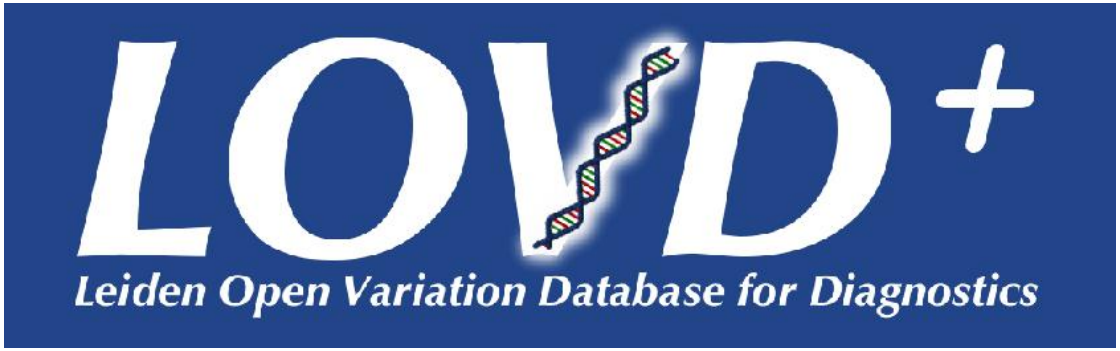
Leiden University Medical Center

NGS analysis using LOVD+



Ivo Fokkema

Claudia Ruivenkamp

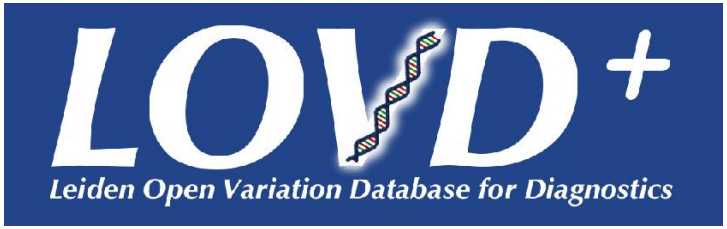
Johan den Dunnen

Analysis NGS data

LOVD+ Leiden Open Variation Database for Diagnostics

- 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*

Analysis NGS data



- 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

LOVD plus — Leiden Open Variation Database for Diagnostics

LDGA Leiden exome diagnostics (production installation)

LOVD v.3.0 Build 17q [Current LOVD status]
Welcome, Ivo F.A.C. Fokkema
Your account | Log out

Genes | Individuals | Diseases | Users | Setup | Documentation

View individuals

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

25 per page ▼ Legend « First ‹ Prev ... 82 83 84 85 86 87 88 89 90 91 92 93 94 95 96 97 98 99 100 101 102 ... Next › Last »

Individual ID	ZIS ID	Lab-ID	Gender	Disease	Gene panels	Custom panel	Analysis status	Analysis by	Analysis started	Analysis approved by	Analysis approved
							!="Confirmed"				
00002322	3487510	Child_12259656	F	ID	GROEI	-	In progress	Quint	2017-09-14	-	
00002323	6517467	Child_12318677	M	ID	ID	-	Closed	Quint	2017-11-10	Claudia Ruivenkamp	2018-02-02
00002324	9049502	Child_12218451	F	ID	ID	KMT2D, KDM6A, GLI3	Closed	Quint	2017-10-24	Mariëtte Hoffer	2017-11-01
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	-	
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, LFNG, 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, LMCD1, LTBP2, 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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Powered by LOVD v.3.0 Build 17q
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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	
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

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

Log entries

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

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)

Filter	Time	Var left
apply_selected_gene_panels ID	0s	625
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

X-linked recessive (v1)

Filter	Time	Var left
chromosome_X	0s	354
apply_selected_gene_panels ID	0s	88
remove_by_quality_lte_100	0s	52
remove_by_indb_count_hc_gte_2	0s	44
remove_by_indb_count_ug_gte_2	0s	38
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 ID	0s	625
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

