

Gene	OMIM	phenotype	AD-AR-XL	ACMG list	Date added
1 ACTC1	<a href="#">ACTC1 (MIM 102540)</a>	<a href="#">Familial hypertrophic cardiomyopathy 11 (MIM 612098)</a>	AD	Y	22/06/2023
2 ACTA2	<a href="#">ACTA2 (MIM 102620)</a>	<a href="#">Aortic aneurysm, familial thoracic 6 (MIM 611788)</a>	AD	Y	
3 ACVRL1	<a href="#">ACVRL1 (MIM 601284)</a>	<a href="#">Hereditary hemorrhagic telangiectasia type 2 (MIM 600376)</a>	AD	Y	
4 APC	<a href="#">APC (MIM 611731)</a>	<a href="#">Adenomatous polyposis coli (MIM 175100)</a>	AD	Y	
5 APOB	<a href="#">APOB (MIM 107730)</a>	<a href="#">Familial hypercholesterolemia 2 (MIM 144010)</a>	AD	Y	
6 ATP7B	<a href="#">ATP7B (MIM 606882)</a>	<a href="#">Wilson disease (MIM 277900)</a>	AR	Y	
7 BAG3	<a href="#">BAG3 (MIM 603883)</a>	<a href="#">Dilated cardiomyopathy 1HH (MIM 613881)</a>	AD	Y	
7		<a href="#">Myofibrillar myopathy 6 (MIM 612954)</a>	AD	Y	
8 BMPR1A	<a href="#">BMPR1A (MIM 601299)</a>	<a href="#">Juvenile polyposis syndrome (MIM 174900)</a>	AD	Y	
9 BRCA1	<a href="#">BRCA1 (MIM 113705)</a>	<a href="#">Breast-ovarian cancer, familial 1 (MIM 604370)</a>	AD	Y	
10 BRCA2	<a href="#">BRCA2 (MIM 600185)</a>	<a href="#">Breast-ovarian cancer, familial 2 (MIM 612555)</a>	AD	Y	
11 BTD	<a href="#">BTD (MIM 609019)</a>	<a href="#">Biotinidase deficiency (MIM 253260)</a>	AR	Y	
12 CACNA1S	<a href="#">CACNA1S (MIM 114208)</a>	<a href="#">Malignant hyperthermia (MIM 601887)</a>	AD	Y	
13 CALM1	<a href="#">CALM1 (MIM 114180)</a>	<a href="#">Long QT syndrome 14 (MIM 616247)</a>	AD	Y	
13		<a href="#">Ventricular tachycardia, catecholaminergic polymorphic (MIM 601419)</a>	AD	Y	
14 CALM2	<a href="#">CALM2 (MIM 114182)</a>	<a href="#">Long QT syndrome 15 (MIM 616249)</a>	AD	Y	
15 CALM3	<a href="#">CALM3 (MIM 114183)</a>	<a href="#">Long QT syndrome 16 (MIM 618782)</a>	AD	Y	
16 CASQ2	<a href="#">CASQ2 (MIM 114251)</a>	<a href="#">Catecholaminergic polymorphic ventricular tachycardia 2 (MIM 611938)</a>	AR	Y	
17 COL3A1	<a href="#">COL3A1 (MIM 120180)</a>	<a href="#">Ehlers-Danlos syndrome, type 4 (MIM 130050)</a>	AD	Y	
18 DES	<a href="#">DES (MIM 125660)</a>	<a href="#">Dilated cardiomyopathy 1I (MIM 604765)</a>	AD	Y	
18		<a href="#">Myofibrillar myopathy 1 (MIM 601419)</a>	AD	Y	
19 DSC2	<a href="#">DSC2 (MIM 125645)</a>	<a href="#">Arrhythmogenic right ventricular cardiomyopathy, type 11 (MIM 610476)</a>	AD	Y	
19		<a href="#">Arrhythmogenic right ventricular cardiomyopathy, type 10 (MIM 610193)</a>	AD	Y	
20 DSG2	<a href="#">DSG2 (MIM 125671)</a>	<a href="#">Arrhythmogenic right ventricular dysplasia 10 (MIM 610193)</a>	AD	Y	
