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| --- | --- | --- | --- | --- | --- |
| **SAMPLE** | **VARIANT ID** | **TYPE OF VARIANT** | **DBSNP** | | |
| **CONDITIONS** | **CLINICAL SIGNIFICANCE**  **(LAST REVIEWED)** | **REVIEW STATUS** |
| 01\_S1\_L001\_  indels\_dbSnp\_ann.vcf | rs11571814 | indels | Hereditary cancer-predisposing syndrome, not provided, Hereditary breast ovarian cancer syndrome, Breast-ovarian cancer, familial, susceptibility to, 2 | Benign/Likely benign (Dec 17, 2021) | criteria provided, multiple submitters, no conflicts |
| Hereditary cancer-predisposing syndrome, not provided, not specified, Hereditary breast ovarian cancer syndrome | Conflicting interpretations of pathogenicity (Nov 7, 2021) | criteria provided, conflicting interpretations |
| rs11571814 | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015 | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| 01\_S1\_L001snps\_dbSnp\_ann.vcf | rs1799943 | snps | Hereditary breast ovarian cancer syndrome | Uncertain significance (Jun 4, 2021) | criteria provided, single submitter |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jun 12, 2000) | no assertion criteria provided |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| rs9534174 | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Nov 19, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
| rs1801406 | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Dec 17, 2010) | no assertion criteria provided |
| rs543304 | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| rs206075 | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | criteria provided, multiple submitters, no conflicts |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Nov 4, 2011) | no assertion criteria provided |
| rs206076 | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | not provided | no assertion provided |
| rs1460816 | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| rs1799955 | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| 02\_S2\_L001\_indels\_dbSnp\_ann.vcf | rs11571814 | indels | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015))  Method: curation |
| 02\_S2\_L001snps\_dbSnp\_ann.vcf | rs144848 | snps | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Jan 7, 2022) | criteria provided, multiple submitters, no conflicts |
| Breast-ovarian cancer, familial, susceptibility to, 2 | not provided | no assertion provided |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| rs206075 | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | criteria provided, multiple submitters, no conflic |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Nov 4, 2011) | no assertion criteria provided |
| rs206076 | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | not provided | no assertion provided |
| rs169547 | not specified, not provided, Breast-ovarian cancer, familial, susceptibility to, 2, Hereditary breast ovarian cancer syndrome | Benign (Apr 19, 2022) | criteria provided, multiple submitters, no conflicts |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| 03\_S3\_L001\_indels\_dbSnp\_ann.vcf | rs11571661 | indels | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015 | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015))  Method: curation |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: inherited | Benign  (Dec 03, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Mar 10, 2015) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Benign  (Oct 09, 2014) | criteria provided, single submitter  ([ACGS Guidelines, 2013](https://submit.ncbi.nlm.nih.gov/ft/byid/zo8lkdib/evaluation_and_reporting_of_sequence_variants_bpgs_june_2013_finalpdf.pdf))  Method: clinical testing |
| [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline  [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Uncertain significance  (Dec 17, 2010) | no assertion criteria provided  Method: clinical testing |
| no assertion criteria provided  Method: clinical testing | Benign  (-) | [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline |
| no assertion provided  Method: not provided | untested  (-) | [Familial cancer of breast](https://www.ncbi.nlm.nih.gov/medgen/C0346153/)  Affected status: not provided  Allele origin: somatic |
| rs11571814 | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| 03\_S3\_L001snps\_dbSnp\_ann.vcf | rs1799943 | snps | Hereditary breast ovarian cancer syndrome | Uncertain significance (Jun 4, 2021) | criteria provided, single submitted |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jun 12, 2000) | no assertion criteria provide |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| rs7872606 | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jun 21, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| [TSC](https://www.ncbi.nlm.nih.gov/medgen/C0041341/)  Affected status: yes  Allele origin: germline | not provided  (-) | no assertion provided  ([Tuberous Sclerosis Database Assertion Criteria 2015](https://submit.ncbi.nlm.nih.gov/ft/byid/krkkw3ff/tuberous_sclerosis_database_assertion_criteria_2015.docx))  Method: curation |
