

## Facts on Familial Dilated Cardiomyopathy (DCM)

**DCM is a cardiac disease affecting at least 1 in 2500 individuals. About 30-60% of DCM is familial.**

[Colombo MG, et al. \(2008\) Cardiovasc Ultrasound 6: 62-71](#)

- **Familial DCM is caused by mutations in any one of at least 20 genes. Mutations in *MYH7*, *TNNT2*, *TPM1*, *MYBPC3*, *ACTC*, *TNNI3*, and *LMNA* are responsible for 15-40% of familial DCM.**
  - *MYH7*, *TNNT2*, *TPM1*, *MYBPC3*, *ACTC* and *LMNA* mutations have been associated with autosomal dominant DCM. *TNNI3* has been associated with autosomal recessive DCM.
    - [Burkett EL, et al. \(2005\) J. Am. Coll. Cardiol. 45:969-981](#)
    - [Fatkin D, et al. \(2002\) Physiol. Rev. 82:945-980](#)
    - [Murphy RT et al. \(2004\) Lancet 363:371-372](#)
    - [Colombo MG, et al. \(2008\) Cardiovasc Ultrasound 6: 62-71](#)

**DCM is a progressive disease that can lead to life-threatening cardiac dysfunction.**

- If not treated properly, DCM has a five-year mortality rate of up to 50%, mostly due to congestive heart failure.
- DCM-related heart failure is among the most common indications for heart transplantation.
- DCM is associated with cardiac arrhythmias and risk of stroke or sudden cardiac death.
  - [Elliott P \(2000\) Heart 84:106-112](#)
- **Early diagnosis and treatment can delay progression of DCM.**
  - Lifestyle changes and pharmacological treatment with ACE inhibitors and/or  $\beta$ -blockers can slow the development of progressive heart failure.
    - [Taylor MRG, et al \(2006\) Orph. J. Rare Dis. 1:27](#)
- **DCM is often diagnosed relatively late.**
  - In many patients, DCM progression is already quite severe at the time of diagnosis.
    - [Taylor MRG, et al \(2006\) Orph. J. Rare Dis. 1:27](#)
- **To promote early diagnosis of DCM, regular cardiac screening of at-risk patients must be maintained throughout their lifetime.**
  - Absence of symptoms cannot guarantee a favorable prognosis.
    - [Crispell KA, et al. \(2002\) J Am. Coll. Cardiol. 39:1503-1507](#)
- **Genetic testing can confirm a clinical diagnosis of familial DCM, even in the absence of a known and conclusive family history, and distinguish at-risk individuals from individuals not at increased risk among family members of confirmed index patients.**
  - At-risk family members can implement lifestyle changes and preventive pharmacologic therapy at an early age.
  - Only family members harboring the familial DCM-associated mutations have to undergo regular cardiac screening.
    - [Burkett EL, et al. \(2005\) J. Am. Coll. Cardiol. 45:969-981](#)
    - [Colombo MG, et al. \(2008\) Cardiovasc Ultrasound 6: 62-71](#)