20		<a href="#">Cardiomyopathy, dilated, 1BB (MIM 612877)</a>	AR	Y	
21 DSP	<a href="#">DSP (MIM 125647)</a>	<a href="#">Arrhythmogenic right ventricular cardiomyopathy, type 8 (MIM 607450)</a>	AD	Y	
21		<a href="#">Dilated cardiomyopathy with woolly hair, palmoplantar keratoderma, and tooth agenesis (MIM 615821)</a>	AD	Y	
22 ENG	<a href="#">ENG (MIM 131195)</a>	<a href="#">Hereditary hemorrhagic telangiectasia type 1 (MIM 187300)</a>	AD	Y	
23 FBN1	<a href="#">FBN1 (MIM 134797)</a>	<a href="#">Marfan's syndrome (MIM 154700)</a>	AD	Y	
24 FLNC	<a href="#">FLNC (MIM 102565)</a>	<a href="#">Dilated cardiomyopathy (MIM 617047)</a>	AD	Y	
24		<a href="#">Myofibrillar myopathy 5 (MIM 609524)</a>	AD	Y	
25 GAA	<a href="#">GAA (MIM 606800)</a>	<a href="#">Pompe disease (MIM 232300)</a>	AR	Y	
26 GLA	<a href="#">GLA (MIM 300644)</a>	<a href="#">Fabry's disease (MIM 301500)</a>	XL	Y	
27 HFE	<a href="#">HFE (MIM 613609)</a>	<a href="#">Hereditary hemochromatosis (c.845G&gt;A; p.C282Y homozygotes only) (MIM 235200)</a>	AR	Y	
28 HNF1A	<a href="#">HNF1A (MIM 142410)</a>	<a href="#">Maturity-Onset of Diabetes of the Young (MIM 600496)</a>	AD	Y	
29 KCNH2	<a href="#">KCNH2 (MIM 152427)</a>	<a href="#">Long QT syndrome 2 (MIM 613688)</a>	AD	Y	
30 KCNQ1	<a href="#">KCNQ1 (MIM 607542)</a>	<a href="#">Long QT syndrome 1 (MIM 192500)</a>	AD	Y	
31 LDLR	<a href="#">LDLR (MIM 606945)</a>	<a href="#">Familial hypercholesterolemia 1 (MIM 143890)</a>	AD	Y	
32 LMNA	<a href="#">LMNA (MIM 150330)</a>	<a href="#">Dilated cardiomyopathy 1A (MIM 115200)</a>	AD	Y	
33 MAX	<a href="#">MAX (MIM 154950)</a>	<a href="#">Pheochromocytoma (MIM 171300)</a>	AD	Y	
34 MEN1	<a href="#">MEN1 (MIM 613733)</a>	<a href="#">Multiple endocrine neoplasia, type 1 (MIM 131100)</a>	AD	Y	
35 MLH1	<a href="#">MLH1 (MIM 120436)</a>	<a href="#">Lynch syndrome 2 (MIM 609310)</a>	AD	Y	
36 MSH2	<a href="#">MSH2 (MIM 609309)</a>	<a href="#">Lynch syndrome 1 (MIM 120435)</a>	AD	Y	
37 MSH6	<a href="#">MSH6 (MIM 600678)</a>	<a href="#">Lynch syndrome 5 (MIM 614350)</a>	AD	Y	
38 MUTYH	<a href="#">MUTYH (MIM 604933)</a>	<a href="#">MYH-associated polyposis (MIM 608456)</a>	AR	Y	
39 MYBPC3	<a href="#">MYBPC3 (MIM 600958)</a>	<a href="#">Familial hypertrophic cardiomyopathy 4 (MIM 115197)</a>	AD	Y	
40 MYH11	<a href="#">MYH11 (MIM 160745)</a>	<a href="#">Aortic aneurysm, familial thoracic 4 (MIM 132900)</a>	AD	Y	
41 MYH7	<a href="#">MYH7 (MIM 160760)</a>	<a href="#">Dilated cardiomyopathy 1S (MIM 613426)</a>	AD	Y	
42 MYH7	<a href="#">MYH7 (MIM 160760)</a>	<a href="#">Familial hypertrophic cardiomyopathy 1 (MIM 192600)</a>	AD	Y	
