

GENE	CONDITION(S)	REFERENCE(S)
APC	Familial adenomatous polyposis (FAP) and attenuated FAP	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ Jasperson KW, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1998 Dec 18, updated 2017 Feb 2. (PMID: 20301519)
ATM	ATM-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018²
AXIN2	Oligodontia-colorectal cancer syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ Bergendal B, et al. <i>Am J Med Genet A</i> . 2011;155A(7):1616-22. (PMID: 21626677) Lammi L, et al. <i>Am J Hum Genet</i> . 2004;74(5):1043-50. (PMID: 15042511) Marvin ML, et al. <i>Am J Med Genet A</i> . 2011;155A(4):898-902. (PMID: 21416598)
BAP1	BAP1 tumor predisposition syndrome	Battaglia A. <i>Clin Med Insights Oncol</i> . 2014;8:37-47. (PMID: 24855403) Rai K, et al. <i>Clin Genet</i> . 2016;89(3):285-94. (PMID: 26096145)
BARD1	BARD1-related cancers	Ratajska M, et al. <i>Breast Cancer Res Treat</i> . 2012;131(1):89-97. (PMID: 21344236)
BMPR1A	Juvenile polyposis syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ Larsen Haidle J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 May 13, updated 2017 May 9. (PMID: 20301642)
BRCA1	Hereditary breast and ovarian cancer syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² Petrucci N, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1998 Sep 4, updated 2016 Dec 15. (PMID: 20301425)
BRCA2	Hereditary breast and ovarian cancer syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² Petrucci N, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1998 Sep 4, updated 2016 Dec 15. (PMID: 20301425)
BRIP1	BRIP1-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018²
CDC73	Hyperparathyroidism-jaw tumor syndrome Familial isolated hyperparathyroidism Parathyroid carcinoma	Jackson MA, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Dec 31, updated 2015 Jan 15. (PMID: 20301744) Li Y, et al. <i>Endocr Relat Cancer</i> . 2015;23(6):R229-47. (PMID: 27207564)
CDH1	Hereditary diffuse gastric cancer syndrome	NCCN®: Gastric Cancer V.5.2017³ NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² Kaurah P, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Nov 4, updated 2014 Jul 31. (PMID: 20301318) van der Post RS, et al. <i>J Med Genet</i> . 2015;52(6):361-74. (PMID: 25979631)
CDK4	Hereditary cutaneous melanoma	Aoude LG, et al. <i>Pigment Cell Melanoma Res</i> . 2015;28(2):148-60. (PMID: 25431349) Potrony M, et al. <i>Ann Transl Med</i> . 2015;3(15):210. (PMID: 26488006) Soura E, et al. <i>J Am Acad Dermatol</i> . 2016;74(3):395-407. (PMID: 26892650)
CDKN2A	Hereditary melanoma-pancreatic cancer syndrome	NCCN®: Pancreatic Adenocarcinoma V.3.2017⁷ Aoude LG, et al. <i>Pigment Cell Melanoma Res</i> . 2015;28(2):148-60. (PMID: 25431349) Canto MI, et al. <i>Gut</i> . 2013;62(3):339-47. (PMID: 23135763) Potrony M, et al. <i>Ann Transl Med</i> . 2015;3(15):210. (PMID: 26488006) Soura E, et al. <i>J Am Acad Dermatol</i> . 2016;74(3):395-407. (PMID: 26892650)
CHEK2	CHEK2-related cancers including breast, colon and other sites	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹
DICER1	DICER1-related pleuropulmonary blastoma familial tumor predisposition syndrome	International Pleuropulmonary Blastoma Registry Recommendations for Surveillance of High-Risk Children and Children with PPB www.ppbregistry.org/health-professionals/recommendations/surveillance
EPCAM	Lynch syndrome (hereditary nonpolyposis colorectal cancer syndrome)	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ Kohlmann W, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Feb 5, updated 2014 May 22. (PMID: 20301390)
FH	Hereditary leiomyomatosis and renal cell cancer	Menko FH, et al. <i>Fam Cancer</i> . 2014;13(4):637-44. (PMID: 25012257) Pithukpakorn M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Jul 31, updated 2015 Aug 6. (PMID: 20301430)
FLCN	Birt-Hogg-Dubé syndrome	Gupta N, et al. <i>Clin Chest Med</i> . 2016;37(3):475-86. (PMID: 27514594) Menko FH, et al. <i>Lancet Oncol</i> . 2009;10(12):1199-206. (PMID: 19959076) Toro JR. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Feb 27, updated 2014 Aug 7. (PMID: 20301695)
GREM1	Hereditary mixed polyposis syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹
HOXB13	Prostate cancer	Cai Q, et al. <i>Oncotarget</i> . 2015;6(39):42312-21. (PMID: 26517352)
KIT	Gastrointestinal stromal tumors	NCCN®: Soft Tissue Sarcoma V.1.2018⁴ ESMO. <i>Ann Oncol</i> . 2014;25 Suppl 3:iii21-6. (PMID: 25210085)