| 04\_S4\_L001\_indels\_dbSnp\_ann.vcf | rs11571661 | indels | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: inherited | Benign  (Dec 03, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Mar 10, 2015) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Benign  (Oct 09, 2014) | criteria provided, single submitter  ([ACGS Guidelines, 2013](https://submit.ncbi.nlm.nih.gov/ft/byid/zo8lkdib/evaluation_and_reporting_of_sequence_variants_bpgs_june_2013_finalpdf.pdf))  Method: clinical testing |
| [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Uncertain significance  (Dec 17, 2010) | no assertion criteria provided  Method: clinical testing |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
| [Familial cancer of breast](https://www.ncbi.nlm.nih.gov/medgen/C0346153/)  Affected status: not provided  Allele origin: somatic | untested  (-) | no assertion provided  Method: not provided |
| [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: inherited | Benign  (Dec 03, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Mar 10, 2015) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Benign  (Oct 09, 2014) | criteria provided, single submitter  ([ACGS Guidelines, 2013](https://submit.ncbi.nlm.nih.gov/ft/byid/zo8lkdib/evaluation_and_reporting_of_sequence_variants_bpgs_june_2013_finalpdf.pdf))  Method: clinical testing |
| [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| 04\_S4\_L001snps\_dbSnp\_ann.vcf | rs9526165 | snps | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| rs1801426 | Lynch syndrome | Benign (Sep 5, 2013) | reviewed by expert panel |
| Breast-ovarian cancer, familial, susceptibility to, 2 | Benign | Breast-ovarian cancer, familial, susceptibility to, 2 |
| 05\_S5\_L001\_indels\_dbSnp\_ann.vcf | rs11571661 | indels | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: inherited | Benign  (Dec 03, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Mar 10, 2015) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
| [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Benign  (Oct 09, 2014) | criteria provided, single submitter  ([ACGS Guidelines, 2013](https://submit.ncbi.nlm.nih.gov/ft/byid/zo8lkdib/evaluation_and_reporting_of_sequence_variants_bpgs_june_2013_finalpdf.pdf))  Method: clinical testing |
| [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
| [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: yes  Allele origin: germline | Uncertain significance  (Dec 17, 2010) | no assertion criteria provided  Method: clinical testing |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
| [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
| [Familial cancer of breast](https://www.ncbi.nlm.nih.gov/medgen/C0346153/)  Affected status: not provided  Allele origin: somatic | untested  (-) | no assertion provided  Method: not provided |
| 05\_S5\_L001snps\_dbSnp\_ann.vcf | rs1799943 | snps | Hereditary breast ovarian cancer syndrome | Uncertain significance (Jun 4, 2021) | criteria provided, single submitter |
|  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jun 12, 2000) | no assertion criteria provided |
|  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs9534174 |  | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  |  |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  |  |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Nov 19, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
| 10\_S3\_L001\_indels\_dbSnp\_ann.vcf | rs11571814 | indels | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs273902789 |  | not specified, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Benign/Likely benign (Dec 13, 2021) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  |  |  | Hereditary cancer-predisposing syndrome, not specified, Hereditary breast ovarian cancer syndrome | Benign/Likely benign (Oct 24, 2021) | criteria provided, multiple submitters, no conflicts |
| 06\_S6\_L001\_indels\_dbSnp\_ann | rs273902789 | indels | not specified, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Benign/Likely benign (Dec 13, 2021) | criteria provided, multiple submitters, no conflicts |
| Hereditary cancer-predisposing syndrome, not specified, Hereditary breast ovarian cancer syndrome | Benign/Likely benign (Oct 24, 2021) | criteria provided, multiple submitters, no conflicts |
| rs11571661 | [Familial cancer of breast](https://www.ncbi.nlm.nih.gov/medgen/C0346153/)  Affected status: not provided  Allele origin: somatic | untested | no assertion provided |
|  | [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/) | Benign | no assertion criteria provided |
| 06\_S6\_L001snps\_dbSnp\_ann | rs1166699 | snps | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jun 18, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification (06012015)](https://submit.ncbi.nlm.nih.gov/ft/byid/dhtz9flo/genedx_interprules_final_061215.pdf))  Method: clinical testing |
|  | rs1799943 |  | Hereditary breast ovarian cancer syndrome | Uncertain significance | criteria provided, single submitter |