43 MYL2	<a href="#">MYL2 (MIM 160781)</a>	<a href="#">Familial hypertrophic cardiomyopathy 10 (MIM 608758)</a>	AD	Y	
44 MYL3	<a href="#">MYL3 (MIM 160790)</a>	<a href="#">Familial hypertrophic cardiomyopathy 8 (MIM 608751)</a>	AD	Y	
45 NF2	<a href="#">NF2 (MIM 607379)</a>	<a href="#">Neurofibromatosis, type 2 (MIM 101000)</a>	AD	Y	
46 OTC	<a href="#">OTC (MIM 300461)</a>	<a href="#">Ornithine carbamoyltransferase deficiency (MIM 311250)</a>	XL	Y	
47 PALB2	<a href="#">PALB2 (MIM 610355)</a>	<a href="#">Hereditary breast cancer (MIM 114480)</a>	AD	Y	
48 PCSK9	<a href="#">PCSK9 (MIM 607786)</a>	<a href="#">Hypercholesterolemia, autosomal dominant, 3 (MIM 603776)</a>	AD	Y	
49 PKP2	<a href="#">PKP2 (MIM 602861)</a>	<a href="#">Arrhythmogenic right ventricular cardiomyopathy, type 9 (MIM 609040)</a>	AD	Y	
50 PMS2	<a href="#">PMS2 (MIM 600259)</a>	<a href="#">Lynch syndrome 4 (MIM 614337)</a>	AD	Y	
51 PRKAG2	<a href="#">PRKAG2 (MIM 602743)</a>	<a href="#">Familial hypertrophic cardiomyopathy 6 (MIM 600858)</a>	AD	Y	

52	<b>PTEN</b>	<b>PTEN (MIM 601728)</b>	<a href="#">PTEN hamartoma tumor syndrome (MIM 158350)</a>	AD	Y
53	RB1	<a href="#">RB1 (MIM 614041)</a>	<a href="#">Retinoblastoma (MIM 180200)</a>	AD	Y
54	RBM20	<a href="#">RBM20 (MIM 613171)</a>	<a href="#">Dilated cardiomyopathy 1DD (MIM 613172)</a>	AD	Y
55	RET	<a href="#">RET (MIM 164761)</a>	<a href="#">Familial medullary thyroid carcinoma (MIM 155240)</a>	AD	Y
56	RET	<a href="#">RET (MIM 164761)</a>	<a href="#">Multiple endocrine neoplasia, type 2a (MIM 171400)</a>	AD	Y
57	RET	<a href="#">RET (MIM 164761)</a>	<a href="#">Multiple endocrine neoplasia, type 2b (MIM 162300)</a>	AD	Y
58	<b>RPE65</b>	<b>RPE65 (MIM 180069)</b>	RPE65-related retinopathy (MIM 204100, MIM 613794)	AR	Y
59	<b>RYR1</b>	<b>RYR1 (MIM 180901)</b>	<a href="#">Malignant hyperthermia (MIM 145600)</a>	AD	Y
60	RYR2	<a href="#">RYR2 (MIM 180902)</a>	<a href="#">Catecholaminergic polymorphic ventricular tachycardia 1 (MIM 604772)</a>		Y
61	SCN5A	<a href="#">SCN5A (MIM 600163)</a>	<a href="#">Brugada syndrome 1 (MIM 601144)</a>		Y
62	SCN5A	<a href="#">SCN5A (MIM 600163)</a>	<a href="#">Dilated cardiomyopathy 1E (MIM 601154)</a>		Y
63	SCN5A	<b>SCN5A (MIM 600163)</b>	<a href="#">Long QT syndrome 3 (MIM 603830)</a>	AD	Y
64	SDHAF2	<a href="#">SDHAF2 (MIM 613019)</a>	<a href="#">Paragangliomas 2 (MIM 601650)</a>	AD	Y
65	SDHB	<a href="#">SDHB (MIM 185470)</a>	Hereditary paraganglioma-pheochromocytoma syndrome (MIM 115310, MIM 171300)	AD	Y
