Updated gene list responsible for ARVC/D pathology

Subtype	Gene	Location	Reference
ARVC1	**TGFB3**	**14q24.3**	Beffagna G, Occhi G, Nava A, et al. Regulatory mutations in transforming growth factor−beta3 gene cause arrhythmogenic right ventricular cardiomyopathy type 1. Cardiovasc Res. 2005;65:366–73.
ARVC2	**RYR2**	**1q43**	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). Hum Mol Genet. 2001;10:189–94.
ARVC3	**Unknown**	**14q12−q22**	Severini GM, Krajinovic M, Pinamonti B, et al. A new locus for arrhythmogenic right ventricular dysplasia on the long arm of chromosome 14. Genomics. 1996;31:193–200.
ARVC4	**TTN**	**2q32.1−q32.3**	Taylor M, Graw S, Sinagra G, et al. Genetic variation in titin in arrhythmogenic right ventricular cardiomyopathy−overlap syndromes. Circulation. 2011;124:876–85.
ARVC5	**TMEM43**	**3p25.1**	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. Am J Hum Genet. 2008;82:809–21.
ARVC6	**Unknown**	**10p14−p12**	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. Am J Hum Genet. 2000;66:148–56.
ARVC7	**DES**	**2q35**	Klauke B, Kossmann S, Gaertner A, et al. De novo desmin−mutation N116S is associated with arrhythmogenic right ventricular cardiomyopathy. Hum Mol Genet. 2010;19:4595–607.
ARVC8 (Carvajal)	**DSP**	**6p24.3**	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. Am J Hum Genet. 2002;71:1200–6.

ARVC9	**PKP2**	**12p11**	**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.**
ARVC10	**DSG2**	**18q12.1**	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.
ARVC11	**DSC2**	**18q12.1**	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.
ARVC12 (Naxos)	**JUP**	**17q21.2**	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.
Others	**PLN**	**6q22.1**	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.

ARVC1	**LMNA**	**1q22**	
	SCN5A	**3p21**	Erkapic 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.
	CTNNA3	**10q22.2**	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.
	CDH2	**14q24.3**	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.