

Tabella 1: CardioScreen® - Cardiomiopatie

Elenco dei geni analizzati e della malattie genetiche investigate

| | MALATTIA | PhenoMIM | GENE |
|----|--|---------------|---------------|
| 1 | Atrial fibrillation, familial, 12 | <u>614050</u> | ABCC9 |
| 2 | Atrial septal defect 5 | <u>612794</u> | ACTC1 |
| 3 | Dilated cardiomyopathy 1AA | <u>612158</u> | ACTN2 |
| 4 | Cardiomyopathy, hypertrophic/Cardiomyopathy, dilated | 609599 | ANKRD1 |
| 5 | Ventricular tachycardia, catecholaminergic polymorphic, 2 | <u>611938</u> | CASQ2 |
| 6 | Cardiomyopathy, familial hypertrophic | <u>192600</u> | CAV3 |
| 7 | Cardiomyopathy, dilated, 1II | <u>615184</u> | CRYAB |
| 8 | Cardiomyopathy, dilated, 1M | <u>607482</u> | CSRP3 |
| 9 | Cardiomyopathy, dilated | 600435 | CTF1 |
| 10 | Cardiomyopathy, dilated, 1I | <u>604765</u> | DES |
| 11 | Arrhythmogenic right ventricular dysplasia 11 | <u>610476</u> | DSC2 |
| 12 | Arrhythmogenic right ventricular dysplasia 10 | <u>610193</u> | DSG2 |
| 13 | Arrhythmogenic right ventricular dysplasia 8 | <u>607450</u> | DSP |
| 14 | Left ventricular noncompaction 1, with or without congenital heart defects | <u>604169</u> | DTNA |
| 15 | Emery-Dreifuss muscular dystrophy 1, X-linked | <u>310300</u> | EMD |
| 16 | Cardiomyopathy, dilated | 602633 | FHL2 |
| 17 | Fabry disease, cardiac variant | <u>301500</u> | GLA |
| 18 | Arrhythmogenic right ventricular dysplasia 12 | <u>611528</u> | JUP |
| 19 | Cardiomyopathy, dilated, 1JJ | <u>615235</u> | LAMA4 |
| 20 | Danon disease | <u>300257</u> | LAMP2 |
| 21 | Cardiomyopathy, dilated, 1C, with or without LVNC | <u>601493</u> | LDB3 |
| 22 | Cardiomyopathy, dilated, 1A | <u>115200</u> | LMNA |
| 23 | Cardiomyopathy, dilated, 1MM | <u>615396</u> | MYBPC3 |
| 24 | Atrial septal defect 3 | <u>614089</u> | MYH6 |
| 25 | Cardiomyopathy, dilated, 1S | <u>613426</u> | MYH7 |
| 26 | Cardiomyopathy, hypertrophic, 10 | <u>608758</u> | MYL2 |
| 27 | Cardiomyopathy, hypertrophic, 8 | <u>608751</u> | MYL3 |
| 28 | Cardiomyopathy, hypertrophic, 1, digenic | 192600 | MYLK2 |
| 29 | Cardiomyopathy, hypertrophic, 16 | <u>613838</u> | MYOZ2 |
| 30 | Cardiomyopathy, dilated, 1CC | <u>613122</u> | NEXN |
| 31 | Arrhythmogenic right ventricular dysplasia 9 | <u>609040</u> | PKP2 |

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| 32 | Cardiomyopathy, dilated, 1P | <u>609909</u> | PLN |
| 33 | Cardiomyopathy, hypertrophic 6 | <u>600858</u> | PRKAG2 |
| 34 | Cardiomyopathy, dilated, 1DD | <u>613172</u> | RBM20 |
| 35 | Arrhythmogenic right ventricular dysplasia 2 | <u>600996</u> | RYR2 |
| 36 | Cardiomyopathy, dilated, 1L | <u>606685</u> | SGCD |
| 37 | Barth syndrome | <u>302060</u> | TAZ |
| 38 | Cardiomyopathy, hypertrophic, 25 | <u>607487</u> | TCAP |
| 39 | Arrhythmogenic right ventricular dysplasia 5 | <u>604400</u> | TMEM43 |
| 40 | Cardiomyopathy, dilated, 1Z | <u>611879</u> | TNNC1 |
| 41 | Cardiomyopathy, dilated, 2A | <u>611880</u> | TNNI3 |
| 42 | Cardiomyopathy, dilated, 1D | <u>601494</u> | TNNT2 |
| 43 | Cardiomyopathy, dilated, 1Y | <u>611878</u> | TPM1 |
| 44 | Cardiomyopathy, dilated, 1G | <u>604145</u> | TTN |
| 45 | Amyloidosis, hereditary, transthyretin-related | <u>105210</u> | TTR |
| 46 | Cardiomyopathy, dilated, 1W | <u>611407</u> | VCL |