|  | rs9534174 |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Nov 19, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  |  |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  | rs1801406 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs206075 |  | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs206076 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs1460816 |  | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  | rs1799955 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs169547 |  | not specified, not provided, Breast-ovarian cancer, familial, susceptibility to, 2, Hereditary breast ovarian cancer syndrome | Benign (Apr 19, 2022) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs9534262 |  | [not provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: unknown  Allele origin: germline | Benign  (Feb 24, 2022) | criteria provided, single submitter  ([arup molecular germline variant investigation process 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/ythvkvge/arup_molecular_germline_variant_investigation_process_2021.pdf))  Method: clinical testing |
|  |  |  | [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
|  | rs9526165 |  | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  | rs11571836 |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 14, 2020) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  |  |  | [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2018) | criteria provided, single submitter  ([ICSL Variant Classification Criteria 13 December 2019](https://submit.ncbi.nlm.nih.gov/ft/byid/r0x0xrmc/icsl_variant_classification_criteria_13_december_2019.pdf))  Method: clinical testing |
|  | rs1799966 |  | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome, not specified | Benign/Likely benign (Dec 8, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs273900734 |  | [not specified](https://www.ncbi.nlm.nih.gov/medgen/CN169374/)  Affected status: yes  Allele origin: germline | Benign  (-) | no assertion criteria provided  Method: clinical testing |
|  | rs3737559 |  | [Breast-ovarian cancer, familial 1](https://www.ncbi.nlm.nih.gov/medgen/C2676676/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  |  |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jun 23, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  | rs1060915 |  | not provided, Hereditary breast ovarian cancer syndrome | Benign (Jun 28, 2020) | criteria provided, single submitter |
|  |  |  | not provided, Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Likely benign (Feb 15, 2019) | criteria provided, multiple submitters, no conflicts |
|  | rs16942 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Oct 15, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs16941 |  | not specified, not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome, Breast-ovarian cancer, familial, susceptibility to, 1 | Uncertain significance (Aug 30, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs799917 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome, not specified, not provided | Conflicting interpretations of pathogenicity (Dec 23, 2021) | criteria provided, conflicting interpretations |
|  |  |  | Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Uncertain significance (Sep 17, 2021) | riteria provided, multiple submitters, no conflicts |
|  | rs16940 |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Feb 15, 2020) | criteria provided, single submitter  ([Sema4 Curation Guidelines](https://submit.ncbi.nlm.nih.gov/ft/byid/cy0kpury/sema4_variant_curation_guidelines-clinvar_april_2022.docx))  Method: curation |
|  |  |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: yes  Allele origin: germline | Benign  (Nov 01, 2021) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: research |
|  | rs1799949 |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2013) | criteria provided, single submitter  ([LabCorp Variant Classification Summary - May 2015](https://submit.ncbi.nlm.nih.gov/ft/byid/rtxspsnt/labcorp_variant_classification_method_-_may_2015.pdf))  Method: clinical testing |
|  |  |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Aug 01, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  | rs1157851269 |  | [RYR1-Related Disorders](https://www.ncbi.nlm.nih.gov/medgen/CN239331/)  Affected status: unknown  Allele origin: germline | Uncertain significance  (Sep 01, 2021) | criteria provided, single submitter  ([Invitae Variant Classification Sherloc (09022015)](https://www.ncbi.nlm.nih.gov/pubmed/28492532))  Method: clinical testing |
| 07\_S7\_L001\_indels\_dbSnp\_ann |  | indels |  |  |  |
| 07\_S7\_L001snps\_dbSnp\_ann | rs9534174 | snps | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  |  |  | |  |  | | --- | --- | | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | [Ambry Genetics](https://www.ncbi.nlm.nih.gov/clinvar/submitters/61756/)  Accession: SCV002608479.1 First in ClinVar: Nov 29, 2022 | | Benign  (Nov 19, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  | rs144848 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Jan 7, 2022) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
| 08\_S8\_L001\_indels\_dbSnp\_ann | rs276174816 | indels |  |  |  |
|  | rs11571661 |  |  |  |  |
| 08\_S8\_L001snps\_dbSnp\_ann | rs1799943 | snps | Hereditary breast ovarian cancer syndrome | Uncertain significance (Jun 4, 2021) | criteria provided, single submitter |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs9534174 |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Nov 19, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  |  |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  | rs144848 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Jan 7, 2022) | criteria provided, multiple submitters, no conflicts |
