COGR Updates WebEx

To participate in the live feedback survey please go to:
www.polleverywhere.com/cogrmanager699

OR

Text “COGRMANAGER699” to 37607

Aims of the COGR:

1. Design of variant assessment procedures
2. Data extraction and transfer
3. Data access and dissemination

Variant Assessment Tool (VAT)

- The VAT is an excel based tool to semi-automatic the process of variant assessment
- It helps streamline and standardize variant assessment
- The VAT provided by COGR is adapted from Partner's Healthcare
- SOP for the tool to be made available
- The VAT is available at: http://opengenetics.ca/resources/
- Note: The VAT takes advantage of the Alamut software currently with the goal of integretion with GeneInsight

Q: Will the VAT be a useful tool for your laboratory?

A- Yes, I am using it

B- Yes, but I haven’t yet implemented it

C- No, I have my own variant assessment protocol

D-No, it is too complicated/time consuming

E- No, I am not interested at this time

F- I am undecided

Q: What would be most helpful for you in regards to understanding the VAT?

A- I am comfortable using the VAT

B- I would like a WebEx demo

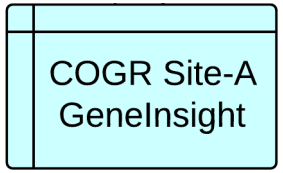
C- I would like a detailed SOP

D- I am not interested in using the VAT at this time

Please email us to set up a WebEx demo of the VAT if you are interested

COGR storage and sharing

- The COGR Consensus Database holds variants with consensus classifications across labs
- Sharing is optional at the discretion of the lab and only enabled upon request



Organization	Upload Status	Sharing status
Alberta Children's Hospital, Calgary AB	Uploading	-
British Columbia Cancer Agency, Vancouver BC	Pending	-
Children’s & Women’s Health Centre of BC, Vancouver BC	Pending	-
Children's Hospital of Eastern Ontario, Ottawa ON	Uploaded	Sharing
Credit Valley Hospital, Trillium Health Centre, Mississauga ON	Uploaded	Sharing
Impact Genetics Inc., Bowmanville ON	Pending	-
McGill University Health Complex, Montréal QC	Uploaded	-
SickKids Hospital and McLaughlin Centre, Toronto ON	Uploaded	Sharing
Hamilton Health Sciences, McMaster University, Hamilton ON	Pending	-
Memorial Health University Medical Center, St. John’s NL	Pending	-
Mount Sinai Hospital, University of Toronto, Toronto ON	Uploaded	Sharing
North York General Hospital, Toronto ON	Pending	-
Ontario Institute of Cancer Research (OICR), Toronto ON	Uploading	-
Kingston General Hospital, Queen’s University, Kingston ON	Uploaded	Sharing
Dept of Medical Genetics, University of Alberta, Edmonton AB	Uploading	-
Regional Health Authority, University of Manitoba, Winnipeg MB	Pending	-
Sainte-Justine Hospital, University of Montreal, Montréal QC	Pending	-
University Hospital, Western University, London ON	Pending	-
Women's College Hospital, University of Toronto, Toronto ON	Uploaded	Sharing

Currently Being Shared

Unique Genes:	Total Unique Variants
BRCA2	1194
BRCA1	959
APC	195
MYBPC3	145
MYH7	126
MSH2	108
MLH1	103
MSH6	100
MUTYH	54
SERPINA1	49
DSP	34
TNNT2	29
TNNI3	23
KRAS	22
TPM1	21
PKP2	19
DSG2	13
PMS2	11
PDGFRA	10
BRAF	7
CDH1	6
KIT	6
BMPR1A	3
SMAD4	3
STK11	2

- Intragenic variants: 3877
  - 3242 unique
- Genes: 25
- CNVs: 3339
- Clinically associated diseases: 13

* Note that the Unique Genes in the table do not include genes associated with the CNVS

Q: What are your plans for data sharing with COGR?

A- I am interested in sharing all of my variants

B- I am interested in sharing a subset of my variants

C- I am not interested in sharing my variants but I want to view other labs' data

D- I am not interested in sharing my variants but want the system to store my own variants

VariantWire

- Data-sharing network for clinical genetic testing labs to share variant and gene interpretations in real time
- Consortium supported by GeneInsight
- VariantWire will be presenting at the upcoming ACMG meeting

Participating Laboratories

Partners HealthCare Laboratory for Molecular Medicine
ARUP Laboratories
Mount Sinai School of Medicine Genetic Testing Laboratory
Mount Sinai Hospital in Toronto
Canadian Open Genetics Repository (consensus database)
Children's Hospital of Eastern Ontario

Variant Classification	#
Pathogenic	1,946
Likely Pathogenic	1,137
Uncertain Significance	4,536
Likely Benign	12,053
Benign	4,657
Resistant or Responsive*	57
Total Variants	24,386

Sharing Variants

- Permission must be explicitly given to the COGR and GeneInsight team to start sharing
- There are ~25000 variants being shared through VariantWire. To access this a secondary permission is needed- please email us if interested
- COGR can assist your laboratory upload variant information to ClinVar, please contact us if you are interested

COGR Variant comparisons

- Variants Identified in Multiple Labs: 522
- Agreements: 264
- Disagreements: 91
- Shared variants not classified by one lab: 167

Variant Disagreements	
Path -Ben*	3
Path -Likely Ben	2
Likely Path - Ben	0
Likely Path - Likely Ben	0
Path -VUS	16
VUS - Ben	16
Likely Path - VUS	1
VUS - Likely Ben	15
Path -Likely Path	6
Likely Ben - Ben	32

