Resource Summary Report

Generated by NIF on Apr 28, 2025

UMD-BRCA1/ BRCA2 databases

RRID:SCR_006128
Type: Tool

Proper Citation

UMD-BRCA1/ BRCA2 databases (RRID:SCR_006128)

Resource Information

URL: http://www.umd.be/BRCA1/

Proper Citation: UMD-BRCA1/ BRCA2 databases (RRID:SCR_006128)

Description: The UMD-BRCA1/BRCA2 databases have been set up in a joined national effort through the network of 16 diagnostic laboratories to provide up-to-date information about mutations of the BRCA1 and BRCA2 genes identified in patients with breast and/or ovarian cancer. These databases currently contain published and unpublished information about the BRCA1/BRCA2 mutations reported in French diagnostic laboratories. This database includes 28 references and 5530 mutations (1440 different mutations and 786 protein variants) The databases of BRCA1 and BRCA2 mutations were built using the Universal Mutation Database tool. For each mutation, information is provided at several levels: * at the gene level: exon and codon number, wild type and mutant codon, mutation event, mutation name and, * at the protein level: wild type and mutant amino acid, binding domain, affected domain. If you want to submit a mutation, please contact R. Lidereau., S. Caputo. or E. Rouleau.

Abbreviations: UMD-BRCA1/ BRCA2 databases

Synonyms: UMD-BRCA1 mutations database, UMD-BRCA1 / BRCA2 databases, UMD-BRCA1/BRCA2 databases

Resource Type: data repository, service resource, storage service resource, database, data or information resource

Defining Citation: PMID:22144684

Keywords: cancer, gene, mutation, exon, codon, wild type, mutant, mutation, protein, amino acid, binding domain, affected domain, brca1, brca2, variant, polymorphism, unclassified

variant, unknown variant, female, woman, bio.tools

Related Condition: Breast cancer, Ovarian cancer

Funding: French National Cancer Institute ;
European Union FP7/2007-2013;
Association dAide a la Recherche Cancerologique de Saint Cloud

Availability: The UMD- BRCA1 Locus Specific Databases constitute the intellectual property of the curators of the database. Any unauthorized copying, Storage or distribution of this material without written permission from the curators would lead to copyright infringement with possible ensuing litigation.

Resource Name: UMD-BRCA1/ BRCA2 databases

Resource ID: SCR_006128

Alternate IDs: nlx_151608, biotools:brca_share

Alternate URLs: https://bio.tools/brca_share

Record Creation Time: 20220129T080234+0000

Record Last Update: 20250428T053214+0000

Ratings and Alerts

No rating or validation information has been found for UMD-BRCA1/ BRCA2 databases.

No alerts have been found for UMD-BRCA1/ BRCA2 databases.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 26 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Al Amri WS, et al. (2024) BRCA1/2 mutations and outcomes among Middle Eastern patients with early-onset breast cancer in Oman. The oncologist, 29(12), e1714.

Grafodatskaya D, et al. (2022) Practice guidelines for BRCA1/2 tumour testing in ovarian cancer. Journal of medical genetics, 59(8), 727.

Paik ES, et al. (2021) Prevalence and clinical characterization of BRCA1 and BRCA2 mutations in Korean patients with epithelial ovarian cancer. Cancer science, 112(12), 5055.

Toss A, et al. (2021) Clinicopathologic Profile of Breast Cancer in Germline ATM and CHEK2 Mutation Carriers. Genes, 12(5).

Salmi F, et al. (2021) Next-generation sequencing of BRCA1 and BRCA2 genes in Moroccan prostate cancer patients with positive family history. PloS one, 16(7), e0254101.

Billaud A, et al. (2021) Functional pre-therapeutic evaluation by genome editing of variants of uncertain significance of essential tumor suppressor genes. Genome medicine, 13(1), 174.

Ndiaye R, et al. (2020) Evidence for an ancient BRCA1 pathogenic variant in inherited breast cancer patients from Senegal. NPJ genomic medicine, 5, 8.

Wappenschmidt B, et al. (2020) Criteria of the German Consortium for Hereditary Breast and Ovarian Cancer for the Classification of Germline Sequence Variants in Risk Genes for Hereditary Breast and Ovarian Cancer. Geburtshilfe und Frauenheilkunde, 80(4), 410.

Nicolussi A, et al. (2019) Next-generation sequencing of BRCA1 and BRCA2 genes for rapid detection of germline mutations in hereditary breast/ovarian cancer. PeerJ, 7, e6661.

Golubeva VA, et al. (2019) Germline Missense Variants in BRCA1: New Trends and Challenges for Clinical Annotation. Cancers, 11(4).

Quaas A, et al. (2019) Alterations in ERBB2 and BRCA and microsatellite instability as new personalized treatment options in small bowel carcinoma. BMC gastroenterology, 19(1), 21.

Gornjec A, et al. (2019) Cytology material is equivalent to tumor tissue in determining mutations of BRCA 1/2 genes in patients with tubo-ovarian high grade serous carcinoma. BMC cancer, 19(1), 296.

Ong PY, et al. (2019) Using next-generation sequencing (NGS) platform to diagnose pathogenic germline BRCA1/2 mutations from archival tumor specimens. Gynecologic oncology, 155(2), 275.

Toland AE, et al. (2018) Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. NPJ genomic medicine, 3, 7.

Quaas A, et al. (2018) Therapy susceptible germline-related BRCA 1-mutation in a case of metastasized mixed adeno-neuroendocrine carcinoma (MANEC) of the small bowel. BMC gastroenterology, 18(1), 75.

Cusin I, et al. (2018) A new bioinformatics tool to help assess the significance of BRCA1 variants. Human genomics, 12(1), 36.

Sung PL, et al. (2017) The frequency of cancer predisposition gene mutations in hereditary breast and ovarian cancer patients in Taiwan: From BRCA1/2 to multi-gene panels. PloS

one, 12(9), e0185615.

Ratajska M, et al. (2017) Detection of BRCA1/2 mutations in circulating tumor DNA from patients with ovarian cancer. Oncotarget, 8(60), 101325.

Eliade M, et al. (2017) The transfer of multigene panel testing for hereditary breast and ovarian cancer to healthcare: What are the implications for the management of patients and families? Oncotarget, 8(2), 1957.

Thouvenot P, et al. (2016) Functional Assessment of Genetic Variants with Outcomes Adapted to Clinical Decision-Making. PLoS genetics, 12(6), e1006096.