

Resource Summary Report

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Familial Hypertrophic Cardiomyopathy DNA Mutation Database

RRID:SCR_002346

Type: Tool

Proper Citation

Familial Hypertrophic Cardiomyopathy DNA Mutation Database (RRID:SCR_002346)

Resource Information

URL: <http://www.angis.org.au/Databases/Heart/>

Proper Citation: Familial Hypertrophic Cardiomyopathy DNA Mutation Database (RRID:SCR_002346)

Description: THIS RESOURCE IS NO LONGER IN SERVICE, documented August 23, 2016. The aim of this locus-specific mutation database was to provide an online resource that contains summarized and updated information on familial hypertrophic cardiomyopathy (FHC)-associated mutations and related data, for researchers and clinicians. It also serves as a means of publishing previously unpublished data, which could be of value in understanding genotype/phenotype correlations. This database contains mutations in various genes known to cause familial hypertrophic cardiomyopathy, a genetic disorder associated with defects in the sarcomere [1]. Only gene symbols approved by HUGO are used and mutations are reported in accordance with guidelines recommended by the Mutation Database Initiative of HUGO and EBI.

Synonyms: FHC Mutation Database

Resource Type: data or information resource, database

Defining Citation: [PMID:10502780](#)

Keywords: familial, gene, gene-, genetic, cardiomyopathy, clinic, correlation, defect, disorder, genotype, hypertrophic, locus, mutation, or disease- specific databases, phenotype, research, sarcomere, system-

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: Familial Hypertrophic Cardiomyopathy DNA Mutation Database

Resource ID: SCR_002346

Alternate IDs: nif-0000-21151

Record Creation Time: 20220129T080212+0000

Record Last Update: 20260905T133117+0000

Ratings and Alerts

No rating or validation information has been found for Familial Hypertrophic Cardiomyopathy DNA Mutation Database.

No alerts have been found for Familial Hypertrophic Cardiomyopathy DNA Mutation Database.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 2 mentions in open access literature.

Listed below are recent publications. The full list is available at [NIF](#).

Xu Q, et al. (2010) Malignant and benign mutations in familial cardiomyopathies: insights into mutations linked to complex cardiovascular phenotypes. Journal of molecular and cellular cardiology, 48(5), 899.

Ababou A, et al. (2008) Myosin binding protein C positioned to play a key role in regulation of muscle contraction: structure and interactions of domain C1. Journal of molecular biology, 384(3), 615.