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

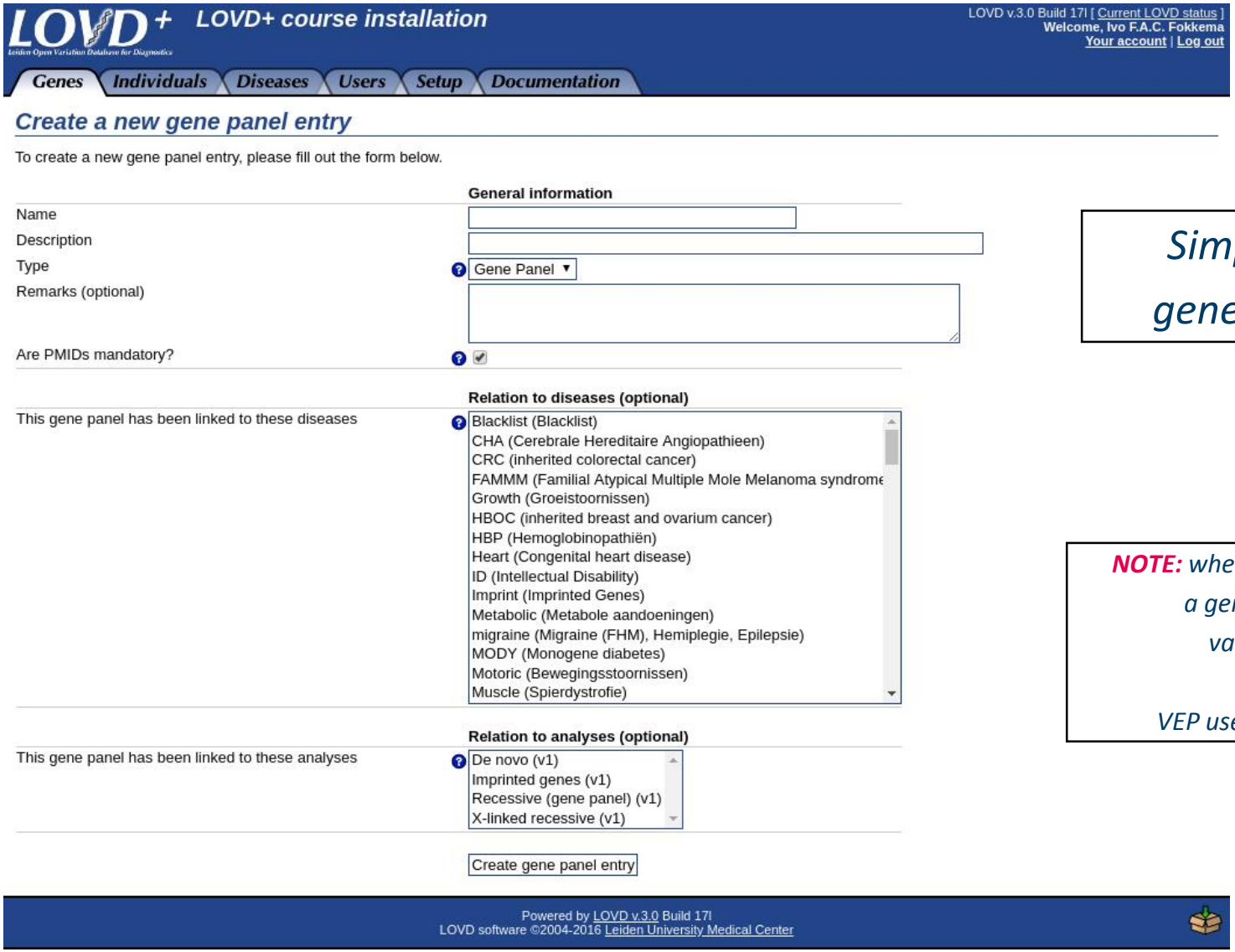
25 per page | Legend

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
-	./.	17	Heterozygous	g.78063998G>A	Alamut	-1.951	100	1.0000	100	1.0000	ID	CCDC40:c.2893G>A
-	./.	17	Heterozygous	g.78064145_78064146insCAC	Alamut	0	100	1.0000	100	1.0000	ID	CCDC40:c.3040_3041insCA
-	./.	19	Heterozygous - Paternal (inferred)	g.11314765C>T	Alamut	3.907	100	1.0000	100	1.0000	Heart; ID; Adams-Oliver syndrome 2	DOCK6:c.5227G>A
-	./.	19	Heterozygous - Maternal (inferred)	g.11358788G>A	Alamut	0.996	100	1.0000	100	1.0000	Heart; ID; Adams-Oliver syndrome 2	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