*These variants have been resolved

Examples of Variant Discrepancies

- Scenario 1
  - Lab A: Pathogenic
  - Lab B: Benign
- Scenario 2:
  - Lab A: VUS
  - Lab B: Pathogenic
- Scenario 3:
  - Lab A: Likely Benign
  - Lab B: Unclassified
  - Lab C: Pathogenic

Discrepancies can be dealt with in many ways:

- Group consensus discussion between all labs/participants
- Creation of a subcommittee
- Discrepancy reports to individual labs

Proposal: Monthly Discrepancies Reports

1. Monthly analysis of discrepant variants across sharing labs
2. Send discrepancy report to all sharing labs
3. Laboratories with discrepant variants to discuss with one another

- The current proposal for resolving discrepancies
- Currently used by ClinVar and VariantWire successfully
- Alternative options:
  - Consensus group discussion
  - Subcommittee

Q: How should COGR resolve discrepant variant information?

A- Discrepancy reports and direct contact only between labs that have discrepant variants classifications

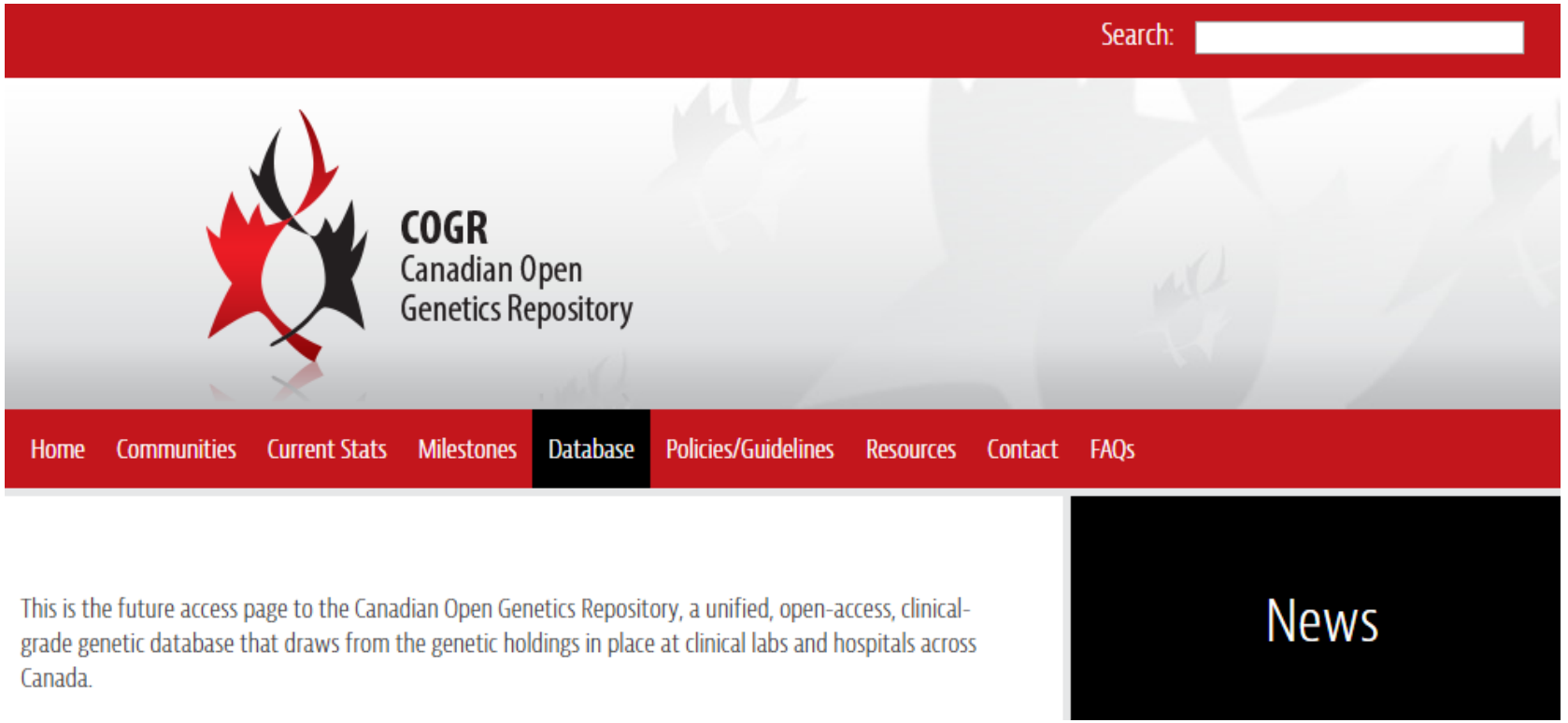
B- Have a subcommittee review discrepant variants

C- Have a group discussion between all COGR participants about discrepant variants

D- Other: ___

Website and public access

Similar to other initiatives like ClinVar COGR aims to post consensus level interpretations on a public website



Q: Please provide suggestions for criteria needed for a consensus variant classification agreement

Open Ended

Issues with Conesus Level Interpretations

- If one lab encounters a variant- is that considered a consensus?
  - What about 2 labs?
  - What is the threshold number of labs?
- Is there a need for a consensus committee?
  - Would that take too much time and resources?
  - Would we need different people to handle variants related to different diseases?
- If two labs have interpretations should evidence be combined?
  - What will be shown on the consensus database?

Would you be interested in attending regularly scheduled web meetings such as this one for COGR updates?

A- Yes-Every 1 or 2 months

B- Yes- Quarterly

C- Yes-Every 6-12 months

D- I am not interested at this time

Q: What are the biggest barriers to your participation in the COGR project and how can we help?

