Tabella 1: CardioScreen® - Prevenzione arresto cardiaco improvviso.

Elenco dei geni analizzati e della malattie genetiche investigate

	DISEASE NAME	PhenoMIM	GENE
1	Atrial fibrillation, familial, 12	614050	ABCC9
2	Sitosterolemia	210250	ABCG5
3	Sitosterolemia	210250	ABCG8
4	Myopathy, actin, congenital, with cores	161800	ACTA1
5	Aortic aneurysm, familial thoracic 6	611788	ACTA2
6	Atrial septal defect 5	612794	ACTC1
7	Cardiomyopathy, dilated, 1AA, with or without LVNC	612158	ACTN2
8	Long QT syndrome-11	611820	AKAP9
9	Alstrom syndrome	203800	ALMS1
10	Cardiac arrhythmia, ankyrin-B-related	600919	ANK2
11	Hyperchylomicronemia, late-onset	144650	APOA5
12	Hypercholesterolemia, due to ligand-defective apo B	144010	APOB
13	Hyperlipoproteinemia, type Ib	207750	APOC2
14	Lipoprotein glomerulopathy	611771	APOE
15	Cardiomyopathy, dilated, 1HH	613881	BAG3
16	Cardiofaciocutaneous syndrome	115150	BRAF
17	Brugada syndrome 3	611875	CACNA1C
18	Brugada syndrome 4	611876	CACNB2
19	Long QT syndrome 14	616247	CALM1
	Ventricular tachycardia, catecholaminergic polymorphic, 4	614916	
20	Cardiomyopathy, hypertrophic, 19	613875	CALR3
21	Ventricular tachycardia, catecholaminergic polymorphic, 2	611938	CASQ2
22	Cardiomyopathy, familial hypertrophic	192600	CAV3
	Long QT syndrome 9	611818	
23	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	613563	CBL
24	Homocystinuria, B6-responsive and nonresponsive types	236200	CBS
25	Hyperalphalipoproteinemia	143470	CETP
26	Ehlers-Danlos syndrome, type III	130020	COL3A1
27	Ehlers-Danlos syndrome, classic type	130000	COL5A1
28	Ehlers-Danlos syndrome, classic type	130000	COL5A2
29	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	615119	COX15
30	Atrioventricular septal defect, partial, with heterotaxy syndrome	606217	CRELD1
31	Cardiomyopathy, dilated, 1II	615184	CRYAB

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CHIAMATA GRATUITA
NUMEROVERDE
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32	Cardiomyopathy, dilated, 1M	607482	CSRP3
33	Cardiomyopathy, dilated, 1I	604765	DES
34	Cardiomyopathy, dilated, 3B	302045	DMD
35	3-methylglutaconic aciduria, type V	610198	DNAJC19
36	Congenital disorder of glycosylation, type Im	610768	DOLK
37	Ventricular fibrillation, paroxysmal familial, 2	612956	DPP6
38	Arrhythmogenic right ventricular dysplasia 11	610476	DSC2
39	Arrhythmogenic right ventricular dysplasia 10	610193	DSG2
40	Arrhythmogenic right ventricular dysplasia 8	607450	DSP
41	Left ventricular noncompaction 1, with or without congenital heart defects	604169	DTNA
42	Cutis laxa, autosomal recessive, type IB	614437	EFEMP2
43	Supravalvar aortic stenosis	185500	ELN
44	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	EMD
45	Cardiomyopathy, dilated, 1J	605362	EYA4
46	Marfan syndrome	154700	FBN1
47	Contractural arachnodactyly, congenital	121050	FBN2
48	Emery-Dreifuss muscular dystrophy 6, X-linked	300696	FHL1
49	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153	FKRP
50	Cardiomyopathy, dilated, 1X	611615	FKTN
51	Friedreich ataxia	229300	FXN
52	Glycogen storage disease II	232300	GAA
53	Cardiomyopathy, dilated, 2B	614672	GATAD1
54	Fasting plasma glucose level QTL 5	613463	GCKR
55	Atrial fibrillation, familial, 11	614049	GJA5
56	Fabry disease, cardiac variant	301500	GLA
57	Pseudohypoparathyroidism	612463	GNAS
58	Brugada syndrome 2	611777	GPD1L
59	Hyperlipoproteinemia, type 1D	615947	GPIHBP1
60	LCHAD deficiency	609016	HADHA
61	Brugada syndrome 8	613123	HCN4
62	Hemochromatosis	235200	HFE
63	Congenital myopathy with excess of muscle spindles	218040	HRAS
64	Neuropathy, distal hereditary motor, type IIA	158590	HSPB8
65	Alagille syndrome	118450	JAG1
66	Cardiomyopathy, hypertrophic, 17	613873	JPH2
67	Arrhythmogenic right ventricular dysplasia 12	611528	JUP
68	Atrial fibrillation, familial, 7	612240	KCNA5

