Cardiology & Pulmonology

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Single Gene

Code	Diseases	Gene
sCe6/0001	Alveolar capillary dysplasia with misalignment of pulmonary veins	FOXF1
sCe6/0002	Arrhythmogenic right ventricular cardiomyopathy type 1	TGFB3
sCe6/0003	Arrhythmogenic right ventricular cardiomyopathy type 10	DSG2
sCe6/0004	Arrhythmogenic right ventricular cardiomyopathy type 11	DSC2
sCe6/0005	Arrhythmogenic right ventricular cardiomyopathy type 12	JUP
sCe6/0006	Arrhythmogenic right ventricular cardiomyopathy type 5	TMEM43
sCe6/0007	Arrhythmogenic right ventricular cardiomyopathy type 8	DSP
sCe6/0008	Arrhythmogenic right ventricular cardiomyopathy type 9	PKP2
sCe6/0009	Arrhythmogenic right ventricular dysplasia type 2	RYR2
sCe6/0010	Atrial fibrillation type 10	SCN5A
sCe6/0011	Atrial fibrillation type 11	GJA5
sCe6/0012	Atrial fibrillation type 12	ABCC9
sCe6/0013	Atrial fibrillation type 3	KCNQ1
sCe6/0014	Atrial fibrillation type 4	KCNE2
sCe6/0015	Atrial fibrillation type 6	NPPA
sCe6/0016	Atrial fibrillation type 7	KCNA5
sCe6/0017	Atrial septal defect type 3	MYH6
sCe6/0018	Atrial septal defect type 4	TBX20
sCe6/0019	Atrial septal defect type 5	ACTC1
sCe6/0020	Atrial septal defect type 8	CITED2
sCe6/0021	Atrial septal defect type 9	GATA6
sCe6/0022	Atrioventricular septal defect type 4	GATA4
sCe6/0023	Atrioventricular septal defect type 5	GATA6
sCe6/0024	Barth syndrome	TAZ
sCe6/0025	Bicuspid aortic valve	TIMP1

sCe6/0026	Brugada syndrome type 1	SCN5A
sCe6/0027	Brugada syndrome type 2	GPD1L
sCe6/0028	Brugada syndrome type 3	CACNA1C
sCe6/0029	Brugada syndrome type 4	CACNB2
sCe6/0030	Brugada syndrome type 5	SCN1B
sCe6/0031	Brugada syndrome type 6	KCNE3
sCe6/0032	Brugada syndrome type 7	SCN3B
sCe6/0033	Brugada syndrome type 8	HCN4
sCe6/0034	Brugada syndrome type 9	SLMAP
sCe6/0035	Cardiac defects, CNOT3 related	CNOT3
sCe6/0036	Cardiac defects, PPP1R8 related	PPP1R8
sCe6/0037	Cardiac valvular dysplesia, X-linked	FLNA
sCe6/0038	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	SCO2
sCe6/0039	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 2	COX15
sCe6/0040	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency type 3	COA5
sCe6/0041	Cardiofaciocutaneous syndrome	KRAS
sCe6/0042	Cardiofaciocutaneous syndrome	BRAF
sCe6/0043	Cardiofaciocutaneous syndrome type 3	MAP2K1
sCe6/0044	Cardiofaciocutaneous syndrome type 4	MAP2K2
sCe6/0045	Cardiomyopathy, apical hypertrophic, and neuropathy, MT-ATP8 related	MT-ATP8
sCe6/0046	Cardiomyopathy, dilated	MYBPC3
sCe6/0047	Cardiomyopathy, dilated type 1	CRYAB
sCe6/0048	Cardiomyopathy, dilated type 1A	LMNA
sCe6/0049	Cardiomyopathy, dilated type 1AA	ACTN2
sCe6/0050	Cardiomyopathy, dilated type 1BB	DSG2
sCe6/0051	Cardiomyopathy, dilated type 1C	LDB3
sCe6/0052	Cardiomyopathy, dilated type 1CC	NEXN

