

ARITMOGENE CARDIOMYOPATHY PANEL¹ DG-3.9.0 (9 GENES)

| <i>Gene</i> | <i>Twist X2 covered >10x</i> | <i>Twist X2 covered >20x</i> | <i>WGS covered >10x</i> | <i>WGS covered >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% | 100.0% | 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 | 99.9% | 99.3% | 99.9% | 98.0% | 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 |
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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 3.9.0

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