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MAX	Hereditary paraganglioma-pheochromocytoma syndrome	Favier J, et al. <i>Nat Rev Endocrinol</i> . 2015 Feb;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle (WA): University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol</i> . 2014 Feb;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab</i> . 2014;99(6):1915-42. (PMID: 24893135)
MEN1	Multiple endocrine neoplasia type 1	NCCN®: Neuroendocrine Tumors V.3.2017 ⁵ Giusti F, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Aug 31, updated 2015 Feb 12. (PMID: 20301710) Thakker RV, et al. <i>J Clin Endocrinol Metab</i> . 2012;97(9):2990-3011. (PMID: 22723327)
MET	Hereditary papillary renal cell carcinoma	Mustafa S, et al. <i>J Community Hosp Intern Med Perspect</i> . 2012;1(4). (PMID: 23882344)
MITF	Hereditary cutaneous malignant melanoma	Potrony M, et al. <i>Ann Transl Med</i> . 2015;3(15):210. (PMID: 26488006) Potrony M, et al. <i>JAMA Dermatol</i> . 2016;152(4):405-12. (PMID: 26650189) Soura E, et al. <i>J Am Acad Dermatol</i> . 2016;74(3):395-407. (PMID: 26892650) Udayakumar D, et al. <i>Hematol Oncol Clin North Am</i> . 2009;23(3):415-29, vii. (PMID: 19464594)
MLH1	Lynch syndrome (hereditary nonpolyposis colorectal cancer syndrome)	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Kohlmann W, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Feb 5, updated 2014 May 22. (PMID: 20301390)
MSH2	Lynch syndrome (hereditary nonpolyposis colorectal cancer syndrome)	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Kohlmann W, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Feb 5, updated 2014 May 22. (PMID: 20301390)
MSH3	MSH3-associated polyposis	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Adam R, et al. <i>Am J Hum Genet</i> . 2016;99(2):337-51. (PMID: 27476653)
MSH6	Lynch syndrome (hereditary nonpolyposis colorectal cancer syndrome)	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Kohlmann W, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Feb 5, updated 2014 May 22. (PMID: 20301390)
MUTYH	MUTYH-associated polyposis	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Nielsen M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2012 Oct 4, updated 2015 Sep 24. (PMID: 23035301)
NBN	NBN-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ²
NF1	Neurofibromatosis type 1 Neurofibromatosis-Noonan syndrome Watson syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ² Ben-Shachar S, et al. <i>Eur J Hum Genet</i> . 2013;21(5):535-9. PMID: 23047742 De Luca A, et al. <i>Am J Hum Genet</i> . 2005;77(6):1092-101. PMID: 16380919 Ekvall S, et al. <i>Am J Med Genet A</i> . 2014;164A(3):579-87. PMID: 24357598 Hersh JH, et al. <i>Pediatrics</i> . 2008;121(3):633-42. PMID: 18310216 Steward DR, et al. <i>Genet Med</i> . 2018;20(7):671-682. PMID: 30006586
NF2	Neurofibromatosis type 2	Blakeley JO, et al. <i>Am J Med Genet A</i> . 2012;158A(1):24-41. (PMID: 22140088) Evans DG, et al. <i>Br J Neurosurg</i> . 2005;19(1):5-12. (PMID: 16147576) Evans DG. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1998 Oct 14, updated 2011 Aug 18. (PMID: 20301380) Hoa M, et al. <i>Otolaryngol Clin North Am</i> . 2012;45(2):315-32, viii. (PMID: 22483819)
NTHL1	NTHL1-associated polyposis	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Belhadj S, et al. <i>Clin Gastroenterol Hepatol</i> . 2017;15(3):461-462. (PMID: 27720914) Kuiper RP, et al. <i>Oncotarget</i> . 2015;6(33):34069-70. (PMID: 26431160) Rivera B, et al. <i>N Engl J Med</i> . 2015;373(20):1985-6. (PMID: 26559593) Weren RD, et al. <i>Nat Genet</i> . 2015 Jun;47(6):668-7. (PMID: 25938944)
PALB2	PALB2-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ²
PDGFRA	Gastrointestinal stromal tumors	NCCN®: Soft Tissue Sarcoma V.1.2018 ⁴ ESMO/European Sarcoma Network Working Group. <i>Ann Oncol</i> . 2014;25 Suppl 3:iii21-iii26. (PMID: 25210085)
PMS2	Lynch syndrome (hereditary nonpolyposis colorectal cancer syndrome)	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018 ² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ Kohlmann W, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Feb 5, updated 2014 May 22. (PMID: 20301390)
POLD1	POLD1-related cancers Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy	POLD1-related cancers: NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018 ¹ MDPL: Pelosini C, et al. <i>Metabolism</i> . 2014;63(11):1385-9. (PMID: 25131834)

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POLE	POLE-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹
PRKAR1A	Carney complex	Siordia JA. <i>J Card Surg.</i> 2015;30(7):560-7. (PMID: 25996461) Stratakis CA, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2003 Feb 5, updated 2015 Jan 29. (PMID: 20301463)
PTCH1	Basal cell nevus syndrome	Baliga SD, et al. <i>J Maxillofac Oral Surg.</i> 2010;9(1):82-6. (PMID: 23139577) Bree AF, et al. <i>Am J Med Genet A.</i> 2011;155A(9):2091-7. (PMID: 21834049) Evans DG, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2002 Jun 20, updated 2015 Oct 1. (PMID: 20301330)
PTEN	PTEN hamartoma tumor syndrome	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² Eng C. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2001 Nov 29, updated 2016 Jun 2. (PMID: 20301661)
RAD51C	RAD51C-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018²
RAD51D	RAD51D-related cancers	NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018²
RB1	Retinoblastoma	Abramson DH, et al. <i>JAMA Ophthalmol.</i> 2015;133(11):1341-7. (PMID: 26378747) Canadian Retinoblastoma Society. <i>Can J Ophthalmol.</i> 2009;44 Suppl 2:S1-88. (PMID: 20237571) Fletcher O, et al. <i>J Natl Cancer Inst.</i> 2004;96(5):357-63. (PMID: 14996857) Lohmann DR, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2000 Jul 18, updated 2015 Nov 19. (PMID: 20301625)
RET	Multiple endocrine neoplasia type 2	NCCN®: Neuroendocrine Tumors V.3.2017⁵ NCCN®: Thyroid Carcinoma V.2.2017⁶ Marguard J, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 1999 Sep 27, updated 2015 Jun 25. (PMID: 20301434) Wells SA Jr, et al. <i>Thyroid.</i> 2015;25(6):567-610. (PMID: 25810047)
SDHA	Hereditary paraganglioma-pheochromocytoma syndrome	Benn DE, et al. <i>Endocr Relat Cancer.</i> 2015;22(4):T91-103. (PMID: 26273102) Favier J, et al. <i>Nat Rev Endocrinol.</i> 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol.</i> 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab.</i> 2014;99(6):1915-42. (PMID: 24893135)
SDHAF2	Hereditary paraganglioma-pheochromocytoma syndrome	Benn DE, et al. <i>Endocr Relat Cancer.</i> 2015;22(4):T91-103. (PMID: 26273102) Favier J, et al. <i>Nat Rev Endocrinol.</i> 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol.</i> 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab.</i> 2014;99(6):1915-42. (PMID: 24893135)
SDHB	Hereditary paraganglioma-pheochromocytoma syndrome	Benn DE, et al. <i>Endocr Relat Cancer.</i> 2015;22(4):T91-103. (PMID: 26273102) Favier J, et al. <i>Nat Rev Endocrinol.</i> 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol.</i> 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab.</i> 2014;99(6):1915-42. (PMID: 24893135)
SDHC	Hereditary paraganglioma-pheochromocytoma syndrome	Benn DE, et al. <i>Endocr Relat Cancer.</i> 2015;22(4):T91-103. (PMID: 26273102) Favier J, et al. <i>Nat Rev Endocrinol.</i> 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol.</i> 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab.</i> 2014;99(6):1915-42. (PMID: 24893135)
SDHD	Hereditary paraganglioma-pheochromocytoma syndrome	Benn DE, et al. <i>Endocr Relat Cancer.</i> 2015;22(4):T91-103. (PMID: 26273102) Favier J, et al. <i>Nat Rev Endocrinol.</i> 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. <i>GeneReviews®</i> [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol.</i> 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab.</i> 2014;99(6):1915-42. (PMID: 24893135)

