

Abstract

Arrhythmogenic cardiomyopathy (ACM) is an inherited cardiac disease characterized by myocardial atrophy, fibro-fatty replacement, and a high risk of ventricular arrhythmias that lead to sudden death. In 2009, genetic data from 57 publications were collected in the arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) Genetic Variants Database (freeware available at http://www.arvcdatabase.info), which comprised 481 variants in eight ACM-associated genes. In recent years, deep genetic sequencing has increased our knowledge of the genetics of ACM, revealing a large spectrum of nucleotide variations for which pathogenicity needs to be assessed. As of April 20, 2014, we have updated the ARVD/C database into the ARVD/C database to contain more than 1,400 variants in 12 ACM-related genes (PKP2, DSP, DSC2, DSG2, JUP, TGFB3, TMEM43, LMNA, DES, TTN, PLN, CTNNA3) as reported in more than 160 references. Of these, only 411 nucleotide variants have been reported as pathogenic, whereas the significance of the other approximately 1,000 variants is still unknown. This comprehensive collection of ACM genetic data represents a valuable source of information on the spectrum of ACM-associated genes and aims to facilitate the interpretation of genetic data and genetic counseling.

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