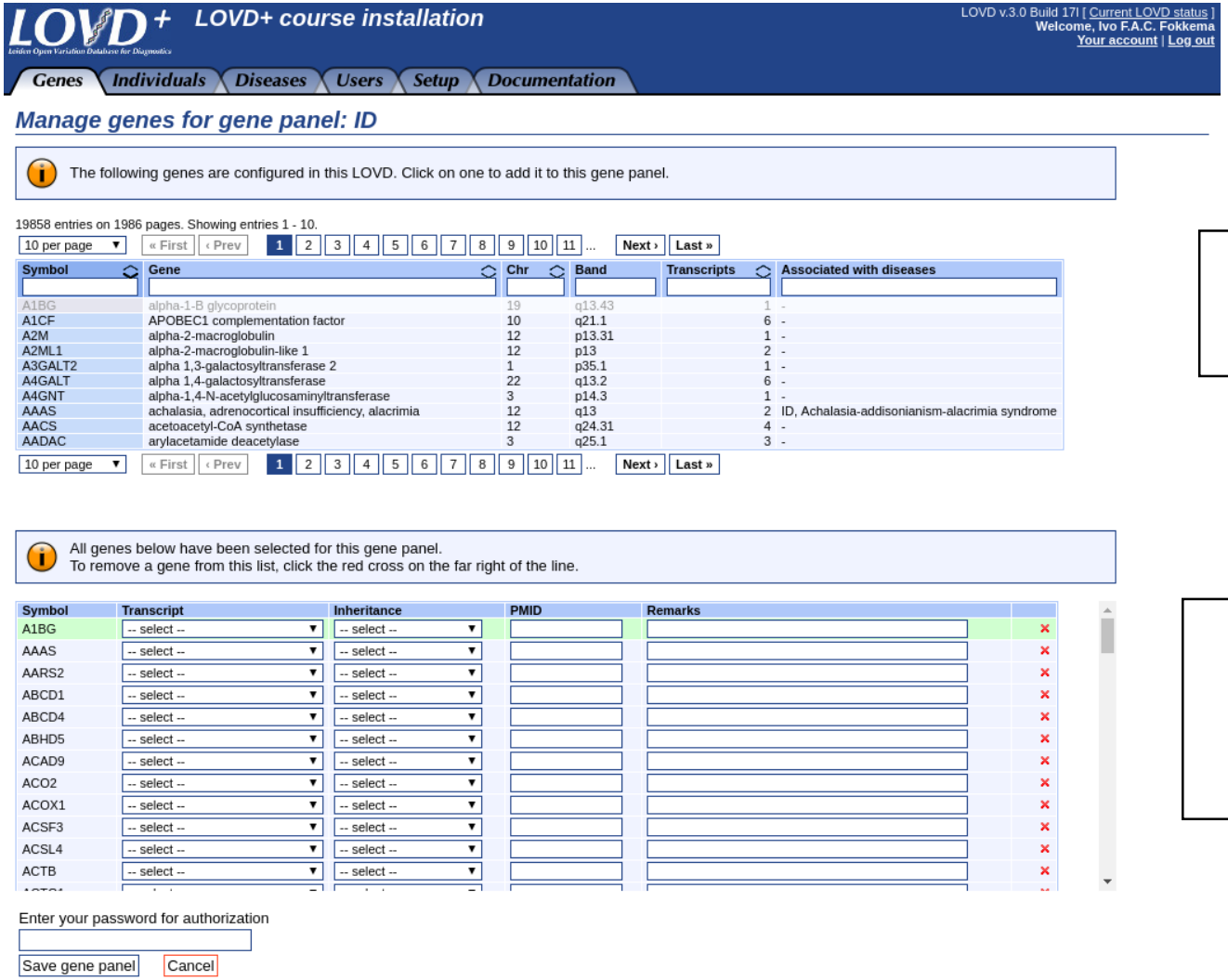


Simple forms for gene panel design

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

VEP uses a gene +/- 5000 bp.