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SMAD4	Juvenile polyposis syndrome Hereditary hemorrhagic telangiectasia	<p>JPS: NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ Larsen Haidle J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 May 13, updated 2017 Mar 9. (PMID: 20301642) Syngal S, et al. <i>Am J Gastroenterol</i>. 2015;110(2):223-62; quiz 263. (PMID: 25645574)</p> <p>HHT: Faughnan ME, et al. <i>J Med Genet</i>. 2011;48(2):73-87. (PMID: 19553198) McDonald J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 26, updated 2017 Feb 2. (PMID: 20301525)</p>
SMARCA4	SMARCA4-related cancers	<p>Berchuck A, et al. <i>Gynecol Oncol Rep</i>. 2015;12:20-2. (PMID: 26076152) Moes-Sosnowska J, et al. <i>Orphanet J Rare Dis</i>. 2015;10:32. (PMID: 25886974) Sredni ST, et al. <i>Pediatr Dev Pathol</i>. 2015;18(1):49-58. (PMID: 25494491) Witkowski L, et al. <i>Nat Genet</i>. 2014;46(5):438-43. (PMID: 24658002)</p>
SMARCB1	Rhabdoid tumor predisposition syndrome Schwannomatosis	<p>RTPS: Teplick A, et al. <i>Eur J Pediatr</i>. 2011;170(3):285-94. (PMID: 21210147)</p> <p>Schwannomatosis: Blakeley JO, et al. <i>Neuro Oncol</i>. 2016;18(5):624-38. (PMID: 26851632) Koontz NA, et al. <i>AJR Am J Roentgenol</i>. 2013;200(6):W646-53. (PMID: 23701098) Reddy RG, et al. <i>BMJ Case Rep</i>. 2013;2013:bcr-2013-008843. (PMID: 23595180)</p>
STK11	Peutz-Jeghers syndrome	<p>NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.1.2018¹ McGarrity TJ, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2001 Feb 23, updated 2016 Jul 14. (PMID: 20301443)</p>
TMEM127	Hereditary paraganglioma-pheochromocytoma syndrome	<p>Favier J, et al. <i>Nat Rev Endocrinol</i>. 2015;11(2):101-11. (PMID: 25385035) Kirmani S, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 May 21, updated 2014 Nov 6. (PMID: 20301715) Lefebvre M, et al. <i>Curr Oncol</i>. 2014;21(1):e8-e17. (PMID: 24523625) Lenders JW, et al. <i>J Clin Endocrinol Metab</i>. 2014;99(6):1915-42. (PMID: 24893135)</p>
TP53	Li-Fraumeni syndrome	<p>NCCN®: Genetic/Familial High-Risk Assessment: Breast and Ovarian V.2.2018² Bougeard G, et al. <i>J Clin Oncol</i>. 2015;33(21):2345-52. (PMID: 26014290) Heymann S, et al. <i>Radiat Oncol</i>. 2010;5:104. (PMID: 21059199) Hisada M, et al. <i>J Natl Cancer Inst</i>. 1998;90(8):606-11. (PMID: 9554443) Kony SJ, et al. <i>Lancet</i>. 1997;350(9071):91-5. (PMID: 9228960) Limacher JM, et al. <i>Int J Cancer</i>. 2001;96(4):238-42. (PMID: 11474498) Schneider K, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1999 Jan 19, updated 2013 Apr 11. (PMID: 20301488) Varley JM. <i>Hum Mutat</i>. 2003;21(3):313-20. (PMID: 12619118) Villani A, et al. <i>Lancet Oncol</i>. 2011;12(6):559-67. (PMID: 21601526)</p>
TSC1	Tuberous sclerosis complex	<p>de Vries P, et al. <i>Eur Child Adolesc Psychiatry</i>. 2005;14(4):183-90. (PMID: 15981129) Hinton RB, et al. <i>J Am Heart Assoc</i>. 2014;3(6):e001493. (PMID: 25424575) Krueger DA, et al. <i>Pediatr Neurol</i>. 2013;49(4):255-65. (PMID: 24053983) Northrup H, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1999 Jul 13, updated 2015 Sep 3. (PMID: 20301399)</p>
TSC2	Tuberous sclerosis complex	<p>de Vries P, et al. <i>Eur Child Adolesc Psychiatry</i>. 2005;14(4):183-90. (PMID: 15981129) Hinton RB, et al. <i>J Am Heart Assoc</i>. 2014;3(6):e001493. (PMID: 25424575) Krueger DA, et al. <i>Pediatr Neurol</i>. 2013;49(4):255-65. (PMID: 24053983) Northrup H, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1999 Jul 13, updated 2015 Sep 3. (PMID: 20301399)</p>
VHL	Von Hippel-Lindau syndrome	<p>Binderup ML, et al. <i>Dan Med J</i>. 2013;60(12):B4763. (PMID: 24355456) Frantzen C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 May 17, updated 2015 Aug 6. (PMID: 20301636) Hes FJ, et al. <i>Neth J Med</i>. 2001;59(5):225-34. (PMID: 11705642) Maher ER, et al. <i>Eur J Hum Genet</i>. 2011;19(6):617-23. (PMID: 21386872)</p>
WT1	WT1-related Wilms tumor	<p>Dome JS, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Dec 19, updated 2016 Oct 20. (PMID: 20301471) Scott RH, et al. <i>Arch Dis Child</i>. 2006;91(12):995-9. (PMID: 16857697) Szychot E, et al. <i>Transl Pediatr</i>. 2014;3(1):12-24. (PMID: 26835318) Wu HY, et al. <i>Curr Opin Urol</i>. 2005;15(4):273-6. (PMID: 15928519)</p>

