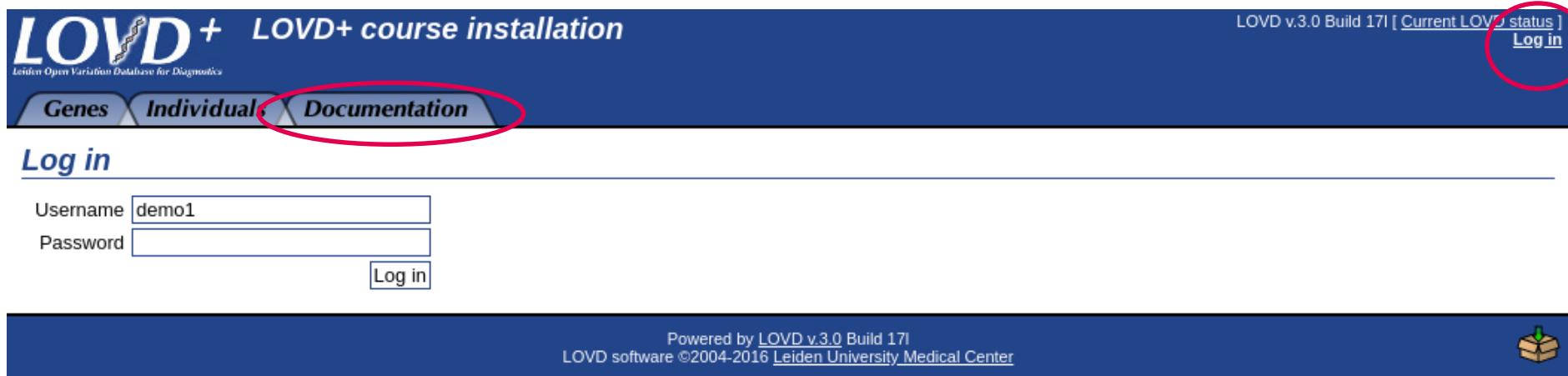
For further information, please contact The Genome Reports Company:

Email: support@genechipreports.org

Website: www.genechipreports.org

[http://courses.lovd.nl/LOVD+/
Practical at the Documentation tab!!](http://courses.lovd.nl/LOVD+/)

Practical at the Documentation tab!!



LOVD + LOVD+ course installation

Leiden Open Variation Database for Diagnostics

Genes Individual Documentation

Log in

Username: demo1

Password:

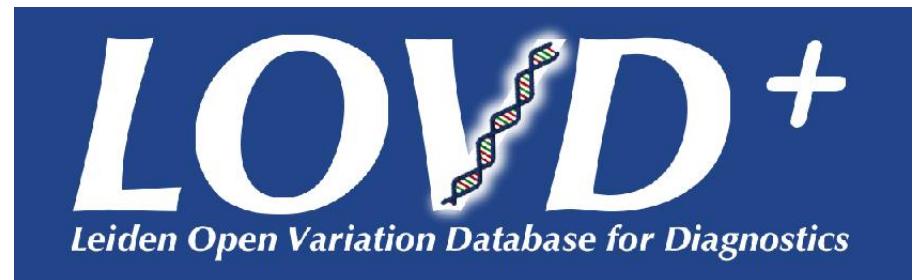
Log in

Powered by LOVD v.3.0 Build 171
LOVD software ©2004-2016 Leiden University Medical Center

Username: demo# (# = your participant ID)
Password: demo

Then press enter key or click "Log in" button

Questions ?



Ivo Fokkema

Claudia Ruivenkamp

Johan den Dunnen