Open Ended

Recap using GeneInsight

- Searching for variants and shared variant information
- Viewing detailed shared variant information
- How to approve variants one by one and in bulk
- Upload Variants (not included)
- Saving (not included)

Search for variants and view shared variant information

Diseases/Drugs

Genes

Variants Create

Saved Searches

Parameters

Save Reset Search

Show Networked Labs Summary

Variant contains

Gene Name/Symbol contains

DSP

Disease/Drug/Area contains

Tests & Panels

Reports

References

Showing results for: Gene Name/Symbol contains DSP and Category = Likely Pathogenic

Variants (17)

Click header to sort. Control+Click (Cmd+Click on Mac) header to remove sort. Delete | Approve Proposed Interp. | Download

Locus	DNA	AA	Genomic	**	Region	Cat (Dis)	Cat Date	Rpt	Fam	Actions	dbSNP	ClinVar	Networked Lab
DSP	c.214C>T	p.Gln72X	g.7555994C>...	R	Ex 02	Lik Path (...	16.May.2008	1	1	Ed | Del			
DSP	c.478C>T	p.Arg160X	g.7559514C>...		Ex 04	Lik Path (...	30.Sep.2011	2	2	Ed | Del			
DSP	c.534_535insA	p.Gly179Argf...	g.7559570_7...		Ex 04	Lik Path (DCM)	21.Sep.2012	1	1	Ed | Del			Under Review - 1 Unknown Signif...
DSP	c.699G>A	p.Trp233X	g.7562986G...		Ex 05	Lik Path (DCM)	02.Jul.2012	2	1	Ed | Del			
DSP	c.712_713insA	p.Ile238Asnf...	g.7562999_7...		Ex 05	Lik Path (DCM)	17.Jul.2012	1	1	Ed | Del			
DSP	c.867C>A	p.Cys289X	g.7565681C>...		Ex 07	Lik Path (DCM)	08.Dec.2012	1	1	Ed | Del			
DSP	c.939+1G>A		g.7565754G...		In 07	Lik Path (...	02.May.2012	2	1	Ed | Del			
DSP	c.1146delT	p.Phe382Le...	g.7568019de...		Ex 10	Lik Path (DCM)	10.Dec.2012	1	1	Ed | Del			Unknown Signif... Likely Pathogen...
DSP	c.1273C>T	p.Arg425X	g.7568676C>...		Ex 11	Lik Path (...	01.Mar.2008	2	1	Ed | Del			
DSP	c.1650G>A	p.Trp550X	g.7570745G...	R	Ex 13	Lik Path (...	09.Oct.2009	1	1	Ed | Del			
DSP	c.2848_2849...	p.Ile950Asnf...	g.7577246_7...		Ex 20	Lik Path (DCM)	25.May.2012	1	1	Ed | Del			
DSP	c.2959T>C	p.Ser987Pro	g.7578093T>...	N	Ex 21	Lik Path (...	17.Aug.2009	2	1	Ed | Del			
DSP	c.3160_3169...	p.Lys1054Se...	g.7579583_7...		Ex 23	Lik Path (...	16.Nov.2007	6	1	Ed | Del			
DSP	c.3474_3475...	p.Glu1159Ar...	g.7579897_7...	N	Ex 23	Lik Path (...	*22.Mar.2012*	13	2	Ed | Del			Deleterious (... Likely Pathogen...
DSP	c.3829C>T	p.Gln1277X	g.7580252C>...		Ex 23	Lik Path (...	23.Dec.2011	1	1	Ed | Del			
DSP	c.4531C>T	p.Gln1511X	g.7580954C>...	N	Ex 23	Lik Path (...	15.Dec.2009	1	1	Ed | Del			
DSP	c.5428C>T	p.Gln1810X	g.7582923C>...		Ex 24	Lik Path (...	08.Apr.2008	1	1	Ed | Del			

Diseases/Drugs
Genes
Variants Create
Saved Searches
Parameters
Save Reset Search
Show Networked Labs Summary
Variant contains
Gene Name/Symbol contains
DSP
Disease/Drug/Area contains
Tests & Panels
Reports
References

Showing results for: Gene Name/Symbol contains DSP and Category = Likely Pathogenic

Variants (17)