GENE	CONDITION(S)	REFERENCE(S)
ACTA2	Thoracic aortic aneurysms and dissections	<p>Erbel R, et al. <i>Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340)</p> <p>Hiratzka LF, et al. <i>Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780)</p> <p>Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>
ACTC1	<p>Atrial septal defect</p> <p>Dilated cardiomyopathy</p> <p>Hypertrophic cardiomyopathy</p> <p>Left ventricular noncompaction cardiomyopathy</p>	<p>ASD:</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Warnes CA, et al. <i>J Am Coll Cardiol</i>. 2008;52(23):e143-263. (PMID: 19038677)</p> <p>DCM:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486)</p> <p>HCM:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725)</p> <p>Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435)</p> <p>Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>LVNC:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
ACTN2	<p>Dilated cardiomyopathy</p> <p>Hypertrophic cardiomyopathy</p>	<p>DCM:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486)</p> <p>Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725)</p> <p>Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435)</p> <p>Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
ACVRL1	<p>Hereditary hemorrhagic telangiectasia</p> <p>Pulmonary hypertension</p>	<p>HHT:</p> <p>Faughnan ME, et al. <i>J Med Genet</i>. 2011;48(2):73-87. (PMID: 19553198)</p> <p>McDonald J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 26, updated 2017 Feb 2. (PMID: 20301525)</p> <p>PH:</p> <p>Austin ED, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Jul 18, updated 2015 Jun 11. (PMID: 20301658)</p> <p>McLaughlin VV, et al. <i>J Am Coll Cardiol</i>. 2009;53(17):1573-619. (PMID: 19389575)</p>
APOB	<p>Familial hypercholesterolemia</p> <p>Familial hypobetalipoproteinemia</p>	<p>FH:</p> <p>Nordestgaard BG, et al. <i>Eur Heart J</i>. 2013;34(45):3478-90a. (PMID: 23956253)</p> <p>Robinson JG. <i>J Manag Care Pharm</i>. 2013;19(2):139-49. (PMID: 23461430)</p> <p>Watts GF, et al. <i>Int J Cardiol</i>. 2014;171(3):309-25. (PMID: 24418289)</p> <p>Youngblom E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2014 Jan 2, updated 2016 Dec 8. (PMID: 24404629)</p> <p>FHBL:</p> <p>Lee J, et al. <i>J Inherit Metab Dis</i>. 2014;37(3):333-9. (PMID: 24288038)</p>

GENE	CONDITION(S)	REFERENCE(S)
BAG3	Dilated cardiomyopathy Myofibrillar myopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>MFN: M King W, et al. <i>Continuum (Minneapolis)</i>. 2013;19(6 Muscle Disease):1650-73. (PMID: 24305452) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375) Selcen D, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Jan 28, updated 2012 Oct 29. (PMID: 20301672)</p>
BMPR2	Pulmonary arterial hypertension	<p>Austin ED, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Jul 18, updated 2015 Jun 11. (PMID: 20301658) McLaughlin VV, et al. <i>J Am Coll Cardiol</i>. 2009;53(17):1573-619. (PMID: 19389575)</p>
CACNA1C	Brugada syndrome Short QT syndrome Timothy syndrome	<p>BrS: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>SQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>Timothy syndrome: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Feb 15, updated 2015 Jul 16. (PMID: 20301577) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
CACNB2	Brugada syndrome Short QT syndrome	<p>BrS: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>SQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
CALM1	Catecholaminergic polymorphic ventricular tachycardia Long QT syndrome	<p>CPVT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
CALM2	Catecholaminergic polymorphic ventricular tachycardia Long QT syndrome	<p>CPVT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>

GENE	CONDITION(S)	REFERENCE(S)
CALM3	Catecholaminergic polymorphic ventricular tachycardia Long QT syndrome	<p>CPVT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	<p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
CAV1	Pulmonary arterial hypertension	<p>Austin ED, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Jul 18, updated 2015 Jun 11. (PMID: 20301658) McLaughlin VV, et al. <i>J Am Coll Cardiol</i>. 2009;53(17):1573-619. (PMID: 19389575)</p>
CAV3	CAV3-related neuromuscular conditions Hypertrophic cardiomyopathy Long QT syndrome	<p>CAV3-related neuromuscular conditions: Bruno C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 May 14, updated 2012 Sep 6. (PMID: 20301559) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>LQT: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
COL3A1	Ehlers-Danlos syndrome, vascular type	<p>Beridze N, et al. <i>Cardiol Rev</i>. 2012;20(1):4-7. (PMID: 22143279) Erbel R, et al. <i>Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Pepin MG, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1999 Sep 2, updated 2015 Nov 19. (PMID: 20301667) Sobey G. <i>Arch Dis Child</i>. 2015;100(1):57-61. (PMID: 24994860)</p>
CRYAB	Dilated cardiomyopathy Myofibrillar myopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486)</p> <p>MFM: M King W, et al. <i>Continuum (Minneapolis)</i>. 2013;19(6 Muscle Disease):1650-73. (PMID: 24305452) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375) Selcen D, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Jan 28, updated 2012 Oct 29. (PMID: 20301672)</p>

GENE	CONDITION(S)	REFERENCE(S)
CSRP3	Dilated cardiomyopathy Hypertrophic cardiomyopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
DES	Dilated cardiomyopathy Myofibrillar myopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>MFM: Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375) Selcen D, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Jan 28, updated 2012 Oct 29. (PMID: 20301672)</p>
DMD	Becker muscular dystrophy Dilated cardiomyopathy Duchenne muscular dystrophy	<p>BMD: Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) American Academy of Pediatrics Section on Cardiology and Cardiac Surgery. <i>Pediatrics</i>. 2005;116(6):1569-73. (PMID: 16322188) Bushby K, et al. <i>Lancet Neurol</i>. 2010;9(1):77-93. (PMID: 19945913) Bushby K, et al. <i>Lancet Neurol</i>. 2010;9(2):177-89. (PMID: 19945914)</p> <p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>DMD: Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) American Academy of Pediatrics Section on Cardiology and Cardiac Surgery. <i>Pediatrics</i>. 2005;116(6):1569-73. (PMID: 16322188) Bushby K, et al. <i>Lancet Neurol</i>. 2010;9(1):77-93. (PMID: 19945913) Bushby K, et al. <i>Lancet Neurol</i>. 2010;9(2):177-89. (PMID: 19945914)</p>
DSC2	Arrhythmogenic right ventricular cardiomyopathy	<p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p>
DSG2	Arrhythmogenic right ventricular cardiomyopathy Dilated cardiomyopathy	<p>ARVC: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>