sCe6/0053	Cardiomyopathy, dilated type 1D	TNNT2
sCe6/0054	Cardiomyopathy, dilated type 1DD	RBM20
sCe6/0055	Cardiomyopathy, dilated type 1E	SCN5A
sCe6/0056	Cardiomyopathy, dilated type 1EE	MYH6
sCe6/0057	Cardiomyopathy, dilated type 1G	TTN
sCe6/0058	Cardiomyopathy, dilated type 1GG	SDHA
sCe6/0059	Cardiomyopathy, dilated type 1HH	BAG3
sCe6/0060	Cardiomyopathy, dilated type 1I	DES
sCe6/0061	Cardiomyopathy, dilated type 1J	EYA4
sCe6/0062	Cardiomyopathy, dilated type 1KK	MYPN
sCe6/0063	Cardiomyopathy, dilated type 1L	SGCD
sCe6/0064	Cardiomyopathy, dilated type 1LL	PRDM16
sCe6/0065	Cardiomyopathy, dilated type 1M	CSRP3
sCe6/0066	Cardiomyopathy, dilated type 1N	TCAP
sCe6/0067	Cardiomyopathy, dilated type 1O	ABCC9
sCe6/0068	Cardiomyopathy, dilated type 1P	PLN
sCe6/0069	Cardiomyopathy, dilated type 1R	ACTC1
sCe6/0070	Cardiomyopathy, dilated type 1S	MYH7
sCe6/0071	Cardiomyopathy, dilated type 1T	TMPO
sCe6/0072	Cardiomyopathy, dilated type 1U	PSEN1
sCe6/0073	Cardiomyopathy, dilated type 1V	PSEN2
sCe6/0074	Cardiomyopathy, dilated type 1W	VCL
sCe6/0075	Cardiomyopathy, dilated type 1X	FKTN
sCe6/0076	Cardiomyopathy, dilated type 1Y	TPM1
sCe6/0077	Cardiomyopathy, dilated type 1Z	TNNC1
sCe6/0078	Cardiomyopathy, dilated type 2A	TNNI3
sCe6/0079	Cardiomyopathy, dilated type 2B	GATAD1
sCe6/0080	Cardiomyopathy, dilated type 3B	DMD
sCe6/0081	Cardiomyopathy, dilated with ataxia	DNAJC19

sCe6/0082	Cardiomyopathy, dilated with hypergonadotropic hypogonadism	LMNA
sCe6/0083	Cardiomyopathy, dilated with woolly hair and keratoderma	DSP
sCe6/0084	Cardiomyopathy, familial hypertrophic	CAV3
sCe6/0085	Cardiomyopathy, familial hypertrophic type 1	MYH7
sCe6/0086	Cardiomyopathy, familial hypertrophic type 10	MYL2
sCe6/0087	Cardiomyopathy, familial hypertrophic type 11	ACTC1
sCe6/0088	Cardiomyopathy, familial hypertrophic type 12	CSRP3
sCe6/0089	Cardiomyopathy, familial hypertrophic type 16	MYOZ2
sCe6/0090	Cardiomyopathy, familial hypertrophic type 17	JPH2
sCe6/0091	Cardiomyopathy, familial hypertrophic type 19	CALR3
sCe6/0092	Cardiomyopathy, familial hypertrophic type 2	TNNT2
sCe6/0093	Cardiomyopathy, familial hypertrophic type 3	TPM1
sCe6/0094	Cardiomyopathy, familial hypertrophic type 4	MYBPC3
sCe6/0095	Cardiomyopathy, familial hypertrophic type 6	PRKAG2
sCe6/0096	Cardiomyopathy, familial hypertrophic type 7	TNNI3
sCe6/0097	Cardiomyopathy, familial hypertrophic type 8	MYL3
sCe6/0098	Cardiomyopathy, familial hypertrophic type 9	TTN
sCe6/0099	Cardiomyopathy, familial restrictive type 1	TNNI3
sCe6/0100	Cardiomyopathy, fatal, MT-TI related	MT-TI
sCe6/0101	Cardiomyopathy, hypertrophic, midventricular, digenic	MYLK2
sCe6/0102	Cardiomyopathy, hypertrophic, MT-TG related	MT-TG
sCe6/0103	Cardiomyopathy, hypertrophic, type 18	PLN
sCe6/0104	Cardiomyopathy, idiopathic dilated, mitochondrial, MT-TH related	MT-TH
sCe6/0105	Cardiomyopathy, infantile hypertrophic, MT-ATP8 related	MT-ATP8
sCe6/0106	Cardiomyopathy, left ventricular noncompaction, MYH7B related	MYH7B
sCe6/0107	Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B
sCe6/0108	Central hypoventilation syndrome, congenital	ASCL1
sCe6/0109	Congenital heart defects multiple types	TAB2
sCe6/0110	Coronary heart disease, susceptibility to, type 6	MMP3

