

Genetics of cardiac arrhythmias (ht46334)

Web only refs

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1. Keating M, Atkinson D, Dunn C, *et al.* Linkage of a cardiac arrhythmia, the long QT syndrome, and the Harvey ras-1 gene. *Science* 1991;**252**(5006):704–6.
2. Wang Q, Curran ME, Splawski I, *et al.* Positional cloning of a novel potassium channel gene: KVLQT1 mutations cause cardiac arrhythmias. *Nat Genet* 1996;**12**(1):17–23.
3. Jiang C, Atkinson D, Towbin JA, *et al.* Two long QT syndrome loci map to chromosomes 3 and 7 with evidence for further heterogeneity. *Nat Genet* 1994;**8**(2):141–7.
4. Sanguinetti MC, Curran ME, Spector PS, *et al.* Spectrum of HERG K⁺-channel dysfunction in an inherited cardiac arrhythmia. *Proc Natl Acad Sci USA* 1996;**93**(5):2208–12.
5. Bennett PB, Yazawa K, Makita N. Molecular mechanism for an inherited cardiac arrhythmia. *Nature* 1995;**376**:683–5.
6. Schott JJ, Charpentier F, Peltier S, *et al.* Mapping of a gene for long QT syndrome. *Am J Hum Genet* 1995;**57**(5):1114–22.
7. Splawski I, Tristani-Firouzi M, Lehmann MH, *et al.* Mutations in the hminK gene cause long QT syndrome and suppress I(Ks) function. *Nat Genet* 1997;**17**:338–40.
8. Abbott GW, Sesti F, Splawski I, *et al.* MiRP1 forms I(kr) potassium channels with HERG and is associated with cardiac arrhythmia. *Cell* 1999;**97**:175–87.
9. Neyroud N, Tesson F, Denjoy I, *et al.* A novel mutation in the potassium channel gene KVLQT1 causes the Jervell and Lange-Nielsen cardioauditory syndrome. *Nat Genet* 1997;**15**(2):186–9.
10. Splawski I, Timothy KW, Vincent GM, *et al.* Molecular basis of the long-QT syndrome associated with deafness. *N Engl J Med* 1997;**336**(22):1562–7.
11. Wollnik B, Schroeder BC, Kubisch C, *et al.* Pathophysiological mechanisms of dominant and recessive KVLQT1 K⁺ channel mutations found in inherited cardiac arrhythmias. *Hum Mol Genet* 1997 Oct;**6**(11):1943–9.
12. Chouabe C, Neyroud N, Guicheney P, *et al.* Properties of KvLQT1 K⁺ channel mutations in Romano-Ward and Jervell and Lange-Nielsen inherited cardiac arrhythmias. *EMBO J* 1997 Sep 1;**16**(17):5472–9.
13. Schulze-Bahr E, Wang Q, Wedekind H, *et al.* KCNE1 mutations cause Jervell and Lange-Nielsen syndrome. *Nat Genet* 1997;**17**(3):267–8.
14. Tyson J, Tranebjaerg L, Bellman S, *et al.* IsK and KvLQT1: mutation in either of the two subunits of the slow component of the delayed rectifier potassium channel can cause Jervell and Lange-Nielsen syndrome. *Hum Mol Genet* 1997;**6**(12):2179–85.
15. Bianchi L, Shen Z, Dennis AT, *et al.* Cellular dysfunction of LQT5-minK mutants: abnormalities of IKs, IKr and trafficking in long QT syndrome. *Hum Mol Genet* 1999 Aug;**8**(8):1499–507.
16. Brugada R, Hong K, Dumaine R. Sudden death associated with Short-QT syndrome linked to mutations in HERG. *Circulation* 2004;**109**:r151–r156.
17. Bellocq C, van Ginneken AC, Bezzina CR, *et al.* Mutation in the KCNQ1 gene leading to the short QT-interval syndrome. *Circulation* 2004 May 25;**109**(20):2394–7.
18. Priori SG, Pandit SV, Rivolta I, *et al.* A Novel Form of Short QT Syndrome (SQT3) Is Caused by a Mutation in the KCNJ2 Gene. *Circ Res* 2005 Mar 10.
19. Chen Q, Kirsch GE, Zhang D, *et al.* Genetic basis and molecular mechanism for idiopathic ventricular fibrillation. *Nature* 1998;**392**(6673):293–6.
20. Weiss R, Barmada MM, Nguyen T, *et al.* Clinical and molecular heterogeneity in the Brugada syndrome: a novel gene locus on chromosome 3. *Circulation* 2002;**105**(6):707–13.
21. Laitinen PJ, Brown KM, Piippo K, *et al.* Mutations of the cardiac ryanodine receptor (RyR2) gene in familial polymorphic ventricular tachycardia. *Circulation* 2001 Jan 30;**103**(4):485–90.