GENE	CONDITION(S)	REFERENCE(S)
DSP	Arrhythmogenic right ventricular cardiomyopathy Dilated cardiomyopathy	<p>ARVC: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
EMD	Emery-Dreifuss muscular dystrophy	<p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Bonne G, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Sep 29, updated 2015 Nov 25. (PMID: 20301609) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375)</p>
ENG	Hereditary hemorrhagic telangiectasia	<p>Faughnan ME, et al. <i>J Med Genet</i>. 2011;48(2):73-87. (PMID: 19553198) McDonald J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 26, updated 2017 Feb 2. (PMID: 20301525)</p>
F2	Hereditary thrombophilia	<p>Kujovich JL. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Jul 25, updated 2014 Aug 14. (PMID: 20301327) Varga EA, et al. <i>Clin Genet</i>. 2012;81(1):7-17. (PMID: 21707594)</p>
F5	Hereditary thrombophilia	<p>Kujovich JL. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 1999 May 14, updated 2010 Mar 9. (PMID: 20301542) Varga EA, et al. <i>Clin Genet</i>. 2012;81(1):7-17. (PMID: 21707594)</p>
F9	Hemophilia Hereditary thrombophilia	<p>Hemophilia: www1.wfh.org/publication/files/pdf/1472.pdf Konkle BA, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Oct 2, updated 2017 Jun 15. (PMID: 20301668)</p> <p>Hereditary thrombophilia: Varga EA, et al. <i>Clin Genet</i>. 2012;81(1):7-17. (PMID: 21707594)</p>
FBN1	FBN1-related conditions	<p>Dietz HC. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2001 Apr 18, updated 2017 Feb 2. (PMID: 20301510) Erbel R, et al. <i>Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Loeys BL, et al. <i>J Med Genet</i>. 2010;47(7):476-85. (PMID: 20591885) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299) Tinkle BT, et al. <i>Pediatrics</i>. 2013;132(4):e1059-72. (PMID: 24081994)</p>
FHL1	Emery-Dreifuss muscular dystrophy Hypertrophic cardiomyopathy Reducing body myopathy	<p>EDMD: Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Bonne G, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Sep 29, updated 2015 Nov 25. (PMID: 20301609) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>RBM: Schessl J, et al. <i>Semin Pediatr Neurol</i>. 2011;18(4):257-63. (PMID: 22172421)</p>

GENE	CONDITION(S)	REFERENCE(S)
FLNC	Dilated cardiomyopathy Distal myopathy Hypertrophic cardiomyopathy Myofibrillar myopathy Restrictive cardiomyopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>MFM: M King W, et al. <i>Continuum (Minneapolis)</i>. 2013;19(6 Muscle Disease):1650-73. (PMID: 24305452) Narayanaswami P, et al. <i>Neurology</i>. 2014;83(16):1453-63. (PMID: 25313375) Selcen D, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Jan 28, updated 2012 Oct 29. (PMID: 20301672)</p> <p>RCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
GDF2	Hereditary hemorrhagic telangiectasia	Faughnan ME, et al. <i>J Med Genet</i> . 2011;48(2):73-87. (PMID: 19553198) McDonald J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 26, updated 2017 Feb 2. (PMID: 20301525)
GLA	Fabry disease	Eng CM, et al. <i>Genet Med</i> . 2006;8(9):539-48. (PMID: 16980809) Hopkin RJ, et al. <i>Mol Genet Metab</i> . 2016;117(2):104-13. (PMID: 26546059) Laney DA, et al. <i>J Genet Couns</i> . 2013;22(5):555-64. (PMID: 23860966) Mehta A, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Aug 5, updated 2017 Jan 5. (PMID: 20301469)
GPD1L	Brugada syndrome	Ackerman MJ, et al. <i>Heart Rhythm</i> . 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i> . 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, et al. <i>Heart Rhythm</i> . 2013;10(12):1932-63. (PMID: 24011539)
HCN4	Brugada syndrome Left ventricular noncompaction cardiomyopathy Sinus node dysfunction or bradycardia	<p>BrS: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LVNC: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>Sinus node dysfunction or bradycardia: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
JUP	Arrhythmogenic right ventricular cardiomyopathy	Ackerman MJ, et al. <i>Heart Rhythm</i> . 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i> . 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i> . 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)
KCNE1	Long QT syndrome	Ackerman MJ, et al. <i>Heart Rhythm</i> . 2011;8(8):1308-39. (PMID: 21787999) Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i> . 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, et al. <i>Heart Rhythm</i> . 2013;10(12):1932-63. (PMID: 24011539)

GENE	CONDITION(S)	REFERENCE(S)
KCNE2	Long QT syndrome	<p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
KCNH2	<p>Brugada syndrome</p> <p>Long QT syndrome</p> <p>Short QT syndrome</p>	<p>BrS:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Brugada R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LQT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>SQT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
KCNJ2	<p>Andersen-Tawil syndrome</p> <p>Catecholaminergic polymorphic ventricular tachycardia</p> <p>Short QT syndrome</p>	<p>ATS:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Nguyen HL, et al. <i>Int J Cardiol</i>. 2013;170(1):1-16. (PMID: 24383070)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>Statland JM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Nov 22, updated 2015 Sep 3. (PMID: 20301441)</p> <p>CPVT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Napolitano C, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>SQT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
KCNQ1	<p>Atrial fibrillation</p> <p>Long QT syndrome</p> <p>Short QT syndrome</p>	<p>AFib:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>January CT, et al. <i>Circulation</i>. 2014;130(23):e199-267. (PMID: 24682347)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LQT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Alders M, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>SQT:</p> <p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Priori SG, et al. <i>Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
LAMP2	Danon disease	D'souza RS, et al. <i>Circ Heart Fail</i> . 2014;7(5):843-9. (PMID: 25228319)
LDLR	Familial hypercholesterolemia	<p>Nordestgaard BG, et al. <i>Eur Heart J</i>. 2013;34(45):3478-90a. (PMID: 23956253)</p> <p>Robinson JG. <i>J Manag Care Pharm</i>. 2013;19(2):139-49. (PMID: 23461430)</p> <p>Watts GF, et al. <i>Int J Cardiol</i>. 2014;171(3):309-25. (PMID: 24418289)</p> <p>Youngblom E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2014 Jan 2, updated 2016 Dec 8. (PMID: 24404629)</p>

