

Tabella 1: CardioScreen® - Cardiomiopatie

Elenco dei geni analizzati e della malattie genetiche investigate

| | MALATTIA | PhenoMIM | GENE |
|----|--|------------------------|---------------|
| 1 | Atrial fibrillation, familial, 12 | 614050 | ABCC9 |
| 2 | Atrial septal defect 5 | 612794 | ACTC1 |
| 3 | Dilated cardiomyopathy 1AA | 612158 | ACTN2 |
| 4 | Ventricular tachycardia, catecholaminergic polymorphic, 2 | 611938 | CASQ2 |
| 5 | Cardiomyopathy, familial hypertrophic | 192600 | CAV3 |
| 6 | Cardiomyopathy, dilated, 1II | 615184 | CRYAB |
| 7 | Cardiomyopathy, dilated, 1M | 607482 | CSRP3 |
| 8 | Cardiomyopathy, dilated, 1I | 604765 | DES |
| 9 | Arrhythmogenic right ventricular dysplasia 11 | 610476 | DSC2 |
| 10 | Arrhythmogenic right ventricular dysplasia 10 | 610193 | DSG2 |
| 11 | Arrhythmogenic right ventricular dysplasia 8 | 607450 | DSP |
| 12 | Left ventricular noncompaction 1, with or without congenital heart defects | 604169 | DTNA |
| 13 | Emery-Dreifuss muscular dystrophy 1, X-linked | 310300 | EMD |
| 14 | Fabry disease, cardiac variant | 301500 | GLA |
| 15 | Arrhythmogenic right ventricular dysplasia 12 | 611528 | JUP |
| 16 | Cardiomyopathy, dilated, 1JJ | 615235 | LAMA4 |
| 17 | Danon disease | 300257 | LAMP2 |
| 18 | Cardiomyopathy, dilated, 1C, with or without LVNC | 601493 | LDB3 |
| 19 | Cardiomyopathy, dilated, 1A | 115200 | LMNA |
| 20 | Cardiomyopathy, dilated, 1MM | 615396 | MYBPC3 |
| 21 | Atrial septal defect 3 | 614089 | MYH6 |
| 22 | Cardiomyopathy, dilated, 1S | 613426 | MYH7 |
| 23 | Cardiomyopathy, hypertrophic, 10 | 608758 | MYL2 |
| 24 | Cardiomyopathy, hypertrophic, 8 | 608751 | MYL3 |
| 25 | Cardiomyopathy, hypertrophic, 1, digenic | 192600 | MYLK2 |
| 26 | Cardiomyopathy, hypertrophic, 16 | 613838 | MYOZ2 |
| 27 | Cardiomyopathy, dilated, 1CC | 613122 | NEXN |
| 28 | Arrhythmogenic right ventricular dysplasia 9 | 609040 | PKP2 |
| 29 | Cardiomyopathy, dilated, 1P | 609909 | PLN |
| 30 | Cardiomyopathy, hypertrophic 6 | 600858 | PRKAG2 |
| 31 | Cardiomyopathy, dilated, 1DD | 613172 | RBM20 |

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|----|--|------------------------|---------------|
| 32 | Arrhythmogenic right ventricular dysplasia 2 | 600996 | RYR2 |
| 33 | Cardiomyopathy, dilated, 1L | 606685 | SGCD |
| 34 | Barth syndrome | 302060 | TAZ |
| 35 | Cardiomyopathy, hypertrophic, 25 | 607487 | TCAP |
| 36 | Arrhythmogenic right ventricular dysplasia 5 | 604400 | TMEM43 |
| 37 | Cardiomyopathy, dilated, 1Z | 611879 | TNNC1 |
| 38 | Cardiomyopathy, dilated, 2A | 611880 | TNNI3 |
| 39 | Cardiomyopathy, dilated, 1D | 601494 | TNNT2 |
| 40 | Cardiomyopathy, dilated, 1Y | 611878 | TPM1 |
| 41 | Cardiomyopathy, dilated, 1G | 604145 | TTN |
| 42 | Amyloidosis, hereditary, transthyretin-related | 105210 | TTR |
| 43 | Cardiomyopathy, dilated, 1W | 611407 | VCL |