Versioning Wireframes

Last updated: October 11th, 2016
Please comment on the wireframes themselves or email Mary Goldman: mary@soe.ucsc.edu

Versioning 1.0:

Viewing different releases

Variants

More **▼** Community

v Help

About BRCA1, BRCA2 and Cancer History of the BRCA Exchange

About this site

Previous Data releases

Webservices for PI data access

Use the search box above to find BRCA1 or BRCA2 variants. For more information about the BRCA genes and cancer, click *More* at the top of the page

search for "c.1105G>A", "brca1" or "IVS7+103

This website is supported by the BRCA Exchange of the Global Alliance for Genomics and Health. The BRCA Exchange advances our understanding of the genetic basis of breast cancer, ovarian cancer and other diseases by pooling data on BRCA1/2 genetic variants and corresponding clinical data from around the world.



Notes	Date	Data Sources	New Variants	New Classifications	Changed Classifications	Changed/updated Variants	Deleted Variants
Version 1	October 6, 2016	ENIGMA, ClinVar, ExLOVD, BIC	120	1	1	1400	2
Version 2	July 1, 2016	ENIGMA, ClinVar, ExLOVD, BIC	120	1	1	1400	2

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Version 2	July 1, 2016	ENIGMA, ClinVar, ExLOVD, BIC	120	1	1	1400	2

Notes:

- Most recent variant listed first
- New/Changed Classifications split into New classifications and Changed Classifications
- Removed date data sources were accessed since this is mostly useful for data provenance. Instead include date in zip file of all other provenance data

Variants

scelerisque blandit felis.

If it is the latest release, note it as the 'Current Release Notes'

Current Release notes

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120 new variants
1 new classification
1400 changed/updated variants
2 deleted variants



Release notes

Note that this is not the most current release. Click here to see the most current release

Previous releases have a note saying that it is not the most current release

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Nunc orci sapien, luctus sed bibendum sed, ornare vitae sem. Etiam scelerisque blandit felis.

> 120 new variants 1 new classification 1 changed classification 1400 changed/updated variants 2 deleted variants





Release notes

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General text describing changes made

Link to the variant table where each of the variants falling into these categories are the only variants in the 'search' results

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120 new variants 1 new classification 1 changed classification 1400 changed/updated variants 2 deleted variants

Expert Reviewed

Notes:

- Instead of having separate links to all the provenance data, make it a single link.
- Removed md5sum of zip file of provenance data since what people care more about is the md5sum of the individual files, not the zip file (especially as unzipping the file can introduce errors). Instead include a md5sum.txt in the zip file itself. Zach will do this.

Versioning 2.0: Indicating that an individual variant has previous versions

Mocked up on an "Expert Reviewed" view of a variant but will be the same on the "All Data" view of a variant as well

Variant Detail

HOME

Gene	BRCA1
HGVS Nucleotide	c.199G>T
Transcript Identifier	NM_007294.3
HGVS RNA	
HGVS Protein	p.(Asp67Tyr)
Protein Identifier	NP_009225.1

Scroll to more information

Date Last Evaluated	10 August 2015	
Assertion Method	ENIGMA BRCA1/2 Class	ification Criteria (2015)
Assertion Method Citation	Enigma Rules version Ma	ar 26, 201 <u>5</u>
Allele Origin	Germline	
ClinVar Accession	SCV000244311	All changes will be listed in the changes column When a variant changes

Previous Versions of this Variant:

NM_007294.3:c.199G>T

Date	Clinical Significance	Changes	3 to 2/1 light 3 to 4/5 darker 5/4 to 3 darkest	
10 August 2015	Benign/ Little Clinical Significance	Clinical Significance		
		Allele Frequency (1000 Genomes): (none) -> 0.0002		

classification, this change

will be highlighted in red

Classifications:

How to view deleted variants

This is the BRCA Exchange Clinical Space, and contains only expert-reviewed variant classifications. The variant classifications shown here represent the initial set submitted by the ENIGMA consortium, ~1000 (primarily missense) variants in BRCA1 and BRCA2. The Clinical space will become more fully populated pending additional expert review.

Show Filters

13500 matching variants

c.571

Gene	HGVS Nucleotide	HGVS Protein	Protein Abbrev	BIC Designation	Clinical Significance
BRCA1	c.571G>A	p.(Val191lle)	V191I	690G>A	Pathogenic

There is 1 deleted variant that match your search. Click here to view these deleted variants.

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BRCA1	c.571G>A	p.(Val191lle)	V191I	690G>A	Pathogenic

There is 1 deleted variant that match your search. Click

Text will dynamically change "is" to "are" if there is more than one deleted variant that matches

Expert Reviewed

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13500 matching variants

c.571

Deleted variant is shown in table with non-deleted variant. Deleted variant is highlighted in orange

Gene	HGVS Nucleotide	HGVS Protein	Protein Abbrev	BIC Designation	Clinical Significance
BRCA1	c.571G>A	p.(Val191lle)	V191I	690G>A	Pathogenic
BRCA1	c.571G>C	p.(Val191Leu)	V191L	690G>C	Not Yet Classified

Variant Detail

Note this variant has been removed from the BRCA Exchange

Gene	BRCA1
HGVS Nucleotide	c.571G>C
Transcript Identifier	NM_007294.3
HGVS RNA	
HGVS Protein	p.(Val191Lue)
Protein Identifier	NP_009225.1