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	Cardiomyopathy, hypertrophic/Cardiomyopathy, dilated	609599	ANKRD1
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

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CHIAMATA GRATUITA
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30	Atrioventricular septal defect, partial, with heterotaxy syndrome	606217	CRELD1
31	Cardiomyopathy, dilated, 1II	615184	CRYAB
32	Cardiomyopathy, dilated, 1M	607482	CSRP3
33	Cardiomyopathy, dilated	600435	CTF1
34	Cardiomyopathy, dilated, 1I	604765	DES
35	Cardiomyopathy, dilated, 3B	302045	DMD
36	3-methylglutaconic aciduria, type V	610198	DNAJC19
37	Congenital disorder of glycosylation, type Im	610768	DOLK
38	Ventricular fibrillation, paroxysmal familial, 2	612956	DPP6
39	Arrhythmogenic right ventricular dysplasia 11	610476	DSC2
40	Arrhythmogenic right ventricular dysplasia 10	610193	DSG2
41	Arrhythmogenic right ventricular dysplasia 8	607450	DSP
42	Left ventricular noncompaction 1, with or without congenital heart defects	604169	DTNA
43	Cutis laxa, autosomal recessive, type IB	614437	EFEMP2
44	Supravalvar aortic stenosis	185500	ELN
45	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	EMD
46	Cardiomyopathy, dilated, 1J	605362	EYA4
47	Marfan syndrome	154700	FBN1
48	Contractural arachnodactyly, congenital	121050	FBN2
49	Emery-Dreifuss muscular dystrophy 6, X-linked	300696	FHL1
50	Cardiomyopathy, dilated	602633	FHL2
51	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153	FKRP
52	Cardiomyopathy, dilated, 1X	611615	FKTN
53	Friedreich ataxia	229300	FXN
54	Glycogen storage disease II	232300	GAA
55	Cardiomyopathy, dilated, 2B	614672	GATAD1
56	Fasting plasma glucose level QTL 5	613463	GCKR
57	Atrial fibrillation, familial, 11	614049	GJA5
58	Fabry disease, cardiac variant	301500	GLA
59	Pseudohypoparathyroidism	612463	GNAS
60	Brugada syndrome 2	611777	GPD1L
61	Hyperlipoproteinemia, type 1D	615947	GPIHBP1
62	LCHAD deficiency	609016	HADHA
63	Brugada syndrome 8	613123	HCN4
64	Hemochromatosis	235200	HFE
65	Congenital myopathy with excess of muscle spindles	218040	HRAS

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66	Neuropathy, distal hereditary motor, type IIA	158590	HSPB8
67	Alagille syndrome	118450	JAG1
68	Cardiomyopathy, hypertrophic, 17	613873	JPH2
69	Arrhythmogenic right ventricular dysplasia 12	611528	JUP
70	Atrial fibrillation, familial, 7	612240	KCNA5
71	Brugada syndrome 9	616399	KCND3
72	Long QT syndrome 5	613695	KCNE1
73	Long QT syndrome 6	613693	KCNE2
74	Brugada syndrome 6	613119	KCNE3
75	Long QT syndrome 2	613688	KCNH2
	Short QT syndrome 1	609620	
	Long QT syndrome 2, acquired, susceptibility to	613688	
76	Atrial fibrillation, familial, 9	613980	KCNJ2
77	Long QT syndrome 13	613485	KCNJ5
78	Atrial fibrillation, familial, 3	607554	KCNQ1
	Long QT syndrome 1	609621	
	Short QT syndrome 2	192500	
79	Cardiofaciocutaneous syndrome 2	615278	KRAS
80	Muscular dystrophy, congenital merosin-deficient	607855	LAMA2
81	Cardiomyopathy, dilated, 1JJ	615235	LAMA4
82	Danon disease	300257	LAMP2
83	Cardiomyopathy, dilated, 1C, with or without LVNC	601493	LDB3
	Cardiomyopathy, hypertrophic, 24	601493	
	Left ventricular noncompaction 3	601493	
84	Hypercholesterolemia, familial	143890	LDLR
85	Hypercholesterolemia, familial, autosomal recessive	603813	LDLRAP1
86	Lipase deficiency, combined	246650	LMF1
87	Cardiomyopathy, dilated, 1A	115200	LMNA
88	Combined hyperlipidemia, familial	144250	LPL
89	Weill-Marchesani syndrome 3, recessive	614819	LTBP2
90	Cardiofaciocutaneous syndrome 3	615279	MAP2K1
91	Cardiofaciocutaneous syndrome 4	615280	MAP2K2
92	Left ventricular noncompaction 7	615092	MIB1
93	Cardiomyopathy, dilated, 1MM	615396	MYBPC3
94	Aortic aneurysm, familial thoracic 4	132900	MYH11
95	Atrial septal defect 3	614089	MYH6
96	Cardiomyopathy, dilated, 1S	613426	MYH7
97	Cardiomyopathy, hypertrophic, 10	608758	MYL2
98	Cardiomyopathy, hypertrophic, 8	608751	MYL3

