

# DILATED CARDIOMYOPATHY PANEL<sup>1</sup> DG-4.0.0 (21 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>  |
|-------------|---------------------------------|---------------------------------|----------------------------|----------------------------|--|
| ACTC1       | 100.0%                          | 100.0%                          | 100.0%                     | 99.5%                      | Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424   |
| ACTN2       | 100.0%                          | 100.0%                          | 99.9%                      | 97.9%                      | Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 |
| BAG3        | 100.0%                          | 100.0%                          | 100.0%                     | 98.2%                      | Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954   |
| DES         | 100.0%                          | 100.0%                          | 100.0%                     | 98.9%                      | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419  |

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|------|--------|--------|--------|-------|--|
| DMD  | 99.1%  | 98.7%  | 97.7%  | 70.8% | Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200  |
| 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  |
| HCN4 | 100.0% | 100.0% | 100.0% | 96.9% | Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123   |

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|------|--------|--------|--------|-------|---|
| JPH2 | 100.0% | 99.9%  | 100.0% | 99.1% | Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873  |
| LMNA | 100.0% | 100.0% | 100.0% | 99.2% | Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112 |

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|-------|--------|--------|--------|-------|---|
| MYH7  | 100.0% | 100.0% | 100.0% | 99.0% | Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358 |
| NEXN  | 100.0% | 100.0% | 99.9%  | 94.7% | Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876   |
| PLN   | 100.0% | 100.0% | 100.0% | 98.7% | Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874  |
| RBM20 | 100.0% | 100.0% | 100.0% | 99.2% | Cardiomyopathy, dilated, 1DD, 613172  |

|       |        |        |        |       |   |
|-------|--------|--------|--------|-------|---|
| SCN5A | 100.0% | 100.0% | 100.0% | 98.6% | Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120 |
| TNNC1 | 100.0% | 100.0% | 100.0% | 98.2% | Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243  |
| TNNI3 | 100.0% | 100.0% | 100.0% | 97.6% | ?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286   |

|       |        |        |        |       |   |
|-------|--------|--------|--------|-------|---|
| TNNT2 | 100.0% | 100.0% | 100.0% | 98.8% | Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494  |
| TPM1  | 100.0% | 100.0% | 100.0% | 98.0% | Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878  |
| TTN   | 99.6%  | 99.1%  | 100.0% | 98.6% | Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Cardiomyopathy, familial hypertrophic, 9, 613765;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;Myopathy, myofibrillar, 9, with early respiratory failure, 603689 |
| VCL   | 100.0% | 100.0% | 100.0% | 98.2% | Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255  |

*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*