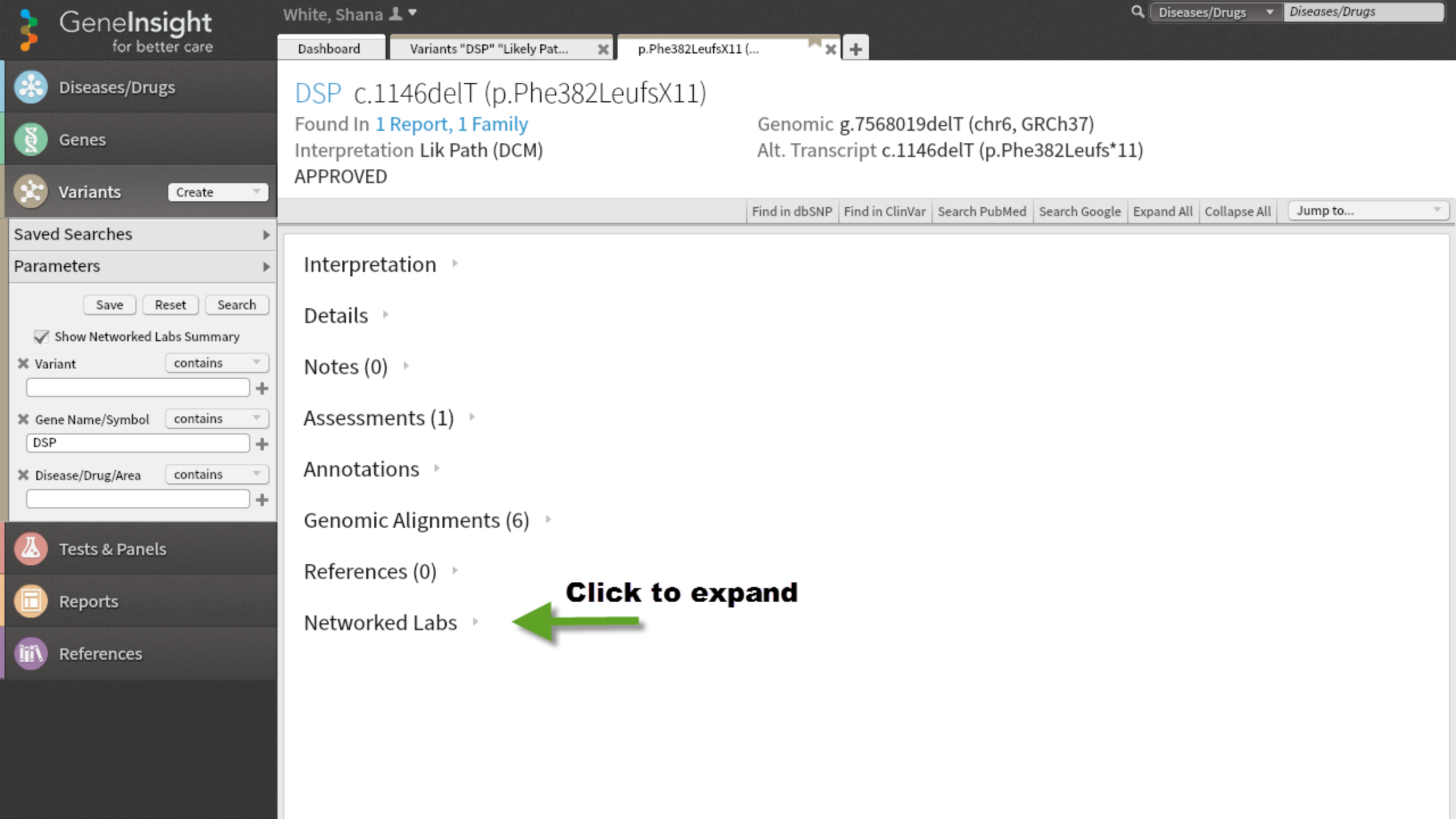
Click header to sort. Control+Click (Cmd+Click on Mac) header to remove sort. Delete | Approve Proposed Interp. | Download

Locus	DNA	AA	Genomic	**	Region	Cat (Dis)	Cat Date	Rpt	Fam	Actions	dbSNP	ClinVar	Networked Lab
DSP	c.214C>T	p.Gln72X	g.7555994C>...	R	Ex 02	Lik Path (...	16.May.2008	1	1	Ed | Del			
DSP	c.478C>T	p.Arg160X	g.7559514C>...		Ex 04	Lik Path (...	30.Sep.2011	2	2	Ed | Del			
DSP	c.534_535insA	p.Gly179Argf...	g.7559570_7...		Ex 04	Lik Path (DCM)	21.Sep.2012	1	1	Ed | Del			Under Review - 1 Unknown Signif...
DSP	c.699G>A	p.Trp233X	g.7562986G...		Ex 05	Lik Path (DCM)	02.Jul.2012	2	1	Ed | Del			
DSP	c.712_713insA	p.Ile238Asnf...	g.7562999_7...		Ex 05	Lik Path (DCM)	17.Jul.2012	1	1	Ed | Del			
DSP	c.867C>A	p.Cys289X	g.7565681C>...		Ex 07	Lik Path (DCM)	08.Dec.2012	1	1	Ed | Del			
DSP	c.939+1G>A		g.7565754G...		In 07	Lik Path (...	02.May.2012	2	1	Ed | Del			
DSP	c.1146delT	p.Phe382Le...	g.7568019de...		Ex 10	Lik Path (DCM)	10.Dec.2012	1	1	Ed | Del			Unknown Signif... Likely Pathogen...
DSP	c.1273C>T	p.Arg425X	g.7568676C>...		Ex 11	Lik Path (...	01.Mar.2008	2	1	Ed | Del			
DSP	c.1650G>A	p.Trp550X	g.7570745G...	R	Ex 13	Lik Path (...	09.Oct.2009	1	1	Ed | Del			
DSP	c.2848_2849...	p.Ile950Asnf...	g.7577246_7...		Ex 20	Lik Path (DCM)	25.May.2012	1	1	Ed | Del			
DSP	c.2959T>C	p.Ser987Pro	g.7578093T>...	N	Ex 21	Lik Path (...	17.Aug.2009	2	1	Ed | Del			
DSP	c.3160_3169...	p.Lys1054Se...	g.7579583_7...		Ex 23	Lik Path (...	16.Nov.2007	6	1	Ed | Del			
DSP	c.3474_3475...	p.Glu1159Ar...	g.7579897_7...	N	Ex 23	Lik Path (...	*22.Mar.2012*	13	2	Ed | Del			Deleterious (... Likely Pathogen...
DSP	c.3829C>T	p.Gln1277X	g.7580252C>...		Ex 23	Lik Path (...	23.Dec.2011	1	1	Ed | Del			
DSP	c.4531C>T	p.Gln1511X	g.7580954C>...	N	Ex 23	Lik Path (...	15.Dec.2009	1	1	Ed | Del			
DSP	c.5428C>T	p.Gln1810X	g.7582923C>...		Ex 24	Lik Path (...	08.Apr.2008	1	1	Ed | Del			

