

# ARITMOGENE CARDIOMYOPATHY PANEL<sup>1</sup> DG-4.1.0 (9 GENES)

<i>Gene</i>	<i>Twist X2 covered &gt;10x</i>	<i>Twist X2 covered &gt;20x</i>	<i>WGS covered &gt;10x</i>	<i>WGS covered &gt;20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
DES	100%	100%	100%	98.2%	Scapulo-peroneal syndrome, neurogenic, Kaeser type, 181400; Cardiomyopathy, dilated, 11, 604765; Myopathy, myofibrillar, 1, 601419
DSC2	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476; Arrhythmogenic right ventricular dysplasia 11, 610476
DSG2	100%	99.9%	100%	99.6%	Cardiomyopathy, dilated, 1BB, 612877; Arrhythmogenic right ventricular dysplasia 10, 610193

DSP	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
FLNC	100%	100%	100%	99.1%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
JUP	100%	100%	100%	98.4%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
PKP2	99%	97.1%	100%	99.3%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLN	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874

TMEM43	100%	100%	100%	99%	Arrhythmogenic right ventricular dysplasia 5, 604400; Auditory neuropathy, autosomal dominant 3, 619832; Emery-Dreifuss muscular dystrophy 7, AD, 614302
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*Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*

*TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.*

*TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.*

*srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*

*srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*

*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023.*

*This list is accurate for panel version DG 4.0.0*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*