

## Kardiogenetik

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### Referenzen

1. Geisterfer-Lowrance AA et al. A molecular basis for familial hypertrophic cardiomyopathy : a beta myosin heavy chain gene missense mutation. *Cell* 1990 ;62 :999-1006
2. Curran ME et al. A molecular basis for cardiac arrhythmia : HERG mutations cause long QT syndrome. *Cell* 1995 ; 80 :795-803.
3. Priori SG et al. HRS/EHRA/APHRS expert consensus statement on the diagnosis and management of patients with inherited primary arrhythmia syndromes. *Europace* 2013 ;15 :1389-406.
4. Elliott PM et al. 2014 ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy : the task force for the diagnosis and management of hypertrophic cardiomyopathy of the European Society of Cardiology (ESC). *Eur Heart J* 2014 ;35 :2733-79.
5. Richards S. et al : Standards and Guidelines for the Interpretation of Sequence Variants : A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 ; 17(5) :405-424.
6. Gesetz über genetische Untersuchungen am Menschen. [www.admin.ch](http://www.admin.ch)
7. Maron BJ. Hypertrophic cardiomyopathy : a systematic review. *JAMA* 2002 ;287 :1308-20.
8. O'Mahony C et al. A novel clinical risk prediction model for sudden cardiac death in hypertrophic cardiomyopathy (HCM Risk-SCD). *Eur Heart J* 2014 ; 35 :2010-20.
9. Maron BJ et al. Genetics of hypertrophic cardiomyopathy after 20 years : clinical perspectives. *J Am Coll Cardiol.* 2012 ;60(8) :705-15.
10. Akhtar M, Elliott P. The genetics of hypertrophic cardiomyopathy. *Global Cardiology Science & Practice.* 2018 ;36 <https://doi.org/10.21542/gcsp.2018.36>
11. Wordsworth S et al. DNA testing for hypertrophic cardiomyopathy : a cost-effectiveness model. *Eur Heart J* 2010 ;31 :926-935.