

Facts on Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C)

- **ARVD/C is a dominantly inherited disease affecting as many as 1 in 1000 individuals.**
[Peters S, et al. \(2004\) Int Journal Cardiology 97:499-501](#)
- **ARVD/C is caused by mutations in any one of at least 7 genes, but mutations in one of 5 genes are most commonly found.**
 - Mutations in *PKP2*, *DSG2*, *DSP2*, *DSC2*, or *TMEM43* account for up to 74% of familial ARVD/C.
[Dalal D, et al. \(2006\) JACC 48:1416-24](#)
[Pilichou K, et al. \(2006\) Circulation 113:1171-79](#)
[Bauce B, et al. \(2005\) Eur Heart Jour 26:1666-75](#)
[Syrris P, et al. \(2006\) AJHG 79:978-84](#)
[Merner N, et al. \(2008\) AJHG 82: 809-21](#)
- **ARVD/C is the second most common cause of sudden cardiac death (SCD) in young adults, including trained athletes. Affected individuals are often unaware of their condition.**
 - About 20% SCD in young adults and athletes is due to ARVD/C.
[Thiene G, et al. \(1988\) NEJM 318: 129-33](#)
[Corrado D, et al. \(1998\) NEJM 339: 364-69](#)
- **Extensive cardiac screening at regular intervals can identify patients at high risk for SCD, who may benefit from implantation of a cardioverter-defibrillator (ICD).**
 - ICDs averted potentially lethal arrhythmias in 72% of 132 high-risk ARVD/C patients.
[Corrado D, et al. \(2003\) Circulation 108: 3084-91](#)
- **Children who are genetically predisposed to ARVD/C may be advised not to participate in certain competitive sports.**
[Maron BJ, et al. \(2004\) Circulation 109:2807-16](#)
- **Genetic testing can confirm a diagnosis of familial ARVD/C in the index patient for a family and identify family members with a predisposition for ARVD/C at any age.**
 - Family members have a ~50% chance of inheriting ARVD/C.
[Awad MM, et al. \(2008\) Nature Clin Practice Cardio Med 5: 258-67](#)
[Hamid MS, et al. \(2002\) JACC 40: 1445-50](#)