|  | rs1801406 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs1214830867 |  | Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Jul 10, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Hereditary breast ovarian cancer syndrome | Likely benign (May 12, 2019) | criteria provided, single submitter |
|  | rs206075 |  | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs206076 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  | rs1460816 |  | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  | rs1799955 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  | rs169547 |  | not specified, not provided, Breast-ovarian cancer, familial, susceptibility to, 2, Hereditary breast ovarian cancer syndrome | Benign (Apr 19, 2022) | criteria provided, multiple submitters, no conflicts |
|  | rs9534262 |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/) | Benign  (Dec 18, 2021) | criteria provided, single submitter  ([Invitae Variant Classification Sherloc (09022015)](https://www.ncbi.nlm.nih.gov/pubmed/28492532))  Method: clinical testing |
|  |  |  | [Breast and/or ovarian cancer](https://www.ncbi.nlm.nih.gov/medgen/CN221562/)  Affected status: unknown  Allele origin: germline | Benign  (Apr 22, 2021) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
|  | rs80359065 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  | rs56268579 |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: yes  Allele origin: germline | Benign  (Nov 16, 2021) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
|  |  |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: unknown  Allele origin: germline | Benign  (Aug 05, 2020) | criteria provided, single submitter  ([Invitae Variant Classification Sherloc (09022015)](https://www.ncbi.nlm.nih.gov/pubmed/28492532))  Method: clinical testing |
|  | rs9526165 |  | [Breast-ovarian cancer, familial 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  ([ENIGMA BRCA1/2 Classification Criteria (2015)](https://submit.ncbi.nlm.nih.gov/ft/byid/c2ffjci6/enigma_rules_2015-03-26.pdf))  Method: curation |
|  | rs11571836 |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Jul 14, 2020) | criteria provided, single submitter  ([GeneDx Variant Classification Process June 2021](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  |  |  | [Breast-ovarian cancer, familial, susceptibility to, 2](https://www.ncbi.nlm.nih.gov/medgen/C2675520/)  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2018) | criteria provided, single submitter  ([ICSL Variant Classification Criteria 13 December 2019](https://submit.ncbi.nlm.nih.gov/ft/byid/r0x0xrmc/icsl_variant_classification_criteria_13_december_2019.pdf))  Method: clinical testing |
|  | rs4986854 |  | Hereditary cancer-predisposing syndrome | Uncertain significance (Feb 4, 2019) | criteria provided, single submitter |
|  | rs1799966 |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Aug 10, 2015) | reviewed by expert pane |
|  | rs273900734 |  | [Not Provided](https://www.ncbi.nlm.nih.gov/medgen/CN517202/)  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  ([GeneDX Variant Classification (06012015)](https://submit.ncbi.nlm.nih.gov/ft/byid/fc81e176/genedx_variant_classification_process_june_2021.pdf))  Method: clinical testing |
|  | rs1060915 |  | not provided, Hereditary breast ovarian cancer syndrome | Benign (Jun 28, 2020) | criteria provided, single submitter |
|  |  |  | not provided, Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Likely benign (Feb 15, 2019) | criteria provided, multiple submitters, no conflicts |
|  | rs16942 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Oct 15, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Aug 10, 2015) | reviewed by expert panel |
|  | rs16941 |  | not specified, not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome, Breast-ovarian cancer, familial, susceptibility to, 1 | Uncertain significance (Aug 30, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs799917 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome, not specified, not provided | Conflicting interpretations of pathogenicity (Dec 23, 2021) | criteria provided, conflicting interpretations |
|  |  |  | Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Uncertain significance (Sep 17, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs16940 |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2021) | criteria provided, single submitter  ([Invitae Variant Classification Sherloc (09022015)](https://www.ncbi.nlm.nih.gov/pubmed/28492532))  Method: clinical testing |
|  |  |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Aug 01, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  | rs201875054 |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Likely benign  (Apr 17, 2017) | criteria provided, single submitter  ([ACMG Guidelines, 2015](https://www.ncbi.nlm.nih.gov/pubmed/25741868))  Method: clinical testing |
