

Table S1. Published ARVC mutations¹

Type	Gene	Locus	Function
ARVC1	TGFβ3	14q23-q24	Transcription factor ligand, modulates cell adhesion and promotes myocardial fibrosis [2]
ARVC2	RYR2	1q42-q43	Calcium channel for calcium-induced calcium release (CICR), induces the release of calcium from the sarcoplasmic reticulum into the cytosol [3]
ARVC3	Unknown	14q12-q22	Unknown
ARVC4	Unknown	2q32.1-q32.3	Unknown
ARVC5	TMEM43	3p25	Unknown
ARVC6	Unknown	10p14-p12	Unknown
ARVC7	Unknown	10q22.3	Unknown
ARVC8	DSP	6p24	Anchors intermediate filaments to desmosomal plaques [4]
ARVC9	PKP2	12p11	Role in transcriptional regulation and links cadherens to intermediate filaments in desmosome plaque [5]
ARVC10	DSG2	18q12.1-q12	Calcium-binding transmembrane glycoprotein, involved in the interaction of plaque proteins and mediates cell-cell adhesion [6]
ARVC11	DSC2	18q12.1	Calcium-dependent glycoprotein member of the cadherin superfamily may contribute to desmosome and gap junction interdependence through connexin43 regulation [7]
ARVC12	JUP	17q21	Common junctional plaque protein. Forms complexes with cadherins and desmosomal cadherins (DSC2 and DSG2), plays a central role in the structure and function of submembranous plaques [8]

The table reflects the current available information of the ARVC mutations. Data was obtained from NCBI gene data base¹.

References

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