Gene panel design

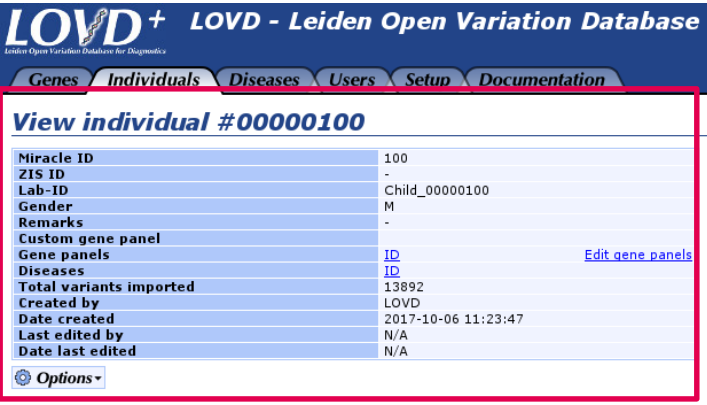


LOVD+ provides full gene panel history

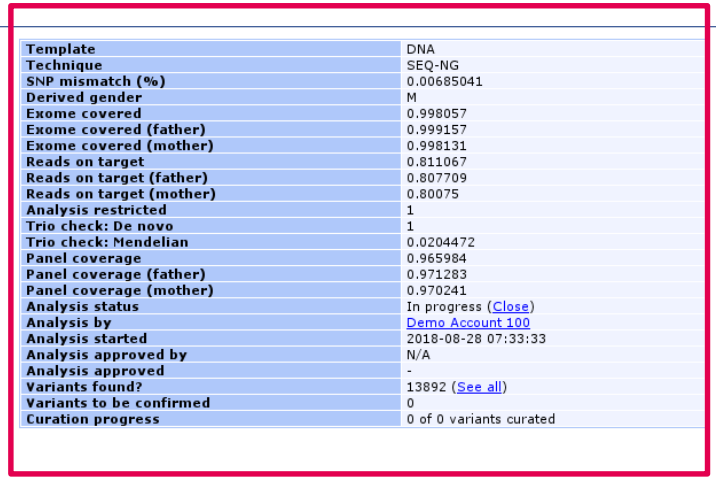
***NOTE:** whether variants are linked to a gene is defined during variant annotation.*

VEP uses a gene +/- 5000 bp.

Details NGS analysis



details Individual



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
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
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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

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	-
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Curation progress	0 of 0 variants curated

coverage

Details WES data

Details NGS

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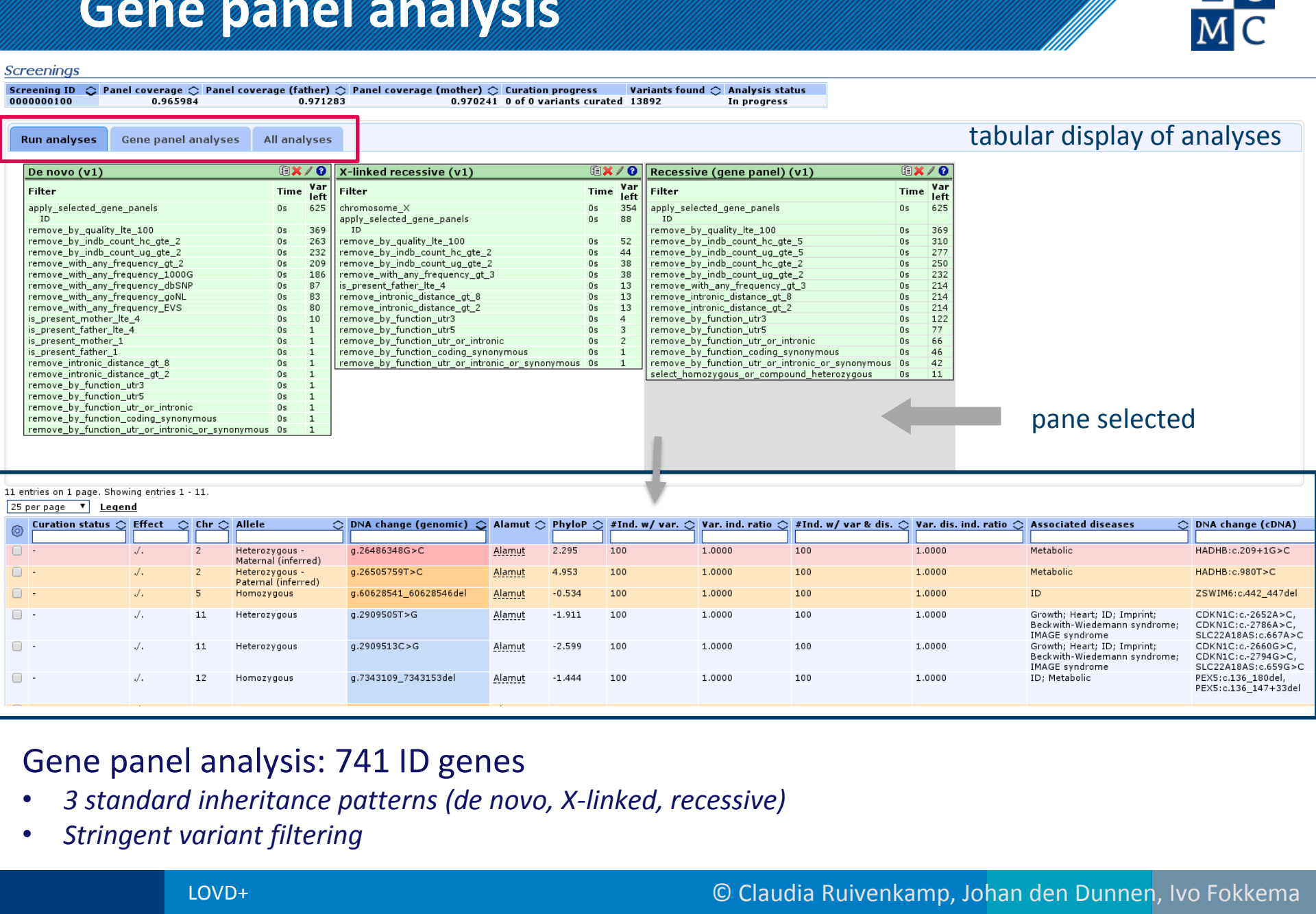
Analysis by	Demo Account 100
Analysis started	2018-08-28 07:33:33
Analysis approved by	N/A
Analysis approved	-
Variants found?	13892 (See all)
Variants to be confirmed	0
Curation progress	0 of 0 variants curated