|  |  |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Likely benign  (Jun 25, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  | rs1799949 |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2021) | criteria provided, single submitter  ([Invitae Variant Classification Sherloc (09022015)](https://www.ncbi.nlm.nih.gov/pubmed/28492532))  Method: clinical testing |
|  |  |  | [Hereditary cancer-predisposing syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0027672/)  Affected status: unknown  Allele origin: germline | Benign  (Aug 01, 2014) | criteria provided, single submitter  ([Ambry General Variant Classification Scheme\_2022](https://submit.ncbi.nlm.nih.gov/ft/byid/5hsvndk6/ambry_general_variant_classification_scheme_2022.pdf))  Method: clinical testing |
|  |  |  | [Hereditary breast ovarian cancer syndrome](https://www.ncbi.nlm.nih.gov/medgen/C0677776/)  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2013) | criteria provided, single submitter  ([LabCorp Variant Classification Summary - May 2015](https://submit.ncbi.nlm.nih.gov/ft/byid/rtxspsnt/labcorp_variant_classification_method_-_may_2015.pdf))  Method: clinical testing |
| 10\_S3\_L001snps\_dbSnp\_ann.vcf | rs9534174 | SNP | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | Not Provided  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  (GeneDx Variant Classification Process June 2021  Method: clinical testing |
|  | rs543304 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs206075 |  | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | |  | | --- | | reviewed by expert panel | |  | |
|  | rs206076 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | |  | | --- | | reviewed by expert panel | |  | |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | |  | | --- | | reviewed by expert panel | |  | |
|  | rs1460817 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs1460816 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs169547 |  | not specified, not provided, Breast-ovarian cancer, familial, susceptibility to, 2, Hereditary breast ovarian cancer syndrome | Benign (Apr 19, 2022) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs9534262 |  | Hereditary breast ovarian cancer syndrome  Affected status: yes  Allele origin: germline | Benign  (Apr 19, 2022) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
|  |  |  | Breast and/or ovarian cancer  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2021) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
|  | rs9526165 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs1799966 |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Aug 10, 2015) | |  | | --- | | reviewed by expert panel | |  | |
|  |  |  | not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome, not specified | Benign/Likely benign (Dec 8, 2021) | criteria provided, multiple submitters, no conflicts |
|  | rs273900734 |  | Breast-ovarian cancer, familial 1  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | not specified  Affected status: yes  Allele origin: germline | Benign  (Nov 01, 2017) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
|  | rs1060915 |  | not provided, Hereditary breast ovarian cancer syndrome | Benign (Jun 28, 2020) | |  | | --- | | criteria provided, single submitter | |  | |
|  |  |  | not provided, Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Likely benign (Feb 15, 2019) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  | rs16942 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome | Uncertain significance (Oct 15, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Aug 10, 2015) | reviewed by expert panel |
|  | rs16941 |  | not specified, not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome, Breast-ovarian cancer, familial, susceptibility to, 1 | Uncertain significance (Aug 30, 2021) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Aug 10, 2015) | reviewed by expert panel |
|  | rs799917 |  | Hereditary cancer-predisposing syndrome, Hereditary breast ovarian cancer syndrome, not specified, not provided | Conflicting interpretations of pathogenicity (Dec 23, 2021) | |  | | --- | | criteria provided, conflicting interpretations | |  | |
|  |  |  | Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Uncertain significance (Sep 17, 2021) | |  | | --- | | criteria provided, multiple submitters, no conflicts | |  | |
|  | rs16940 |  | not provided  Affected status: unknown  Allele origin: germline | Benign  (Feb 14, 2022) | criteria provided, single submitter  (arup molecular germline variant investigation process 2021  Method: clinical testing |
|  |  |  | Hereditary breast ovarian cancer syndrome  Affected status: unknown  Allele origin: germline | Benign  (Dec 18, 2021) | criteria provided, single submitter  (Invitae Variant Classification Sherloc (09022015)  Method: clinical testing |
|  | rs1799949 |  | Breast-ovarian cancer, familial 1  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | Hereditary cancer-predisposing syndrome  Affected status: unknown  Allele origin: germline | Benign  (Nov 21, 2014) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
|  | rs1800062 |  | Breast-ovarian cancer, familial, susceptibility to, 1 | not provided | |  | | --- | | no assertion provided | |  | |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 1 | Benign (Jan 12, 2015) | reviewed by expert panel |
