

# ARITMOGENE CARDIOMYOPATHY PANEL<sup>1</sup> DG-4.0.0 (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         | 100.0%                          | 100.0%                          | 100.0%                     | 98.9%                      | Scapulo-peroneal syndrome, neurogenic, Kaeser type, 181400; Cardiomyopathy, dilated, 11, 604765; Myopathy, myofibrillar, 1, 601419                              |
| DSC2        | 100.0%                          | 100.0%                          | 100.0%                     | 98.6%                      | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476; Arrhythmogenic right ventricular dysplasia 11, 610476 |
| DSG2        | 100.0%                          | 99.9%                           | 100.0%                     | 99.0%                      | Cardiomyopathy, dilated, 1BB, 612877; Arrhythmogenic right ventricular dysplasia 10, 610193   |

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|------|--------|--------|--------|-------|--|
| DSP  | 100.0% | 100.0% | 100.0% | 98.0% | 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 | 100.0% | 100.0% | 100.0% | 99.4% | 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  | 100.0% | 100.0% | 100.0% | 99.4% | Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528   |
| PKP2 | 98.4%  | 97.7%  | 99.9%  | 97.9% | Arrhythmogenic right ventricular dysplasia 9, 609040   |
| PLN  | 100.0% | 100.0% | 100.0% | 98.7% | Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874   |

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|--------|--------|--------|--------|-------|--|
| TMEM43 | 100.0% | 100.0% | 100.0% | 98.7% | Arrhythmogenic right ventricular dysplasia 5, 604400; Auditory neuropathy, autosomal dominant 3, 619832; Emery-Dreifuss muscular dystrophy 7, AD, 614302 |
|--------|--------|--------|--------|-------|--|

*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*