Information and controls

Details WES data

Gene panel analysis



Gene panel analysis: 741 ID genes

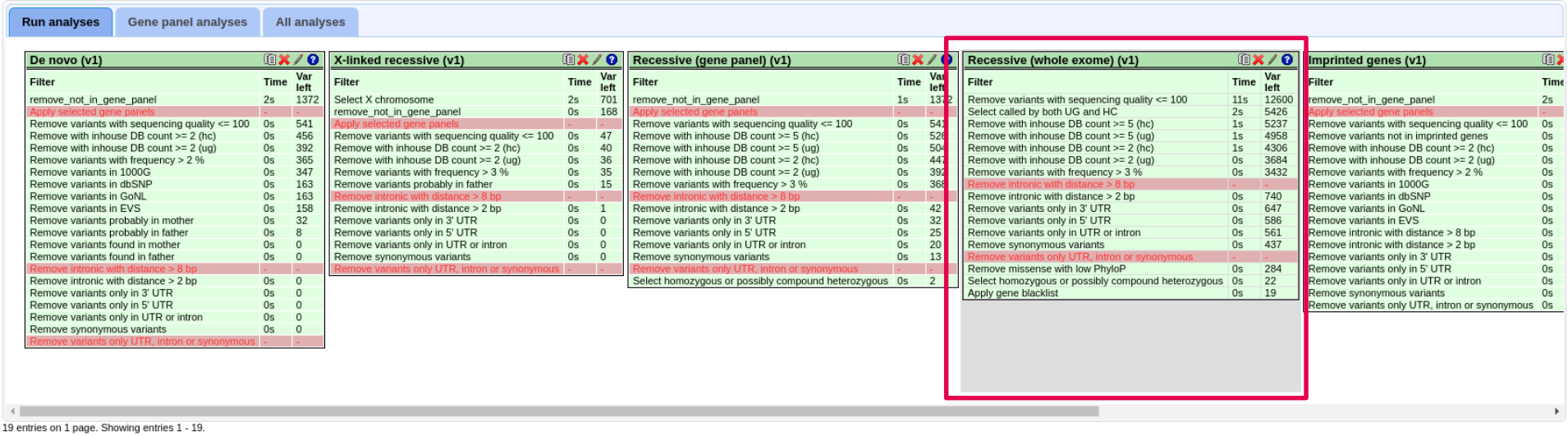
- *3 standard inheritance patterns (de novo, X-linked, recessive)*
- *Stringent variant filtering*

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



Only (!) with informed consent

(*de novo*, X-linked, recessive)

Variant coloring

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	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..., TRAK1:c.1844_1845insGGA..., TRAK1:c.1963+103_1963+104insGGA..., TRAK1:c.2066_2067insGGA...	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_60628182insGCG	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	ANG
-?/. (-?)	12	Heterozygous	g.42499690T>C	Alamut	3.696	1814	0.3141	1477	0.7030	-	GXYLT1:c.701A>G, GXYLT1:c.794A>G	p.(Tyr234Cys), p.(Tyr265Cys)	missense	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	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	GXYI
-?/. (-?)	12	Homozygous	g.132313098_132313099insGCT...	Alamut	-0.53	2453	0.4247	1607	0.7649	-	MMP17:c.59_60insGCT...	p.?	coding	MMP
-?/. (-?)	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	NIPA
-?/.	17	Homozygous	g.44408795G>A	Alamut	-0.405	1854	0.3210	1493	0.7106	-	ARL17B:c.259+21391C>T, LRRC37A: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

alamut visual

1 entry on 1 page. Showing entry 1.

