

## Updated gene list responsible for ARVC/D pathology

Subtype	Gene	Location	Reference
<b>ARVC1</b>	<b>TGFB3</b>	<b>14q24.3</b>	Beffagna G, Occhi G, Nava A, et al. Regulatory mutations in transforming growth factor-β3 gene cause arrhythmogenic right ventricular cardiomyopathy type 1. <i>Cardiovasc Res.</i> 2005;65:366–73.
<b>ARVC2</b>	<b>RYR2</b>	<b>1q43</b>	Tiso N, Stephan DA, Nava A, et al. Identification of mutations in the cardiac ryanodine receptor gene in families affected with arrhythmogenic right ventricular cardiomyopathy type 2 (ARVD2). <i>Hum Mol Genet.</i> 2001;10:189–94.
<b>ARVC3</b>	<b>Unknown</b>	<b>14q12–q22</b>	Severini GM, Krajinovic M, Pinamonti B, et al. A new locus for arrhythmogenic right ventricular dysplasia on the long arm of chromosome 14. <i>Genomics.</i> 1996;31:193–200.
<b>ARVC4</b>	<b>TTN</b>	<b>2q32.1–q32.3</b>	Taylor M, Graw S, Sinagra G, et al. Genetic variation in titin in arrhythmogenic right ventricular cardiomyopathy–overlap syndromes. <i>Circulation.</i> 2011;124:876–85.
<b>ARVC5</b>	<b>TMEM43</b>	<b>3p25.1</b>	Merner ND, Hodgkinson KA, Haywood AF, et al. Arrhythmogenic right ventricular cardiomyopathy type 5 is a fully penetrant, lethal arrhythmic disorder caused by a missense mutation in the TMEM43 gene. <i>Am J Hum Genet.</i> 2008;82:809–21.
<b>ARVC6</b>	<b>Unknown</b>	<b>10p14–p12</b>	Li D, Ahmad F, Gardner MJ, et al. The locus of a novel gene responsible for arrhythmogenic right-ventricular dysplasia characterized by early onset and high penetrance maps to chromosome 10p12–p14. <i>Am J Hum Genet.</i> 2000;66:148–56.
<b>ARVC7</b>	<b>DES</b>	<b>2q35</b>	Klauke B, Kossmann S, Gaertner A, et al. De novo desmin–mutation N116S is associated with arrhythmogenic right ventricular cardiomyopathy. <i>Hum Mol Genet.</i> 2010;19:4595–607.
<b>ARVC8 (Carvajal)</b>	<b>DSP</b>	<b>6p24.3</b>	Rampazzo A, Nava A, Malacrida S, et al. Mutation in human desmoplakin domain binding to plakoglobin causes a dominant form of arrhythmogenic right ventricular cardiomyopathy. <i>Am J Hum Genet.</i> 2002;71:1200–6.

<b>ARVC9</b>	<b>PKP2</b>	<b>12p11</b>	<b>Gerull B, Heuser A, Wichter T, et al. Mutations in the desmosomal protein plakophilin-2 are common in arrhythmogenic right ventricular cardiomyopathy. Nat Genet. 2004;36:1162–4.</b>
<b>ARVC10</b>	<b>DSG2</b>	<b>18q12.1</b>	Pilichou K, Nava A, Basso C, et al. Mutations in desmoglein-2 gene are associated with arrhythmogenic right ventricular cardiomyopathy. Circulation. 2006;113:1171–9.
<b>ARVC11</b>	<b>DSC2</b>	<b>18q12.1</b>	Syrris P, Ward D, Evans A, et al. Arrhythmogenic right ventricular dysplasia/cardiomyopathy associated with mutations in the desmosomal gene desmocollin-2. Am J Hum Genet. 2006;79:978–84.
<b>ARVC12 (Naxos)</b>	<b>JUP</b>	<b>17q21.2</b>	van der Zwaag PA, van Rijsingen IA, Asimaki A, et al. Phospholamban R14del mutation in patients diagnosed with dilated cardiomyopathy or arrhythmogenic right ventricular cardiomyopathy: evidence supporting the concept of arrhythmogenic cardiomyopathy. Eur J Heart Fail. 2012;14:1199–207.
<b>Others</b>	<b>PLN</b>	<b>6q22.1</b>	Quarta G, Syrris P, Ashworth M, et al. Mutations in the Lamin A/C gene mimic arrhythmogenic right ventricular cardiomyopathy. Eur Heart J. 2012;33:1128–36.

<b>ARVC1</b>	<b>LMNA</b>	<b>1q22</b>	
	<b>SCN5A</b>	<b>3p21</b>	Erkaptic D, Neumann T, Schmitt J, et al. Electrical storm in a patient with arrhythmogenic right ventricular cardiomyopathy and SCN5A mutation. Europace. 2008;10:884–7.
	<b>CTNNA3</b>	<b>10q22.2</b>	van Hengel J, Calore M, Bauce B, et al. Mutations in the area composita protein alphaT-catenin are associated with arrhythmogenic right ventricular cardiomyopathy. Eur Heart J. 2013;34:201–10.
	<b>CDH2</b>	<b>14q24.3</b>	Mayosi BM, Fish M, Shaboodien G, et al. Identification of cadherin 2 (CDH2) mutations in arrhythmogenic right ventricular cardiomyopathy. Circ Cardiovasc Genet. 2017;10:pii: e001605.