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99	Aortic aneurysm, familial thoracic 7	613780	MYLK
100	Cardiomyopathy, hypertrophic, 1, digenic	192600	MYLK2
101	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy	606346	MYO6
102	Cardiomyopathy, hypertrophic, 16	613838	MYOZ2
103	Cardiomyopathy, dilated, 1KK	615248	MYPN
104	Cardiomyopathy, dilated, 1CC	613122	NEXN
105	Atrial septal defect 7, with or without AV conduction defects	108900	NKX2-5
106	Heterotaxy, visceral, 5	270100	NODAL
107	Aortic valve disease 1	109730	NOTCH1
108	Atrial fibrillation, familial, 6	612201	NPPA
109	Autoimmune lymphoproliferative syndrome type IV	614470	NRAS
110	Hypercholesterolemia, familial, 3	603776	PCSK9
111	Arrhythmogenic right ventricular dysplasia 9	609040	PKP2
112	Cardiomyopathy, dilated, 1P	609909	PLN
113	Cardiomyopathy, dilated, 1LL	615373	PRDM16
114	Cardiomyopathy, hypertrophic 6	600858	PRKAG2
115	Myxoma, intracardiac	255960	PRKAR1A
116	LEOPARD syndrome 1	151100	PTPN11
117	Cardiomyopathy, dilated, 1NN	615916	RAF1
118	Cardiomyopathy, dilated, 1DD	613172	RBM20
119	Central core disease	117000	RYR1
120	Arrhythmogenic right ventricular dysplasia 2	600996	RYR2
121	IVIC syndrome	147750	SALL4
122	Atrial fibrillation, familial, 13	615377	SCN1B
	Brugada syndrome 5	612838	
	Cardiac conduction defect, nonspecific	612838	
123	Atrial fibrillation, familial, 14	615378	SCN2B
124	Atrial fibrillation, familial, 16	613120	SCN3B
125	Atrial fibrillation, familial, 17	611819	SCN4B
126	Atrial fibrillation, familial, 10	614022	SCN5A
127	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	604377	SCO2
128	Cardiomyopathy, dilated, 1GG	613642	SDHA
129	Myopathy, congenital, with fiber-type disproportion	255310	SEPN1
130	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB
131	Cardiomyopathy, dilated, 1L	606685	SGCD
132	Muscular dystrophy, limb-girdle, type 2C	253700	SGCG
133	Noonan-like syndrome with loose anagen hair	607721	SHOC2
134	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	615418	SLC25A4

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135	Arterial tortuosity syndrome	208050	SLC2A10
136	Loeys-Dietz syndrome, type 3	613795	SMAD3
137	Myhre syndrome	139210	SMAD4
138	Long QT syndrome 12	612955	SNTA1
139	Noonan syndrome 4	610733	SOS1
140	Barth syndrome	302060	TAZ
141	Atrial septal defect 4	611363	TBX20
142	Ulnar-mammary syndrome	181450	TBX3
143	Holt-Oram syndrome	142900	TBX5
144	Cardiomyopathy, hypertrophic, 25	607487	TCAP
145	Loeys-Dietz syndrome, type 4	614816	TGFB2
146	Arrhythmogenic right ventricular dysplasia 1	107970	TGFB3
147	Loeys-Dietz syndrome, type 1	609192	TGFBR1
148	Loeys-Dietz syndrome, type 2	610168	TGFBR2
149	Arrhythmogenic right ventricular dysplasia 5	604400	TMEM43
150	Cardiomyopathy, dilated, 1T	613740	TMPO
151	Cardiomyopathy, dilated, 1Z	611879	TNNC1
152	Cardiomyopathy, dilated, 1FF	613286	TNNI3
153	Cardiomyopathy, dilated, 1D	601494	TNNT2
154	Cardiomyopathy, dilated, 1Y	611878	TPM1
155	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	615441	TRDN
156	Progressive familial heart block, type IB	604559	TRPM4
157	Cardiomyopathy, dilated, 1G	604145	TTN
158	Amyloidosis, hereditary, transthyretin-related	105210	TTR
159	Cardiomyopathy, dilated, 1W	611407	VCL
	Cardiomyopathy, hypertrophic, 15	613255	
160	Congenital heart defects, nonsyndromic, 1, X-linked	306955	ZIC3

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