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)	Protein	GVS function
Requires Confirmation	+?/?	12	Heterozygous	g.49579770C>T	Alamut http://127.0.0.1:10000/show?request=12:49579770C>T	5.702	1	0.0002	1	0.0005	HART; ID	TUBA1A:c.379G>A	p.(Asp127Asn)	missense

25 per page ▼ Legend

gene

Integrative Genomics Viewer

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

annotations

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 --

Upload file

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)

Filter	Time	Var left
apply_selected_gene_panels ID	0s	625
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)

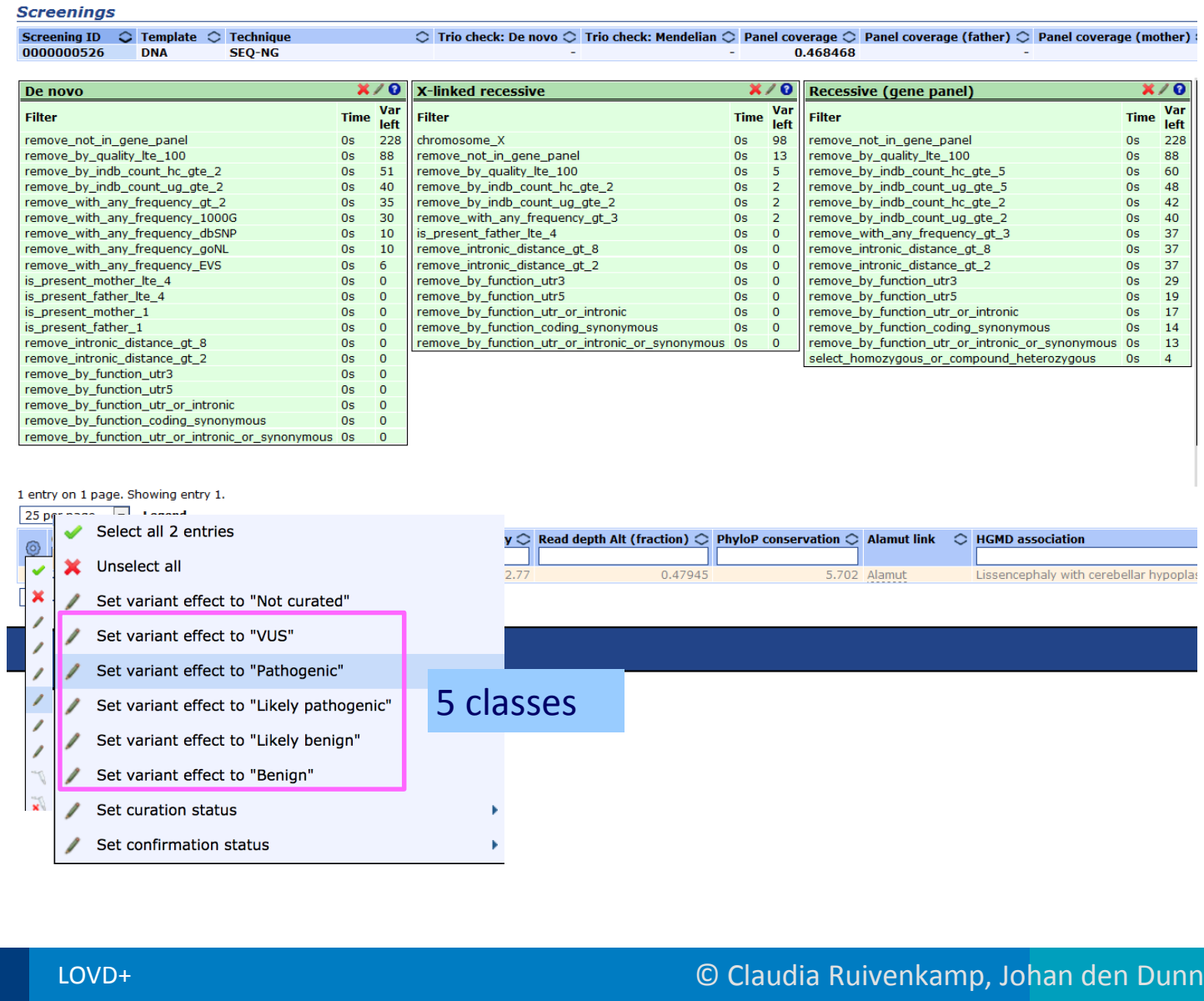
Filter	Time	Var left
apply_selected_gene_panels ID	0s	625
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_5	0s	310
remove_by_indb_count_ug_gte_5	0s	277
remove_by_indb_count_hc_gte_2	0s	250
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_3	0s	214
remove_intronic_distance_gt_8	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)