GENE	CONDITION(S)	REFERENCE(S)
LDLRAP1	Familial hypercholesterolemia	<p>Nordestgaard BG, <i>et al. Eur Heart J.</i> 2013;34(45):3478-90a. (PMID: 23956253)</p> <p>Robinson JG. <i>J Manag Care Pharm.</i> 2013;19(2):139-49. (PMID: 23461430)</p> <p>Youngblom E, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2014 Jan 2, updated 2016 Dec 8. (PMID: 24404629)</p>
LMNA	Congenital muscular dystrophy Dilated cardiomyopathy Emery-Dreifuss muscular dystrophy Limb-Girdle muscular dystrophy	<p>CMD: Kang PB, <i>et al. Neurology.</i> 2015;84(13):1369-78. (PMID: 25825463)</p> <p>Sparks SE, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2001 Jan 22, updated 2012 Aug 23. (PMID: 20301468)</p> <p>DCM: Ackerman MJ, <i>et al. Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Jun 12, updated 2016 Jul 7. (PMID: 20301717)</p> <p>Hershberger RE, <i>et al. J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666)</p> <p>EDMD: Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Bonne G, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Sep 29, updated 2015 Nov 25. (PMID: 20301609)</p> <p>Narayanaswami P, <i>et al. Neurology.</i> 2014;83(16):1453-63. (PMID: 25313375)</p> <p>LGMD: Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Narayanaswami P, <i>et al. Neurology.</i> 2014;83(16):1453-63. (PMID: 25313375)</p> <p>Pegoraro E, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 8, updated 2012 Aug 30. (PMID: 20301582)</p>
MYBPC3	Dilated cardiomyopathy Hypertrophic cardiomyopathy Left ventricular noncompaction cardiomyopathy	<p>DCM: Ackerman MJ, <i>et al. Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486)</p> <p>Hershberger RE, <i>et al. J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, <i>et al. Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725)</p> <p>Gersh BJ, <i>et al. Circulation.</i> 2011;124(24):2761-96. (PMID: 22068435)</p> <p>Hershberger RE, <i>et al. J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666)</p> <p>LVNC: Ackerman MJ, <i>et al. Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999)</p> <p>Al-Khatib SM, <i>et al. J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296)</p> <p>Hershberger RE, <i>et al. J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666)</p>
MYH11	Thoracic aortic aneurysms and dissections	<p>Erbel R, <i>et al. Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340)</p> <p>Hiratzka LF, <i>et al. Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780)</p> <p>Milewicz DM, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>

GENE	CONDITION(S)	REFERENCE(S)
<p style="writing-mode: vertical-rl; transform: rotate(180deg);">INVITAE GENETIC HEALTH SCREEN (continued)</p> <p style="writing-mode: vertical-rl; transform: rotate(180deg);">INVITAE CARDIO SCREEN (continued)</p> <p>MYH7</p>	<p>Dilated cardiomyopathy</p> <p>Hypertrophic cardiomyopathy</p> <p>Laing distal myopathy</p> <p>Left ventricular noncompaction cardiomyopathy</p> <p>Myosin storage myopathy</p>	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>Laing distal myopathy: Lamont P, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Oct 17, updated 2015 Mar 12. (PMID: 20301606) Tajsharghi H, et al. <i>Acta Neuropathol</i>. 2013;125(1):3-18. (PMID: 22918376) Wang CH, et al. <i>J Child Neurol</i>. 2012;27(3):363-82. (PMID: 22431881)</p> <p>LVNC: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>MSM: Lamont P, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Oct 17, updated 2015 Mar 12. (PMID: 20301606) Tajsharghi H, et al. <i>Acta Neuropathol</i>. 2013;125(1):3-18. (PMID: 22918376) Wang CH, et al. <i>J Child Neurol</i>. 2012;27(3):363-82. (PMID: 22431881)</p>
MYL2	Hypertrophic cardiomyopathy	<p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
MYL3	Hypertrophic cardiomyopathy	<p>Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
MYLK	Thoracic aortic aneurysms and dissections	<p>Erbel R, et al. <i>Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>
NKX2-5	NKX2-5 related cardiovascular conditions	<p>Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) January CT, et al. <i>Circulation</i>. 2014;130(23):e199-267. (PMID: 24682347) Warnes CA, et al. <i>J Am Coll Cardiol</i>. 2008;52(23):e143-263. (PMID: 19038677)</p>
PCSK9	Familial hypercholesterolemia	<p>Nordestgaard BG, et al. <i>Eur Heart J</i>. 2013;34(45):3478-90a. (PMID: 23956253) Robinson JG. <i>J Manag Care Pharm</i>. 2013;19(2):139-49. (PMID: 23461430) Watts GF, et al. <i>Int J Cardiol</i>. 2014;171(3):309-25. (PMID: 24418289) Youngblom E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2014 Jan 2, updated 2016 Dec 8. (PMID: 24404629)</p>

GENE	CONDITION(S)	REFERENCE(S)
PKP2	Arrhythmogenic right ventricular cardiomyopathy Brugada syndrome Dilated cardiomyopathy	<p>ARVC: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>BrS: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, <i>et al. Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
PLN	Arrhythmogenic right ventricular cardiomyopathy Dilated cardiomyopathy Hypertrophic cardiomyopathy	<p>ARVC: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, <i>et al. Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
PRKAG2	Hypertrophic cardiomyopathy Wolff-Parkinson-White syndrome	<p>Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, <i>et al. Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
PRKG1	Thoracic aortic aneurysms and dissections	<p>Erbel R, <i>et al. Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, <i>et al. Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Milewicz DM, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>
PROC	Hereditary thrombophilia	<p>Varga EA, <i>et al. Clin Genet</i>. 2012;81(1):7-17. (PMID: 21707594)</p>
PROS1	Hereditary thrombophilia	<p>Varga EA, <i>et al. Clin Genet</i>. 2012;81(1):7-17. (PMID: 21707594)</p>
RBM20	Dilated cardiomyopathy	<p>Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>