Hover over

Summary By Category
Unknown Significance (DCM) - 1
Likely Pathogenic (DCM) - 2

All Categories
Unknown Significance (DCM) - LAB-DEMO-D
Likely Pathogenic (DCM) - LAB-DEMO-C
Likely Pathogenic (DCM) - LAB-DEMO-B


GeneInsight
for better care
White, Shana
Diseases/Drugs
Diseases/Drugs
Dashboard
Variants "DSP" "Likely Pat...
p.Phe382LeufsX11 (...
Diseases/Drugs
Genes
Variants
Create
Saved Searches
Parameters
Save
Reset
Search
Show Networked Labs Summary
Variant
contains
Gene Name/Symbol
contains
DSP
Disease/Drug/Area
contains
Tests & Panels
Reports
References
DSP c.1146delT (p.Phe382LeufsX11)
Found In 1 Report, 1 Family
Interpretation Lik Path (DCM)
APPROVED
Genomic g.7568019delT (chr6, GRCh37)
Alt. Transcript c.1146delT (p.Phe382Leufs*11)
Find in dbSNP
Find in ClinVar
Search PubMed
Search Google
Expand All
Collapse All
Jump to...
Interpretation
Details
Notes (0)
Assessments (1)
Annotations
Genomic Alignments (6)
References (0)
Networked Labs
Click to expand

DSP c.1146delT (p.Phe382LeufsX11)

Found In 1 Report, 1 Family
Interpretation Lik Path (DCM)
APPROVED

Genomic g.7568019delT (chr6, GRCh37)
Alt. Transcript c.1146delT (p.Phe382Leufs*11)

Interpretation

Details

Notes (0)

Assessments (1)

Annotations

Genomic Alignments (6)

References (0)

Networked Labs

Warning: The Network Laboratories section below displays information for outside laboratories or content providers. GeneInsight has not validated this content. It is important that you review and validate this content before using it for any purpose. GeneInsight provides access to these external information sources for convenience only; the inclusion of a laboratory or content provider is not intended to be, and should not be taken as, an endorsement or validation by GeneInsight of any content, product or service that may be available. Terms of use applicable at third party websites are not controlled by GeneInsight and may be different. Any questions regarding the information provided should be directed to the networked laboratory or content provider.

Click to expand

Ext. Source	Transcript	DNA	AA	Region	DNA Type	AA Type	Interp	# Rpts	# Fams
LAB-DEMO-D	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Unknown Significance (DCM)	1	1
LAB-DEMO-C	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Likely Pathogenic (DCM)	1	1
LAB-DEMO-B	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Likely Pathogenic (DCM)	1	1

Parameters

Save Reset Search

Show Networked Labs Summary

Variant contains

Gene Name/Symbol contains

DSP

Disease/Drug/Area contains

Tests & Panels

Reports

References

Networked Labs

Warning: The Network Laboratories section below displays information for outside laboratories or content providers. GeneInsight has not validated this content. It is important that you review and validate this content before using it for any purpose. GeneInsight provides access to these external information sources for convenience only; the inclusion of a laboratory or content provider is not intended to be, and should not be taken as, an endorsement or validation by GeneInsight of any content, product or service that may be available. Terms of use applicable at third party websites are not controlled by GeneInsight and may be different. Any questions regarding the information provided should be directed to the networked laboratory or content provider.

LAB-DEMO-B Information Close

ALIASES

REPORT ALLELE NAME, DNA CHANGE, AND AMINO ACID CHANGE

p.Phe382fs

REPORTS
1

FAMILIES
1

SPLICING IMPACT
Predicted unlikely impact

SOURCE

Current Interpretation Import

CONTENT APPROVED 10.Dec.2012 06:05 PM by Matthew Lebo

REVISION APPROVED 19.Mar.2013 03:32 PM by Geneticist One

REASON(S) FOR UPDATE
New Evidence

CATEGORY
Likely Pathogenic

DISEASES/DRUGS
DCM

PHENOTYPE

EXCLUDE FROM REPORTS
No

INHERITANCE
Autosomal dominant

SCORE

EVIDENCE

The Phe382fs variant in DSP has not been reported in the literature nor previously identified by our laboratory. This frameshift variant is predicted to alter the protein's amino acid sequence beginning at position 382 and lead to a premature termination codon 11 amino acids downstream. This alteration is then predicted to lead to a truncated or absent protein. Frameshift and nonsense variants in DSP are common in patients with ARVC (http://arvcdatabase.info/), but recent evidence supports that they can also cause DCM (Elliott 2010, Garcia-Pavia 2011). In summary, this variant is likely to be pathogenic, though additional studies are required to fully establish its clinical significance.

Ext. Source	Transcript	DNA	AA	Region	DNA Type	AA Type	Interp	# Rpts	# Fa
LAB-DEMO-D	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Unknown Significance (DCM)	1	1
LAB-DEMO-C	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Likely Pathogenic (DCM)	1	1
LAB-DEMO-B	NM_004415.2	c.1146delT	p.Phe382LeufsX11	Ex 10	Del	FS	Likely Pathogenic (DCM)	1	1

Approving variant interpretations

- Variant interpretations must be approved for their interpretation to be visible to other labs

Single variant approval

Diseases/Drugs
Genes
Variants Create
Saved Searches
Parameters
Save Reset Search
Show Networked Labs Summary
Variant contains
Gene Name/Symbol contains
Category equals
Disease/Drug/Area contains
Genome Region equals
Interpretation Status equals
Proposed Revision
OR Proposed Initial Interp...
Tests & Panels
Reports

HRAS c.123G>A (p.Arg41Arg)

Found In 0 Reports, 0 Families
Interpretation Lik Ben
PENDING APPROVAL

Genomic g.533933C>T (chr11, GRCh37)

Find in dbSNP | Find in ClinVar | Search PubMed | Search Google | Expand All | Collapse All | Jump to...