22. Priori SG, Napolitano C, Tiso N, *et al.* Mutations in the cardiac ryanodine receptor gene (hRyR2) underlie catecholaminergic polymorphic ventricular tachycardia. *Circulation* 2001 Jan 16;**103**(2):196–200.
23. Wehrens XH, Lehmann SE, Huang F, *et al.* FKBP12.6 deficiency and defective calcium release channel (ryanodine receptor) function linked to exercise-induced sudden cardiac death. *Cell* 2003 Jun 27;**113**(7):829–40.
24. George CH, Higgs GV, Lai FA. Ryanodine receptor mutations associated with stress-induced ventricular tachycardia mediate increased calcium release in stimulated cardiomyocytes. *Circ Res* 2003 Sep 19;**93**(6):531–40.
25. Jiang D, Xiao B, Zhang L, *et al.* Enhanced basal activity of a cardiac Ca²⁺ release channel (ryanodine receptor) mutant associated with ventricular tachycardia and sudden death. *Circ Res* 2002 Aug 9;**91**(3):218–25.
26. Lahat H, Pras E, Olender T, *et al.* A missense mutation in a highly conserved region of CASQ2 is associated with autosomal recessive catecholamine-induced polymorphic ventricular tachycardia in Bedouin families from Israel. *Am J Hum Genet* 2001 Dec;**69**(6):1378–84.
27. Viatchenko-Karpinski S, Terentyev D, Gyorke I, *et al.* Abnormal calcium signaling and sudden cardiac death associated with mutation of calsequestrin. *Circ Res* 2004 Mar 5;**94**(4):471–7.
28. Schulze-Bahr E, Neu A, Friederich P, *et al.* Pacemaker channel dysfunction in a patient with sinus node disease. *J Clin Invest* 2003 May;**111**(10):1537–45.
29. Benson DW, Wang DW, Dymment M, *et al.* Congenital sick sinus syndrome caused by recessive mutations in the cardiac sodium channel gene (SCN5A). *J Clin Invest* 2003 Oct;**112**(7):1019–28.
30. Brink PA, Ferreira A, Moolman JC, *et al.* Gene for progressive familial heart block type I maps to chromosome 19q13. *Circulation* 1995 Mar 15;**91**(6):1633–40.
31. Schott JJ, Alshinawi C, Kyndt F, *et al.* Cardiac conduction defects associate with mutations in SCN5A. *Nat Genet* 1999 Sep;**23**(1):20–1.
32. Tan HL, Bink-Boelkens MT, Bezzina CR, *et al.* A sodium-channel mutation causes isolated cardiac conduction disease. *Nature* 2001 Feb 22;**409**(6823):1043–7.
33. Kyndt F, Schott JJ, Probst V, *et al.* A new locus for isolated cardiac conduction defect maps to 16q23–24 [abstract]. *Circulation* 2000;**102**(18 Suppl II):II–358.
34. Brugada R, Tapscott T, Czernuszewicz GZ, *et al.* Identification of a genetic locus for familial atrial fibrillation. *N Engl J Med* 1997 Mar 27;**336**(13):905–11.
35. Chen YH, Xu SJ, Bendahhou S, *et al.* KCNQ1 gain-of-function mutation in familial atrial fibrillation. *Science* 2003 Jan 10;**299**(5604):251–4.
36. Ellinor PT, Shin JT, Moore RK, *et al.* Locus for atrial fibrillation maps to chromosome 6q14–16. *Circulation* 2003 Jun 17;**107**(23):2880–3.
37. Yang Y, Xia M, Jin Q, *et al.* Identification of a KCNE2 gain-of-function mutation in patients with familial atrial fibrillation. *Am J Hum Genet* 2004 Nov;**75**(5):899–905.
38. Wilde AA, Jongbloed RJ, Doevendans PA, *et al.* Auditory stimuli as a trigger for arrhythmic events differentiate HERG-related (LQTS2) patients from KVLQT1-related patients (LQTS1). *J Am Coll Cardiol* 1999 Feb;**33**(2):327–32.
39. Moss AJ, Robinson JL, Gessman L, *et al.* Comparison of clinical and genetic variables of cardiac events associated with loud noise versus swimming among subjects with the long QT syndrome. *Am J Cardiol* 1999 Oct 15;**84**(8):876–9.
40. Benhorin J, Taub R, Goldmit M, *et al.* Effects of flecainide in patients with new SCN5A mutation: mutation-specific therapy for long-QT syndrome? *Circulation* 2000 Apr 11;**101**(14):1698–706.
41. van den Berg MP, Wilde AA, Viersma TJW, *et al.* Possible bradycardic mode of death and successful pacemaker treatment in a large family with features of long QT syndrome type 3 and Brugada syndrome. *J Cardiovasc Electrophysiol* 2001 Jun;**12**(6):630–6.

42. Veldkamp MW, Viswanathan PC, Bezzina C, *et al.* Two distinct congenital arrhythmias evoked by a multidysfunctional Na(+) channel. *Circ Res* 2000 May 12;**86**(9):E91–E97.
43. Veldkamp MW, Wilders R, Baartscheer A, *et al.* Contribution of sodium channel mutations to bradycardia and sinus node dysfunction in LQT3 families. *Circ Res* 2003 May 16;**92**(9):976–83.
44. Arking DE, Chugh SS, Chakravarti A, *et al.* Genomics in sudden cardiac death. *Circ Res* 2004 Apr 2;**94**(6):712–23.