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69	Brugada syndrome 9	616399	KCND3
70	Long QT syndrome 5	613695	KCNE1
71	Long QT syndrome 6	613693	KCNE2
72	Brugada syndrome 6	613119	KCNE3
73	Long QT syndrome 2	613688	KCNH2
	Short QT syndrome 1	609620	
	Long QT syndrome 2, acquired, susceptibility to	613688	
74	Atrial fibrillation, familial, 9	613980	KCNJ2
75	Long QT syndrome 13	613485	KCNJ5
76	Atrial fibrillation, familial, 3	607554	KCNQ1
	Long QT syndrome 1	609621	
	Short QT syndrome 2	192500	
77	Cardiofaciocutaneous syndrome 2	615278	KRAS
78	Muscular dystrophy, congenital merosin-deficient	607855	LAMA2
79	Cardiomyopathy, dilated, 1JJ	615235	LAMA4
80	Danon disease	300257	LAMP2
81	Cardiomyopathy, dilated, 1C, with or without LVNC	601493	LDB3
	Cardiomyopathy, hypertrophic, 24	601493	
	Left ventricular noncompaction 3	601493	
82	Hypercholesterolemia, familial	143890	LDLR
83	Hypercholesterolemia, familial, autosomal recessive	603813	LDLRAP1
84	Lipase deficiency, combined	246650	LMF1
85	Cardiomyopathy, dilated, 1A	115200	LMNA
86	Combined hyperlipidemia, familial	144250	LPL
87	Weill-Marchesani syndrome 3, recessive	614819	LTBP2
88	Cardiofaciocutaneous syndrome 3	615279	MAP2K1
89	Cardiofaciocutaneous syndrome 4	615280	MAP2K2
90	Left ventricular noncompaction 7	615092	MIB1
91	Cardiomyopathy, dilated, 1MM	615396	MYBPC3
92	Aortic aneurysm, familial thoracic 4	132900	MYH11
93	Atrial septal defect 3	614089	MYH6
94	Cardiomyopathy, dilated, 1S	613426	MYH7
95	Cardiomyopathy, hypertrophic, 10	608758	MYL2
96	Cardiomyopathy, hypertrophic, 8	608751	MYL3
97	Aortic aneurysm, familial thoracic 7	613780	MYLK
98	Cardiomyopathy, hypertrophic, 1, digenic	192600	MYLK2
99	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy	606346	MYO6
100	Cardiomyopathy, hypertrophic, 16	613838	MYOZ2
101	Cardiomyopathy, dilated, 1KK	615248	MYPN

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102	Cardiomyopathy, dilated, 1CC	613122	NEXN
103	Atrial septal defect 7, with or without AV conduction defects	108900	NKX2-5
104	Heterotaxy, visceral, 5	270100	NODAL
105	Aortic valve disease 1	109730	NOTCH1
106	Atrial fibrillation, familial, 6	612201	NPPA
107	Autoimmune lymphoproliferative syndrome type IV	614470	NRAS
108	Hypercholesterolemia, familial, 3	603776	PCSK9
109	Arrhythmogenic right ventricular dysplasia 9	609040	PKP2
110	Cardiomyopathy, dilated, 1P	609909	PLN
111	Cardiomyopathy, dilated, 1LL	615373	PRDM16
112	Cardiomyopathy, hypertrophic 6	600858	PRKAG2
113	Myxoma, intracardiac	255960	PRKAR1A
114	LEOPARD syndrome 1	151100	PTPN11
115	Cardiomyopathy, dilated, 1NN	615916	RAF1
116	Cardiomyopathy, dilated, 1DD	613172	RBM20
117	Central core disease	117000	RYR1
118	Arrhythmogenic right ventricular dysplasia 2	600996	RYR2
119	IVIC syndrome	147750	SALL4
120	Atrial fibrillation, familial, 13	615377	SCN1B
	Brugada syndrome 5	612838	
	Cardiac conduction defect, nonspecific	612838	
121	Atrial fibrillation, familial, 14	615378	SCN2B
122	Atrial fibrillation, familial, 16	613120	SCN3B
123	Atrial fibrillation, familial, 17	611819	SCN4B
124	Atrial fibrillation, familial, 10	614022	SCN5A
125	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	SCO2
126	Cardiomyopathy, dilated, 1GG	613642	SDHA
127	Myopathy, congenital, with fiber-type disproportion	255310	SEPN1
128	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB
129	Cardiomyopathy, dilated, 1L	606685	SGCD
130	Muscular dystrophy, limb-girdle, type 2C	253700	SGCG
131	Noonan-like syndrome with loose anagen hair	607721	SHOC2
132	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	615418	SLC25A4
133	Arterial tortuosity syndrome	208050	SLC2A10
134	Loeys-Dietz syndrome, type 3	613795	SMAD3
135	Myhre syndrome	139210	SMAD4
136	Long QT syndrome 12	612955	SNTA1
137	Noonan syndrome 4	610733	SOS1

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138	Barth syndrome	302060	**TAZ**
139	Atrial septal defect 4	611363	**TBX20**
140	Ulnar-mammary syndrome	181450	**TBX3**
141	Holt-Oram syndrome	142900	**TBX5**
142	Cardiomyopathy, hypertrophic, 25	607487	**TCAP**
143	Loeys-Dietz syndrome, type 4	614816	**TGFB2**
144	Arrhythmogenic right ventricular dysplasia 1	107970	**TGFB3**
145	Loeys-Dietz syndrome, type 1	609192	**TGFBR1**
146	Loeys-Dietz syndrome, type 2	610168	**TGFBR2**
147	Arrhythmogenic right ventricular dysplasia 5	604400	**TMEM43**
148	Cardiomyopathy, dilated, 1T	613740	**TMPO**
149	Cardiomyopathy, dilated, 1Z	611879	**TNNC1**
150	Cardiomyopathy, dilated, 1FF	613286	**TNNI3**
151	Cardiomyopathy, dilated, 1D	601494	**TNNT2**
152	Cardiomyopathy, dilated, 1Y	611878	**TPM1**
153	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	615441	**TRDN**
154	Progressive familial heart block, type IB	604559	**TRPM4**
155	Cardiomyopathy, dilated, 1G	604145	**TTN**
156	Amyloidosis, hereditary, transthyretin-related	105210	**TTR**
157	Cardiomyopathy, dilated, 1W	611407	**VCL**
	Cardiomyopathy, hypertrophic, 15	613255	
158	Congenital heart defects, nonsyndromic, 1, X-linked	306955	**ZIC3**

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