Interpretation

Edit | Approve | Reject

Proposed

PROPOSED 20.Feb.2015 05:15 by Shana White

CATEGORY
Likely Benign

DISEASES/DRUGS

PHENOTYPE

EXCLUDE FROM REPORTS
No

INHERITANCE

SCORE

EVIDENCE
This variant has been identified in a high frequency of controls.

Click to approve

Change History

Show Text-Only Change History

Category	Date	Author	Reason(s) for Update
Likely Benign	20.Feb.2015 05:15 PM	Shana White	

Details

Notes (0)

Assessments (0)

Annotations

Bulk approval

Diseases/Drugs

Genes

Variants Create

Saved Searches

Parameters

Save | Reset | Search

Show Networked Labs Summary

Variant contains

Gene Name/Symbol contains

Category equals

Disease/Drug/Area contains

Genome Region equals

Interpretation Status equals

Proposed Revision

OR Proposed Initial Interp...

Tests & Panels

Reports

Showing results for: Interpretation Status = Proposed Revision or Proposed Initial Interpretation

Variants (796)

Click header to sort. Control+Click (Cmd+Click on Mac) header to remove sort. Delete | Approve Proposed Interp. | Copy Selected Rows | Download

Locus	DNA	AA	Genomic	*1	Region	Cat (Dis)	Cat Date	Rpt	Fam	Actions	dbSNP	ClinVar	Alias	Ref
	del(3)(p11)			R		Unk Sig		0	0	Ed | Del				
ABCC9	c.49G>A	p.Asp17Asn	g.22089560...	R	Ex 22	(Not Cat.) (...		0	0	Ed | Del				
ABCC9	c.169C>T	p.Gln57X	g.22086831...	N	Ex 02	Unk Sig, 5 (...	20.Jan.2011	4	1	Ed | Del				
ABCC9	c.816+11G>A		g.22068591...	N	In 05	Lik Ben	19.Mar.2012	1	1	Ed | Del				
ABCC9	c.852T>A	p.Ser284Ser	g.22065965...	RN	Ex 06	Lik Ben		0	0	Ed | Del	rs113562970			
ABCC9	c.1165-6T>G		g.22063252...	RN	In 07	Ben		0	0	Ed | Del	rs35857705			
ABCC9	c.2425-13G>A		g.22012613...	R	In 19	Unk Sig, 4		1	1	Ed | Del				
ABCC9	c.2826T>C	p.Tyr942Tyr	g.22001124...	RN	Ex 23	Lik Ben		0	0	Ed | Del				
ACTA2	c.1110C>T	p.Ser370Ser	g.90695004...	RN	Ex 09	Lik Ben		0	0	Ed | Del				
ACTA2	c.1118A>C	p.His373Pro	g.90694996...	RN	Ex 09	Ben		0	0	Ed | Del	rs1062398			
ACTC	c.42C>T	p.Asn14Asn	g.35086968...	R	Ex 01	Lik Ben		0	0	Ed | Del				
ACTC	c.76G>A	p.Asp26Asn	g.35086934...	RN	Ex 01	Unk Sig, 5	18.Jun.2010	4	1	Ed | Del				tng 2009
ACTC	c.383C>T	p.Thr128Ile	g.35085517...	N	Ex 02	Unk Sig, 5 (...	09.Mar.2009	4	1	Ed | Del				
ACTC	c.756T>G	p.Ile252Met	g.35084343...	N	Ex 04	Unk Sig, 5 (...	01.Mar.2008	2	1	Ed | Del				
ACTC	c.793C>G	p.Gln265Glu	g.35084306...		Ex 04	Unk Sig, 4	01.Mar.2008	2	2	Ed | Del				
ACTC	c.850A>T	p.Ile284Phe	g.35083455...	N	Ex 05	Unk Sig (...	30.Sep.2008	3	1	Ed | Del				
ACTG1	c.1128A>G	p.X376X		RXN	Ex 06	Ben		2	2	Ed | Del	rs11549223			
ACTN2	c.136G>T	p.Ala46Ser	g.23688116...	R	Ex 02	Unk Sig, 5		0	0	Ed | Del				
ACTN2	c.355G>A	p.Ala119Thr		DN	Ex 03	Path (HCM)	23.Dec.2009	0	0	Ed | Del				Chiu 2009
ACTN2	c.546T>C	p.Asp182Asp	g.23689098...	RN	Ex 06	Lik Ben		0	0	Ed | Del	rs34263845			
ACTN2	c.1298C>T	p.Ser433Leu	g.23690796...	X	Ex 12	Ben	30.Sep.2011	1	1	Ed | Del	rs143749154			
ACTN2	c.1485G>A	p.Thr495Thr	g.23691104...	R	Ex 13	Lik Ben (...		1	1	Ed | Del	rs201179281			
ACTN2	c.1740C>T	p.Asn580Asn	g.23691485...	RN	Ex 15	Lik Ben		0	0	Ed | Del	rs3738546			

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Diseases/Drugs

Genes

Variants Create

Saved Searches

Parameters

Save | Reset | Search

Show Networked Labs Summary

Variant contains

Gene Name/Symbol contains

Category equals

Disease/Drug/Area contains

Genome Region equals

Interpretation Status equals

Proposed Revision

OR Proposed Initial Interp...