GENE	CONDITION(S)	REFERENCE(S)
RYR2	Arrhythmogenic right ventricular cardiomyopathy Catecholaminergic polymorphic ventricular tachycardia Left ventricular noncompaction cardiomyopathy	<p>ARVC: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666) McNally E, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>CPVT: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Napolitano C, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2004 Oct 14, updated 2016 Oct 13. (PMID: 20301466) Priori SG, <i>et al. Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>LVNC: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
SCN5A	Atrial fibrillation Brugada syndrome Dilated cardiomyopathy Long QT syndrome	<p>AFib: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) January CT, <i>et al. Circulation</i>. 2014;130(23):e199-267. (PMID: 24682347) Priori SG, <i>et al. Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>BrS: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Brugada R, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Mar 31, updated 2016 Nov 17. (PMID: 20301690) Priori SG, <i>et al. Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p> <p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>LQT: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Alders M, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 20, updated 2015 Jun 18. (PMID: 20301308) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Priori SG, <i>et al. Heart Rhythm</i>. 2013;10(12):1932-63. (PMID: 24011539)</p>
SERPINC1	Hereditary thrombophilia	Varga EA, <i>et al. Clin Genet</i> . 2012;81(1):7-17. (PMID: 21707594)
SGCD	Limb-Girdle muscular dystrophy	Al-Khatib SM, <i>et al. J Am Coll Cardiol</i> . 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Narayanaswami P, <i>et al. Neurology</i> . 2014;83(16):1453-63. (PMID: 25313375) Pegoraro E, <i>et al</i> . In: Pagon RA, <i>et al</i> , eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 8, updated 2012 Aug 30. (PMID: 20301582)
SMAD3	Loews-Dietz syndrome Thoracic aortic aneurysms and dissections/	<p>LDS: Erbel R, <i>et al. Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, <i>et al. Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Loews BL, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Feb 28, updated 2013 Jul 11. (PMID: 20301312) Milewicz DM, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299) van de Laar IM, <i>et al. J Med Genet</i>. 2012;49(1):47-57. (PMID: 22167769)</p> <p>TAAD: Erbel R, <i>et al. Eur Heart J</i>. 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, <i>et al. Circulation</i>. 2010;121(13):e266-369. (PMID: 20233780) Milewicz DM, <i>et al</i>. In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299) van de Laar IM, <i>et al. J Med Genet</i>. 2012;49(1):47-57. (PMID: 22167769)</p>

GENE	CONDITION(S)	REFERENCE(S)
SMAD4	Hereditary hemorrhagic telangiectasia Juvenile polyposis syndrome	<p>HHT: Faughnan ME, et al. <i>J Med Genet.</i> 2011;48(2):73-87. (PMID: 19553198) McDonald J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 26, updated 2017 Feb 2. (PMID: 20301525)</p> <p>JPS: NCCN®: Genetic/Familial High-Risk Assessment: Colorectal V.3.2017¹ Larsen Haidle J, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 May 13, updated 2017 Mar 19. (PMID: 20301642) Syngal S, et al. <i>Am J Gastroenterol.</i> 2015;110(2):223-62; quiz 263. (PMID: 25645574)</p>
TCAP	Limb-Girdle muscular dystrophy	<p>Al-Khatib SM, et al. <i>J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Narayanaswami P, et al. <i>Neurology.</i> 2014;83(16):1453-63. (PMID: 25313375) Pegoraro E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Jun 8, updated 2012 Aug 30. (PMID: 20301582)</p>
TGFB2	Loeys-Dietz syndrome	<p>Erbel R, et al. <i>Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340) Hiratzka L, et al. <i>Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780) Loeys BL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Feb 28, updated 2013 Jul 11. (PMID: 20301312)</p>
TGFB3	Arrhythmogenic right ventricular cardiomyopathy Rienhoff syndrome	<p>ARVC: Ackerman MJ, et al. <i>Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p> <p>Rienhoff syndrome: Kuechler A, et al. <i>Mol Cell Probes.</i> 2015;29(5):330-4. (PMID: 26184463)</p>
TGFB1	Loeys-Dietz syndrome Multiple self-healing squamous epithelioma Thoracic aortic aneurysms and dissections	<p>LDS: Erbel R, et al. <i>Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780) Loeys BL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Feb 28, updated 2013 Jul 11. (PMID: 20301312) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p> <p>MSSE: Ferguson-Smith MA, et al. <i>Int J Biochem Cell Biol.</i> 2014;53:520-5. (PMID: 24747516) Kwiek B, et al. <i>J Am Acad Dermatol.</i> 2016;74(6):1220-33. (PMID: 26853179)</p> <p>TAAD: Erbel R, et al. <i>Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>
TGFB2	Loeys-Dietz syndrome Thoracic aortic aneurysms and dissections	<p>LDS: Erbel R, et al. <i>Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780) Loeys BL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Feb 28, updated 2013 Jul 11. (PMID: 20301312) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p> <p>TAAD: Erbel R, et al. <i>Eur Heart J.</i> 2014;35(41):2873-926. (PMID: 25173340) Hiratzka LF, et al. <i>Circulation.</i> 2010;121(13):e266-369. (PMID: 20233780) Milewicz DM, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Feb 13, updated 2016 Dec 29. (PMID: 20301299)</p>
TMEM43	Arrhythmogenic right ventricular cardiomyopathy	<p>Ackerman MJ, et al. <i>Heart Rhythm.</i> 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol.</i> 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail.</i> 2009;15(2):83-97. (PMID: 19254666) McNally E, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Apr 18, updated 2017 May 25. (PMID: 20301310)</p>

GENE	CONDITION(S)	REFERENCE(S)
TNNC1	Dilated cardiomyopathy Hypertrophic cardiomyopathy	<p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, <i>et al. Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
TNNI3	Dilated cardiomyopathy Hypertrophic cardiomyopathy Restrictive cardiomyopathy	<p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, <i>et al. Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>RCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
TNNT2	Dilated cardiomyopathy Hypertrophic cardiomyopathy Left ventricular noncompaction cardiomyopathy Restrictive cardiomyopathy	<p>DCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, <i>et al.</i> In: Pagon RA, <i>et al</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, <i>et al. Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>LVNC: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, <i>et al. J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>RCM: Ackerman MJ, <i>et al. Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Hershberger RE, <i>et al. J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>