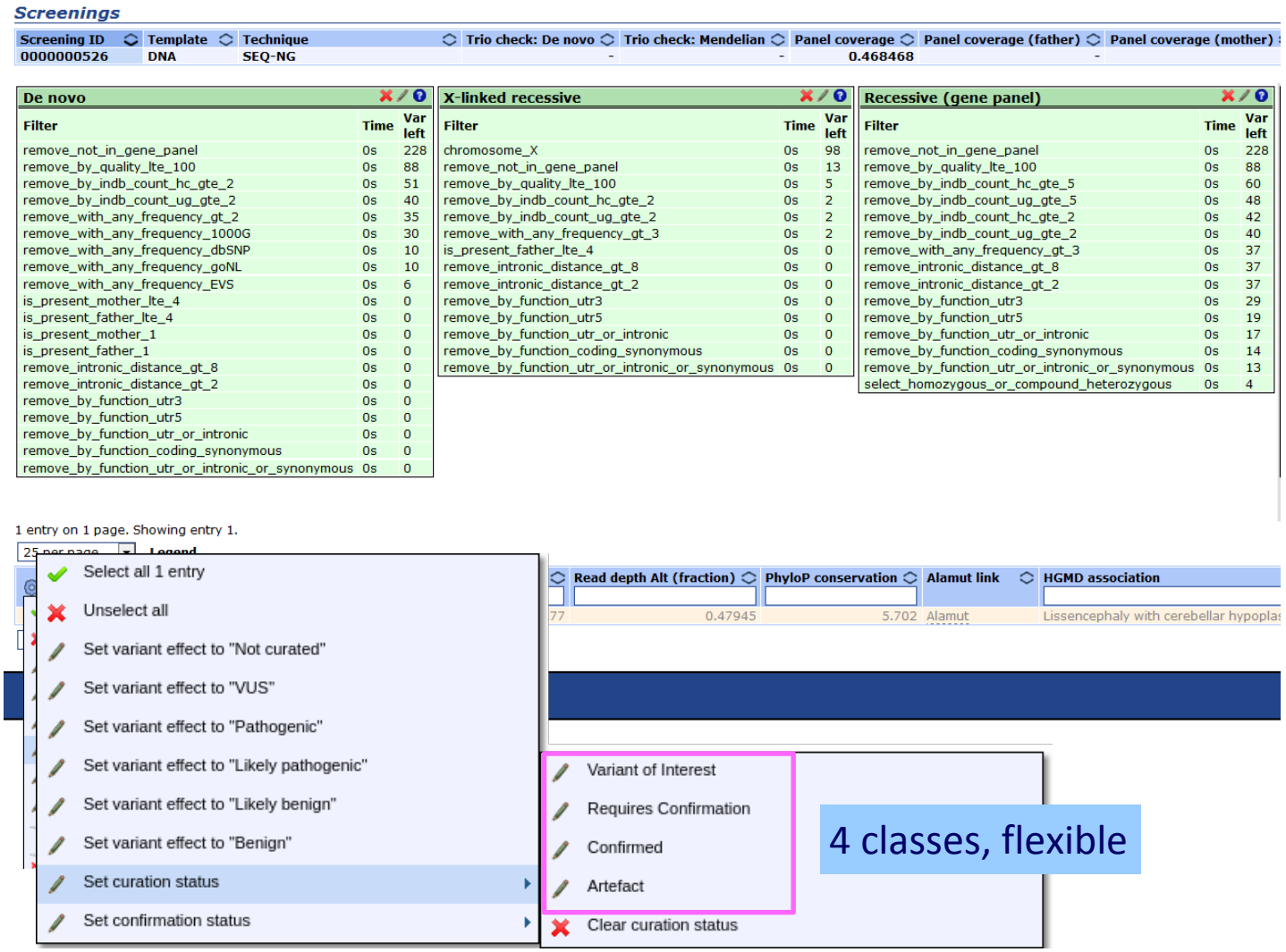
Filter	Time	Var left
apply_selected_gene_panels ID	0s	625
remove_by_quality_lte_100	0s	369
remove_by_indb_count_hc_gte_2	0s	263
remove_by_indb_count_ug_gte_2	0s	232
remove_with_any_frequency_gt_2	0s	209
remove_with_any_frequency_1000G	-	-
remove_with_any_frequency_dbSNP	-	-
remove_with_any_frequency_goNL	-	-
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

