

# WES ARITMOGENE CARDIOMYOPATHY <sup>1</sup> DG 3.8.1

| Gene | Twist X2 covered >10x | Twist X2 covered >20x | WGS covered >10x | WGS covered >20x | Associated Phenotype description and OMIM disease ID   |
|------|-----------------------|-----------------------|------------------|------------------|--|
| DES  | 100.0%                | 100.0%                | 100.0%           | 99.8%            | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419  |
| DSC2 | 100.0%                | 100.0%                | 100.0%           | 99.6%            | 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%           | 99.6%            | Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193   |
| DSP  | 100.0%                | 100.0%                | 100.0%           | 99.2%            | 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% | 99.9% | 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% | 99.7% | Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528  |
| PKP2   | 99.9%  | 99.3%  | 100.0% | 99.4% | Arrhythmogenic right ventricular dysplasia 9, 609040  |
| PLN    | 100.0% | 100.0% | 100.0% | 99.5% | Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874  |
| TMEM43 | 100.0% | 100.0% | 100.0% | 99.7% | Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302  |

Gene symbols used follow HGCN 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 3.7.0.