66	SDHC	<a href="#">SDHC (MIM 602413)</a>	<a href="#">Paragangliomas 3 (MIM 605373)</a>	AD	Y
67	<b>SDHD</b>	<b>SDHD (MIM 602690)</b>	Hereditary paraganglioma-pheochromocytoma syndrome (MIM 168000, MIM 171300)	AD	Y
68	SMAD3	<a href="#">SMAD3 (MIM 603109)</a>	<a href="#">Loeys-Dietz syndrome type 3 (MIM 613795)</a>	AD	Y
69	SMAD4	<a href="#">SMAD4 (MIM 600993)</a>	<a href="#">Juvenile polyposis syndrome (MIM 174900)</a>	AD	Y
70	SMAD4	<a href="#">SMAD4 (MIM 600993)</a>	<a href="#">Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (MIM 175050)</a>	AD	Y
71	STK11	<a href="#">STK11 (MIM 602216)</a>	<a href="#">Peutz-Jeghers syndrome (MIM 175200)</a>	AD	Y
72	TGFBR1	<a href="#">TGFBR1 (MIM 190181)</a>	<a href="#">Loeys-Dietz syndrome type 1A (MIM 609192)</a>	AD	Y
73	TGFBR2	<a href="#">TGFBR2 (MIM 190182)</a>	<a href="#">Loeys-Dietz syndrome type 1B (MIM 610168)</a>	AD	Y
74	TMEM127	<a href="#">TMEM127 (MIM 613403)</a>	<a href="#">Pheochromocytoma (MIM 171300)</a>	AD	Y
75	TMEM43	<a href="#">TMEM43 (MIM 612048)</a>	<a href="#">Arrhythmogenic right ventricular cardiomyopathy, type 5 (MIM 604400)</a>	AD	Y
76	TNNC1	<a href="#">TNNC1 (MIM 191040)</a>	<a href="#">Dilated cardiomyopathy 1Z (MIM 611879)</a>	AD	Y
77	<b>TNNI3</b>	<b>TNNI3 (MIM 191044)</b>	<a href="#">Familial hypertrophic cardiomyopathy 7 (MIM 613690)</a>	AD	Y
78	TNNT2	<a href="#">TNNT2 (MIM 191045)</a>	<a href="#">Dilated cardiomyopathy 1D (MIM 601494)</a>	AD	Y
79	TNNT2	<a href="#">TNNT2 (MIM 191045)</a>	<a href="#">Familial hypertrophic cardiomyopathy 2 (MIM 115195)</a>	AD	Y
80	TP53	<a href="#">TP53 (MIM 191170)</a>	<a href="#">Li-Fraumeni syndrome 1 (MIM 151623)</a>	AD	Y
81	TPM1	<a href="#">TPM1 (MIM 191010)</a>	<a href="#">Familial hypertrophic cardiomyopathy 3 (MIM 115196)</a>	AD	Y
82	<b>TRDN</b>	<b>TRDN (MIM 603283)</b>	<a href="#">Catecholaminergic polymorphic ventricular tachycardia 5 (MIM 615441)</a>	AR	Y
83	TRDN	<a href="#">TRDN (MIM 603283)</a>	Long QT syndrome		Y
84	TSC1	<a href="#">TSC1 (MIM 605284)</a>	<a href="#">Tuberous sclerosis 1 (MIM 191100)</a>	AD	Y
85	TSC2	<a href="#">TSC2 (MIM 191092)</a>	<a href="#">Tuberous sclerosis 2 (MIM 613254)</a>	AD	Y
86	<b>TTN</b>	<b>TTN (MIM 188840)</b>	<a href="#">Dilated cardiomyopathy (truncating variants only) (MIM 604145)</a>	AD	Y
87	TTR	<a href="#">TTR (MIM 176300)</a>	<a href="#">Hereditary transthyretin-related amyloidosis (MIM 105210)</a>	AD	Y
88	<b>VHL</b>	<b>VHL (MIM 608537)</b>	<a href="#">Von Hippel-Lindau syndrome (MIM 193300)</a>	AD	Y
89	WT1	<a href="#">WT1 (MIM 607102)</a>	<a href="#">Wilms' tumor (MIM 194070)</a>	AD	Y