Tests & Panels

Reports

Showing results for: Interpretation Status = Proposed Revision or Proposed Initial Interpretation

Variants (796)

Click header to sort. Control+Click (Cmd+Click on Mac) header to remove sort. Delete | Approve Proposed Interp. | Copy Selected Rows | Download

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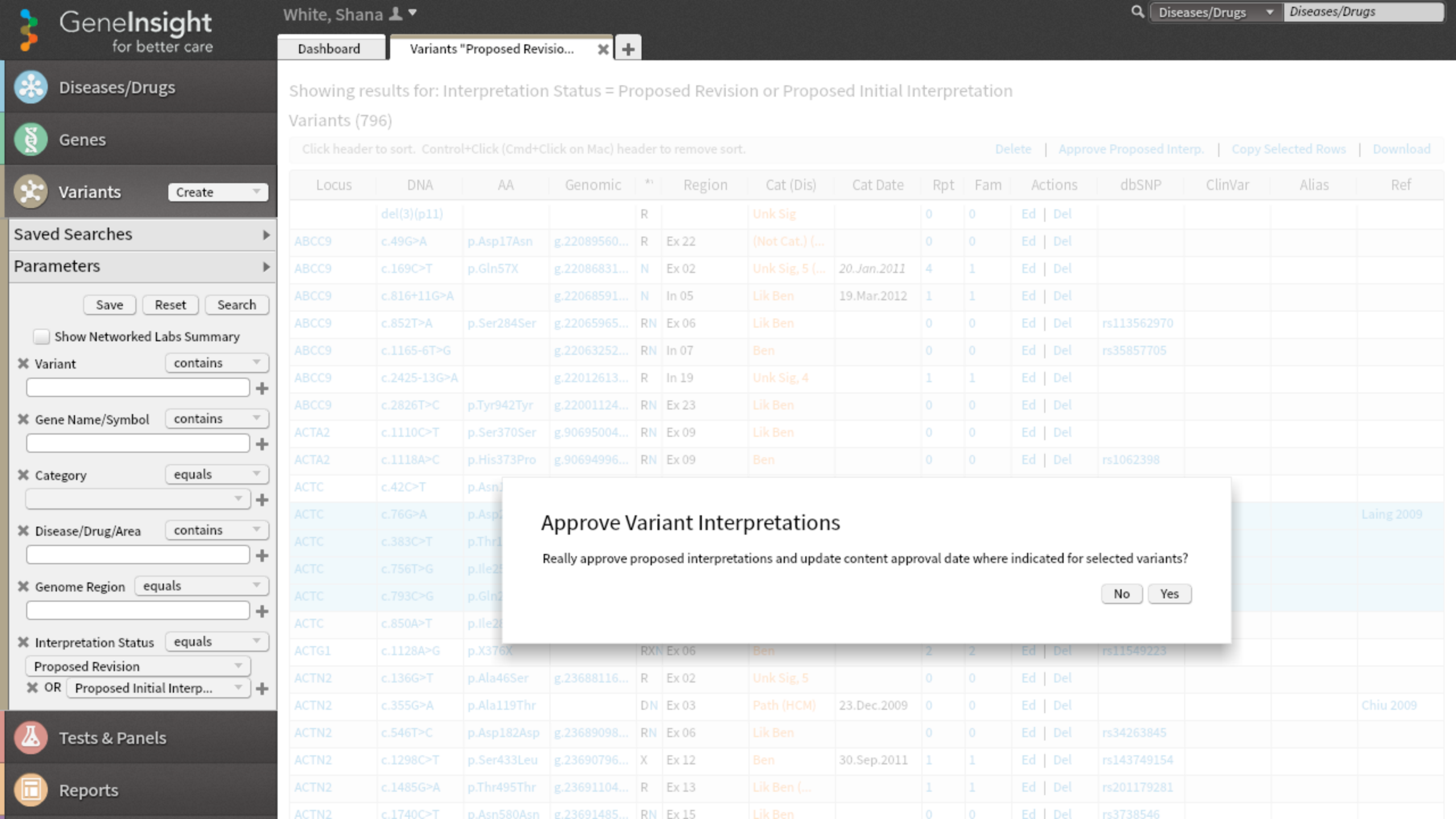
Locus	DNA	AA	Genomic	*1	Region	Cat (Dis)	Cat Date	Rpt	Fam	Actions	dbSNP	ClinVar	Alias	Ref
	del(3)(p11)			R		Unk Sig		0	0	Ed | Del				
ABCC9	c.49G>A	p.Asp17Asn	g.22089560...	R	Ex 22	(Not Cat.) (...		0	0	Ed | Del				
ABCC9	c.169C>T	p.Gln57X	g.22086831...	N	Ex 02	Unk Sig, 5 (...	20.Jan.2011	4	1	Ed | Del				
ABCC9	c.816+11G>A		g.22068591...	N	In 05	Lik Ben	19.Mar.2012	1	1	Ed | Del				
ABCC9	c.852T>A	p.Ser284Ser	g.22065965...	RN	Ex 06	Lik Ben		0	0	Ed | Del	rs113562970			
ABCC9	c.1165-6T>G		g.22063252...	RN	In 07	Ben		0	0	Ed | Del	rs35857705			
ABCC9	c.2425-13G>A		g.22012613...	R	In 19	Unk Sig, 4		1	1	Ed | Del				
ABCC9	c.2826T>C	p.Tyr942Tyr	g.22001124...	RN	Ex 23	Lik Ben		0	0	Ed | Del				
ACTA2	c.1110C>T	p.Ser370Ser	g.90695004...	RN	Ex 09	Lik Ben		0	0	Ed | Del				
ACTA2	c.1118A>C	p.His373Pro	g.90694996...	RN	Ex 09	Ben		0	0	Ed | Del	rs1062398			
ACTC	c.42C>T	p.Asn14Asn	g.35086968...	R	Ex 01	Lik Ben		0	0	Ed | Del				
ACTC	c.76G>A	p.Asp26Asn	g.35086934...	RN	Ex 01	Unk Sig, 5	18.Jun.2010	4	1	Ed | Del				Laing 2009
ACTC	c.383C>T	p.Thr128Ile	g.35085517...	N	Ex 02	Unk Sig, 5 (...	09.Mar.2009	4	1	Ed | Del				
ACTC	c.756T>G	p.Ile252Met	g.35084343...	N	Ex 04	Unk Sig, 5 (...	01.Mar.2008	2	1	Ed | Del				
ACTC	c.793C>G	p.Gln265Glu	g.35084306...		Ex 04	Unk Sig, 4	01.Mar.2008	2	2	Ed | Del				
ACTC	c.850A>T	p.Ile284Phe	g.35083455...	N	Ex 05	Unk Sig (...	30.Sep.2008	3	1	Ed | Del				
ACTG1	c.1128A>G	p.X376X		RXN	Ex 06	Ben		2	2	Ed | Del	rs11549223			
ACTN2	c.136G>T	p.Ala46Ser	g.23688116...	R	Ex 02	Unk Sig, 5		0	0	Ed | Del				
ACTN2	c.355G>A	p.Ala119Thr		DN	Ex 03	Path (HCM)	23.Dec.2009	0	0	Ed | Del				Chiu 2009
ACTN2	c.546T>C	p.Asp182Asp	g.23689098...	RN	Ex 06	Lik Ben		0	0	Ed | Del	rs34263845			
ACTN2	c.1298C>T	p.Ser433Leu	g.23690796...	X	Ex 12	Ben	30.Sep.2011	1	1	Ed | Del	rs143749154			
ACTN2	c.1485G>A	p.Thr495Thr	g.23691104...	R	Ex 13	Lik Ben (...		1	1	Ed | Del	rs201179281			
ACTN2	c.1740C>T	p.Asn580Asn	g.23691485...	RN	Ex 15	Lik Ben		0	0	Ed | Del	rs3738546			