| 11\_S4\_L001\_indels\_dbSnp\_ann.vcf | rs276174878 | indels | Hereditary cancer-predisposing syndrome, not provided, Hereditary breast ovarian cancer syndrome, Breast-ovarian cancer, familial, susceptibility to, 2 | Benign/Likely benign (Dec 17, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Hereditary cancer-predisposing syndrome, not provided, not specified, Hereditary breast ovarian cancer syndrome | Conflicting interpretations of pathogenicity (Nov 7, 2021) | criteria provided, conflicting interpretations |
|  | rs11571661 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | not specified  Affected status: unknown  Allele origin: germline | Uncertain significance  (Apr 20, 2017) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
|  | rs276174878 |  | Hereditary cancer-predisposing syndrome, not provided, Hereditary breast ovarian cancer syndrome, Breast-ovarian cancer, familial, susceptibility to, 2 | Benign/Likely benign (Dec 17, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Hereditary cancer-predisposing syndrome, not provided, not specified, Hereditary breast ovarian cancer syndrome | Conflicting interpretations of pathogenicity (Nov 7, 2021) | criteria provided, conflicting interpretations |
|  | rs1799943 | snps | Hereditary breast ovarian cancer syndrome | Uncertain significance (Jun 4, 2021) | criteria provided, single submitter |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jun 12, 2000) | no assertion criteria provided |
|  | rs9534174 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | Not Provided  Affected status: yes  Allele origin: germline | Benign  (Jul 09, 2018) | criteria provided, single submitter  (GeneDx Variant Classification Process June 2021  Method: clinical testing |
|  | rs1801406 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Dec 17, 2010) | no assertion criteria provided |
|  | rs206075 |  | Not provided, Hereditary breast ovarian cancer syndrome, Hereditary cancer-predisposing syndrome | Likely benign (Sep 13, 2021) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  | rs206076 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs1460816 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs1799955 |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Likely benign (Jun 29, 2017) | reviewed by expert panel |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs169547 |  | not specified, not provided, Breast-ovarian cancer, familial, susceptibility to, 2, Hereditary breast ovarian cancer syndrome | Benign (Apr 19, 2022) | criteria provided, multiple submitters, no conflicts |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2 | Benign (Jan 12, 2015) | reviewed by expert panel |
|  | rs9534262 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  |  |  | Breast-ovarian cancer, familial 2  (Autosomal dominant inheritance)  Affected status: unknown  Allele origin: unknown | Benign  (Jan 02, 2014) | criteria provided, single submitter  (Counsyl Autosomal Dominant Disease Classification criteria (2015)  Method: literature only |
|  | rs9526165 |  | Breast-ovarian cancer, familial 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2015) | reviewed by expert panel  (ENIGMA BRCA1/2 Classification Criteria (2015)  Method: curation |
|  | rs11571836 |  | Not Provided  Affected status: yes  Allele origin: germline | Benign  (Jul 14, 2020) | criteria provided, single submitter  (GeneDx Variant Classification Process June 2021  Method: clinical testing |
|  |  |  | Breast-ovarian cancer, familial, susceptibility to, 2  Affected status: unknown  Allele origin: germline | Benign  (Jan 12, 2018) | criteria provided, single submitter  (ICSL Variant Classification Criteria 13 December 2019  Method: clinical testing |
|  | rs3826551 |  | Not Provided  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  (GeneDX Variant Classification (06012015)  Method: clinical testing |
|  |  |  | not provided  Affected status: unknown  Allele origin: germline | Benign  (Dec 10, 2021) | criteria provided, single submitter  (Invitae Variant Classification Sherloc (09022015)  Method: clinical testing |
|  | rs11551758 |  | not provided  Affected status: unknown  Allele origin: germline | Benign  (Jun 26, 2017) | criteria provided, single submitter  (Invitae Variant Classification Sherloc (09022015)  Method: clinical testing |
|  |  |  | Not Provided  Affected status: yes  Allele origin: germline | Benign  (Mar 03, 2015) | criteria provided, single submitter  (GeneDx Variant Classification Process June 2021)  Method: clinical testing |
|  | rs6503640 |  | not provided  Affected status: unknown  Allele origin: germline | Benign  (Aug 16, 2021) | criteria provided, single submitter  (Invitae Variant Classification Sherloc (09022015)  Method: clinical testing |
|  | rs78951648 |  | not specified  Affected status: yes  Allele origin: germline | Likely benign  (Oct 16, 2017) | criteria provided, single submitter  (GeneDx Variant Classification (06012015)  Method: clinical testing |
|  |  |  | Hereditary cancer-predisposing syndrome  Affected status: unknown  Allele origin: germline | Likely benign  (Nov 06, 2017) | criteria provided, single submitter  (ACMG Guidelines, 2015  Method: clinical testing |
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