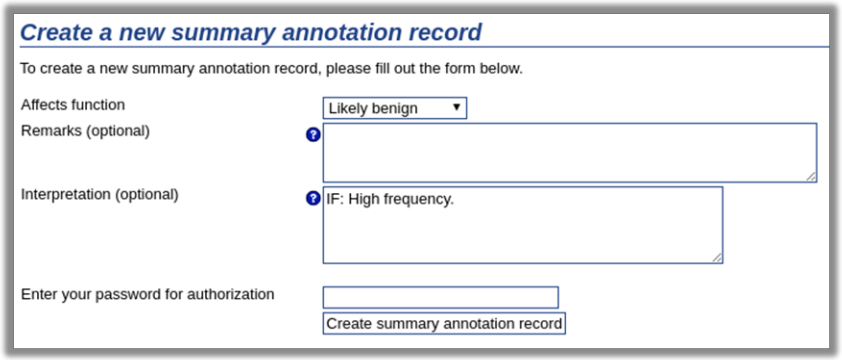


Variant reporting



And more …

- Different user levels (multi-user analysis)
  - *Read only account*
  - *Analyzer*
  - *Manager*
  - *Administrator*



- 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

- 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:*

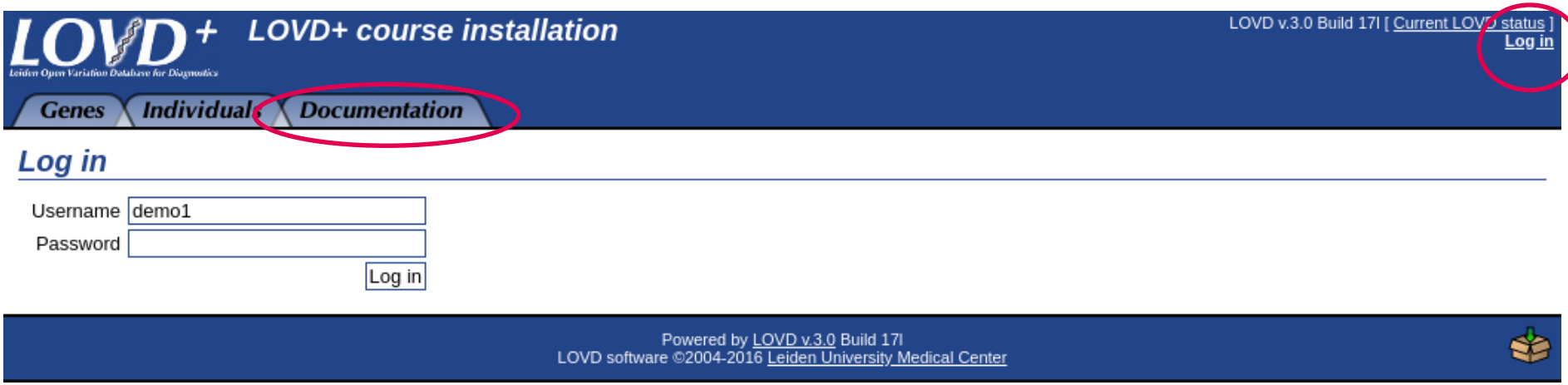
For further information, please contact The Genome Reports Company:

Email: support@geneticreports.org
Website: www.geneticreports.org

LOVD+ practical

http://courses.lovd.nl/LOVD+/

Practical at the Documentation tab!!



Username: demo# (# = your participant ID)

Password: demo

Then press enter key or click "Log in" button

Leiden University Medical Center

Questions ?

Ivo Fokkema

Claudia Ruivenkamp

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