INVITAE CARDIO SCREEN (continued)	INVITAE GENETIC HEALTH SCREEN (continued)	GENE	CONDITION(S)	REFERENCE(S)
		TPM1	Dilated cardiomyopathy Hypertrophic cardiomyopathy Left ventricular noncompaction cardiomyopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>LVNC: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>
VCL	Dilated cardiomyopathy Hypertrophic cardiomyopathy	<p>DCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Hershberger RE, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 Jul 27, updated 2015 Sep 24. (PMID: 20301486) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p> <p>HCM: Ackerman MJ, et al. <i>Heart Rhythm</i>. 2011;8(8):1308-39. (PMID: 21787999) Al-Khatib SM, et al. <i>J Am Coll Cardiol</i>. 2017 Oct 25. [Epub ahead of print] (PMID: 29097296) Cirino AL, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2008 Aug 5, updated 2014 Jan 16. (PMID: 20301725) Gersh BJ, et al. <i>Circulation</i>. 2011;124(24):2761-96. (PMID: 22068435) Hershberger RE, et al. <i>J Card Fail</i>. 2009;15(2):83-97. (PMID: 19254666)</p>		
ATP7B	Wilson disease	<p>Saudubray JM, Baumgartner MR, Walter, J. <i>Inborn Metabolic Diseases: Diagnosis and Treatment</i>, 6th ed. Berlin, 2016.</p> <p>Weiss KH. Wilson Disease. 1999 Oct 22 [Updated 2016 Jul 29]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1999, Oct, 22, updated 2016, Jul 29. (PMID:22340672)</p>		
CACNAT5	Hypokalemic periodic paralysis Malignant hyperthermia susceptibility	<p>HOKPP: Levitt JO. <i>J Transl Med</i>. 2008;6:18. (PMID: 18426576) Vicart S, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2002 Apr 30, updated 2014 Jul 31. (PMID: 20301512)</p> <p>MHS: Glahn KP, et al. <i>Br J Anaesth</i>. 2010;105(4):417-20. (PMID: 20837722) Rosenberg H, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Dec 19, updated 2013 Jan 31. (PMID: 20301325) Schneiderbanger D, et al. <i>Ther Clin Risk Manag</i>. 2014;10:355-62. (PMID: 24868161)</p>		
HAMP	Hereditary hemochromatosis	<p>Bacon BR, et al. <i>Hepatology</i>. 2011;54(1):328-43. (PMID: 21452290) Goldberg YP. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Feb 17, updated 2011 Aug 11. (PMID: 20301349)</p>		
HFE	Hereditary hemochromatosis	<p>Bacon BR, et al. <i>Hepatology</i>. 2011;54(1):328-43. (PMID: 21452290) Seckington R, et al. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2000 Apr 3, updated 2015 Sep 17. (PMID: 20301613)</p>		
HJV	Hereditary hemochromatosis	<p>Bacon BR, et al. <i>Hepatology</i>. 2011;54(1):328-43. (PMID: 21452290) Goldberg YP. In: Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Feb 17, updated 2011 Aug 11. (PMID: 20301349)</p>		
OTC	Ornithine transcarbamylase deficiency	<p>Saudubray JM, Baumgartner MR, Walter, J. <i>Inborn Metabolic Diseases: Diagnosis and Treatment</i>, 6th ed. Berlin, 2016.</p> <p>Lamb S, et al. <i>BMJ Case Rep</i>. 2013; Jan 2. (PMID: 23283608) Lichter-Konecki U, Caldovic L, Morizono H, et al. <i>Ornithine Transcarbamylase Deficiency</i>. 2013 Aug 29 [Updated 2016 Apr 14]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 2013, Aug 29, updated 2016, Apr 14. (PMID: 24006547)</p>		

INVITAE GENETIC HEALTH SCREEN (continued)	GENE	CONDITION(S)	REFERENCE(S)
	RYR1	Central core disease Centronuclear myopathy Malignant hyperthermia susceptibility	<p>CCD: Malicdan MCV, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 May 16, updated 2014 Dec 4. (PMID: 20301565) Wang CH, <i>et al.</i> <i>J Child Neurol.</i> 2012;27(3):363-82. (PMID: 22431881)</p> <p>CNM: Malicdan MCV, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2007 May 16, updated 2014 Dec 4. (PMID: 20301565) Wang CH, <i>et al.</i> <i>J Child Neurol.</i> 2012;27(3):363-82. (PMID: 22431881)</p> <p>MHS: Glahn KP, <i>et al.</i> <i>Br J Anaesth.</i> 2010;105(4):417-20. (PMID: 20837722) Rosenberg H, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2003 Dec 19, updated 2013 Jan 31. (PMID: 20301325) Schneiderbanger D, <i>et al.</i> <i>Ther Clin Risk Manag.</i> 2014;10:355-62. (PMID: 24868161)</p>
	SERPINA1	Alpha-1-antitrypsin deficiency	<p>American Thoracic Society, <i>et al.</i> <i>Am J Respir Crit Care Med.</i> 2003;168(7):818-900. (PMID: 14522813) Marciniuk DD, <i>et al.</i> <i>Can Respir J.</i> 2012;19(2):109-16. (PMID: 22536580) Stoller JK, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2006 Oct 27, updated 2017 Jan 19. (PMID: 20301692)</p>
	SLC40A1	Hereditary hemochromatosis	Bacon BR, <i>et al.</i> <i>Hepatology.</i> 2011;54(1):328-43. (PMID: 21452290)
	TFR2	Hereditary hemochromatosis	<p>Bacon BR, <i>et al.</i> <i>Hepatology.</i> 2011;54(1):328-43. (PMID: 21452290) Camaschella C, <i>et al.</i> In: Pagon RA, <i>et al.</i>, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington; 2005 Aug 29, updated 2011 Jun 9. (PMID: 20301523)</p>

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3. Referenced with permission from the NCCN: Gastric Cancer. Version 5.2017. © National Comprehensive Cancer Network, Inc. 2017. All rights reserved. Accessed October 10, 2017. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.nccn.org).
4. Referenced with permission from the NCCN: Soft Tissue Sarcoma. Version 1.2018. © National Comprehensive Cancer Network, Inc. 2017. All rights reserved. Accessed October 10, 2017. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.nccn.org).
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6. Referenced with permission from the NCCN: Thyroid Carcinoma. Version 2.2017. © National Comprehensive Cancer Network, Inc. 2017. All rights reserved. Accessed August 4, 2017. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.nccn.org).
7. Referenced with permission from the NCCN: Pancreatic Adenocarcinoma. Version 3.2017. © National Comprehensive Cancer Network, Inc. 2017. All rights reserved. Accessed October 10, 2017. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.nccn.org).

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