sCe6/0111	Danon disease	LAMP2
sCe6/0112	Diaphragmatic hernia type 3	ZFPM2
sCe6/0113	Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	DSP
sCe6/0114	Dopamine beta-hydroxylase (DBH) deficiency	DBH
sCe6/0115	Familial atrial fibrillation type 13	SCN1B
sCe6/0116	Heart block, progressive, familial, type 1A	SCN5A
sCe6/0117	Heart-hand syndrome, Slovenian type	LMNA
sCe6/0118	Jervell and Lange-Nielsen syndrome type 1	KCNQ1
sCe6/0119	Jervell and Lange-Nielsen syndrome type 2	KCNE1
sCe6/0120	Long QT syndrome type 1	KCNQ1
sCe6/0121	Long QT syndrome type 10	SCN4B
sCe6/0122	Long QT syndrome type 11	AKAP9
sCe6/0123	Long QT syndrome type 12	SNTA1
sCe6/0124	Long QT syndrome type 13	KCNJ5
sCe6/0125	Long QT syndrome type 15	CALM2
sCe6/0126	Long QT syndrome type 2	KCNH2
sCe6/0127	Long QT syndrome type 3	SCN5A
sCe6/0128	Long QT syndrome type 4	ANK2
sCe6/0129	Long QT syndrome type 5	KCNE1
sCe6/0130	Long QT syndrome type 6	KCNE2
sCe6/0131	Long QT syndrome type 8	CACNA1C
sCe6/0132	Long QT syndrome type 9	CAV3
sCe6/0133	Marfan syndrome	FBN1
sCe6/0134	MASS syndrome	FBN1
sCe6/0135	McKusick-Kaufman syndrome	MKKS
sCe6/0136	MELAS syndrome	MT-TF
sCe6/0137	MELAS syndrome	MT-TC
sCe6/0138	MELAS syndrome, MT-TL1 related	MT-TL1
sCe6/0139	MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1

sCe6/0140	MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2
sCe6/0141	Mitochondrial myopathy and sideroblastic anemia type 1	PUS1
sCe6/0142	Mitochondrial myopathy, infantile, transient, MT-TE related	MT-TE
sCe6/0143	Mitochondrial myopathy, isolated	MT-TD
sCe6/0144	Mitochondrial myopathy, MT-TA related	MT-TA
sCe6/0145	Mitochondrial myopathy, MT-TM related	MT-TM
sCe6/0146	Moyamoya disease type 5	ACTA2
sCe6/0147	Multisystemic smooth muscle dysfunction syndrome	ACTA2
sCe6/0148	Myopathy, MT-TQ related	MT-TQ
sCe6/0149	Myopathy, tubular aggregate, type 2	ORAI1
sCe6/0150	Pancreatic agenesis and congenital heart defects	GATA6
sCe6/0151	Progressive familial heart block	TRPM4
sCe6/0152	Pulmonary fibrosis, idiopathic	SFTPA1
sCe6/0153	Pulmonary newborn hypertension	CRHR1
sCe6/0154	Pulmonary venoocclusive disease type 2	EIF2AK4
sCe6/0155	Sengers syndrome	AGK
sCe6/0156	Short QT syndrome type 1	KCNH2
sCe6/0157	Short QT syndrome type 2	KCNQ1
sCe6/0158	Short QT syndrome type 3	KCNJ2
sCe6/0159	Sick sinus syndrome type 1	SCN5A
sCe6/0160	Sick sinus syndrome type 3	MYH6
sCe6/0161	Sinoatrial node dysfunction and deafness	CACNA1D
sCe6/0162	Sudden infant death syndrome, susceptibility to	SCN5A
sCe6/0163	Sudden infant death with dysgenesis of the testes syndrome	TSPYL1
sCe6/0164	Testicular anomalies with or without congenital heart disease	GATA4
sCe6/0165	Tetralogy of Fallot	ZFPM2
sCe6/0166	Tetralogy of Fallot	ALDH1A2
sCe6/0167	Tetralogy of Fallot	GATA6
sCe6/0168	Tetralogy of Fallot	GATA4

sCe6/0169	Thoracic aortic aneurysm dissection	SMAD2
sCe6/0170	Transposition of the great arteries, dextro-looped 1	MED13L
sCe6/0171	Ventricular fibrillation, paroxysmal familial type 1	SCN5A
sCe6/0172	Ventricular septal defect type 1	GATA4
sCe6/0173	Ventricular septal defect type 2	CITED2
sCe6/0174	Ventricular tachycardia, catecholaminergic polymorphic type 1	RYR2
sCe6/0175	Ventricular tachycardia, catecholaminergic polymorphic type 2	CASQ2
sCe6/0176	Ventricular tachycardia, catecholaminergic polymorphic type 4	CALM1
sCe6/0177	Ventricular tachycardia, catecholaminergic polymorphic type 5	TRDN
sCe6/0178	Wolff -Parkinson-White syndrome	PRKAG2
sCe7/0001	Angioedema, hereditary	SERPING1
sCe7/0002	Antithrombin III deficiency	SERPINC1
sCe7/0003	Aortic aneurysm, familial thoracic type 3	TGFBR2
sCe7/0004	Aortic aneurysm, familial thoracic type 4	MYH11
sCe7/0005	Aortic aneurysm, familial thoracic type 5	TGFBR1
sCe7/0006	Aortic aneurysm, familial thoracic type 6	ACTA2
sCe7/0007	Aortic aneurysm, familial thoracic type 7	MYLK
sCe7/0008	Aortic aneurysm, familial thoracic type 8	PRKG1
sCe7/0009	Aortic aneurysm, familial thoracic, MAT2A related	MAT2A
sCe7/0010	Aortic valve disease type 1	NOTCH1
sCe7/0011	Aortic valve disease type 2	SMAD6
sCe7/0012	Arterial calcification type 1, generalized, infantile	ENPP1
sCe7/0013	Arterial calcification type 2, generalized, infantile	ABCC6
sCe7/0014	Arterial Tortuosity Syndrome	SLC2A10
sCe7/0015	Atherosclerosis, SOAT1 related	SOAT1
sCe7/0016	Bernard Soulier syndrome type A1	GP1BA
sCe7/0017	Bernard Soulier syndrome type A2	GP1BA
sCe7/0018	Bernard Soulier syndrome type B	GP1BB
sCe7/0019	Bernard Soulier syndrome type C	GP9