GeneInsight
for better care
White, Shana
Diseases/Drugs
Diseases/Drugs
Dashboard
Variants "Proposed Revisio...
Diseases/Drugs
Genes
Variants
Create
Saved Searches
Parameters
Save
Reset
Search
Show Networked Labs Summary
Variant
contains
Gene Name/Symbol
contains
Category
equals
Disease/Drug/Area
contains
Genome Region
equals
Interpretation Status
equals
Proposed Revision
OR
Proposed Initial Interp...
Tests & Panels
Reports
Showing results for: Interpretation Status = Proposed Revision or Proposed Initial Interpretation
Variants (796)
Click header to sort. Control+Click (Cmd+Click on Mac) header to remove sort.
Delete | Approve Proposed Interp. | Copy Selected Rows | Download
Locus DNA AA Genomic *1 Region Cat (Dis) Cat Date Rpt Fam Actions dbSNP ClinVar Alias Ref
del(3)(p11) R Unk Sig 0 0 Ed | Del
ABCC9 c.49G>A p.Asp17Asn g.22089560... R Ex 22 (Not Cat.) (... 0 0 Ed | Del
ABCC9 c.169C>T p.Gln57X g.22086831... N Ex 02 Unk Sig, 5 (... 20.Jan.2011 4 1 Ed | Del
ABCC9 c.816+11G>A g.22068591... N In 05 Lik Ben 19.Mar.2012 1 1 Ed | Del
ABCC9 c.852T>A p.Ser284Ser g.22065965... RN Ex 06 Lik Ben 0 0 Ed | Del rs113562970
ABCC9 c.1165-6T>G g.22063252... RN In 07 Ben 0 0 Ed | Del rs35857705
ABCC9 c.2425-13G>A g.22012613... R In 19 Unk Sig, 4 1 1 Ed | Del
ABCC9 c.2826T>C p.Tyr942Tyr g.22001124... RN Ex 23 Lik Ben 0 0 Ed | Del
ACTA2 c.1110C>T p.Ser370Ser g.90695004... RN Ex 09 Lik Ben 0 0 Ed | Del
ACTA2 c.1118A>C p.His373Pro g.90694996... RN Ex 09 Ben 0 0 Ed | Del rs1062398
ACTC c.42C>T p.Asn
ACTC c.76G>A p.Asp Laing 2009
ACTC c.383C>T p.Thr1
ACTC c.756T>G p.Ile2
ACTC c.793C>G p.Gln2
ACTC c.850A>T p.Ile2
ACTG1 c.1128A>G p.X376X RXN Ex 06 Ben 2 2 Ed | Del rs11549223
ACTN2 c.136G>T p.Ala46Ser g.23688116... R Ex 02 Unk Sig, 5 0 0 Ed | Del
ACTN2 c.355G>A p.Ala119Thr DN Ex 03 Path (HCM) 23.Dec.2009 0 0 Ed | Del Chiu 2009
ACTN2 c.546T>C p.Asp182Asp g.23689098... RN Ex 06 Lik Ben 0 0 Ed | Del rs34263845
ACTN2 c.1298C>T p.Ser433Leu g.23690796... X Ex 12 Ben 30.Sep.2011 1 1 Ed | Del rs143749154
ACTN2 c.1485G>A p.Thr495Thr g.23691104... R Ex 13 Lik Ben (... 1 1 Ed | Del rs201179281
ACTN2 c.1740C>T p.Asn580Asn g.23691485... RN Ex 15 Lik Ben 0 0 Ed | Del rs3738546
Approve Variant Interpretations
Really approve proposed interpretations and update content approval date where indicated for selected variants?
No
Yes