sCe7/0020	CADASIL	NOTCH3
sCe7/0021	Capillary malformation-arteriovenous malformation	RASA1
sCe7/0022	CARASIL	HTRA1
sCe7/0023	Carotid intimal medial thickness type 1	PPARG
sCe7/0024	Cerebral cavernous malformations type 1	KRIT1
sCe7/0025	Cerebral cavernous malformations type 2	CCM2
sCe7/0026	Cerebral cavernous malformations type 3	PDCD10
sCe7/0027	Coarctation of the aorta	MCTP2
sCe7/0028	Coronary artery disease in familial hypercholesterolemia, protection against	ABCA1
sCe7/0029	Fabry disease	GLA
sCe7/0030	Factor II deficiency	F2
sCe7/0031	Factor VII deficiency	F7
sCe7/0032	Glycoprotein Ia C807T polymorphism	ITGA2
sCe7/0033	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	JAM3
sCe7/0034	Homocystinuria	MTHFR
sCe7/0035	Homocystinuria due to cystathionine beta-synthase deficiency	CBS
sCe7/0036	Homocystinuria-megaloblastic anemia, cbl E type	MTRR
sCe7/0037	Hypertension early onset	NR3C2
sCe7/0038	Hypertension, ADD2 related	ADD2
sCe7/0039	Hypertension, salt-sensitive essential, susceptibility to	CYP3A5
sCe7/0040	Loeys-Dietz syndrome type 4	TGFB2
sCe7/0041	Moyamoya disease type 2, susceptibility to	RNF213
sCe7/0042	Moyamoya type 6 with achalasia	GUCY1A3
sCe7/0043	Myoglobinuria acute recurrent	LPIN1
sCe7/0044	Myopathy with lactic acidosis hereditary	ISCU
sCe7/0045	Orthostatic intolerance	SLC6A2
sCe7/0046	Parkes Weber syndrome	RASA1
sCe7/0047	Plasminogen activator inhibitor type 1	SERPINE1
sCe7/0048	Polyarteritis nodosa, childhood-onset	CECR1

sCe7/0049	Protein C Deficiency, AD	PROC
sCe7/0050	Protein S Deficiency, autosomal dominant	PROS1
sCe7/0051	Pseudohypoaldosteronism type 2D	KLHL3
sCe7/0052	Pseudohypoaldosteronism type 2E	CUL3
sCe7/0053	Pulmonary hypertension, primary type	BMPR2
sCe7/0054	Pulmonary venoocclusive disease type 1	BMPR2
sCe7/0055	Sneddon syndrome	CECR1
sCe7/0056	Stormorken syndrome	STIM1
sCe7/0057	Supravalvar aortic stenosis	ELN
sCe7/0058	Thrombophilia due to thrombin defect	F2
sCe7/0059	Transposition of the great arteries, dextro-looped 1	MED13L
sCe7/0060	Vascular system defects due to CALCRL deficiency	CALCRL
sCe7/0061	Vascular system defects due to GNA13 deficiency	GNA13

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe6/0001	Arrhythmia, hereditary panel	25
pCe6/0002	Arrhythmogenic right ventricular cardiomyopathy panel	25
pCe6/0003	Brugada syndrome panel	25
pCe6/0004	Cardiomyopathy dilated panel	25
pCe6/0005	Cardiomyopathy hypertrophic panel	25
pCe6/0006	Catecholaminergic polymorphic ventricular tachycardia panel	25
pCe6/0007	Comprehensive pulmonary disease panel	25
pCe6/0008	Congenital heart defects panel	25
pCe6/0009	Long QT syndrome panel	25
pCe6/0010	Pulmonary hypertension panel	25
pCe7/0001	Dolichoectasia panel	25
pCe7/0002	Familial thoracic aortic aneurysm panel	25
pCe7/0003	Hereditary hemorrhagic telangiectasia panel	25