

6. Literaturverzeichnis

[1] Maron, B.J., J.M. Gardin, J. M. Flack, S. S. Gidding, T. T. Kurosaki, D. E. Bild: Prevalence of hypertrophic cardiomyopathy in a general population of young adults: echocardiographic analysis of 4111 subjects in the CARDIA Study. *Circulation* 92 (1995) 785-789.

[2] **Teare** D. Asymmetrical hypertrophy of the heart in young adults. *Br Heart J* 1958; 20:1-18..

[3] **Richardson**, P., W. McKenna, M. Bristow et al.: Report of the 1995 World Health Organisation/ International Society and federation of Cardiology Task Force on the Definition and Classification of Cardiomyopathies. *Circulation* 93 (1996) 841-842

[4] **Maron**, B.J. Hypertrophic cardiomyopathy: A systematic review. *JAMA* 2002; 287; 24; 1965-1991

[5] **Maron**, B.J., Bonow RO, Cannon III RO et al. (1987)) Hypertrophic cardiomyopathy. Interrelations of clinical manifestations, pathophysiology and therapy (first and second part). *N Engl J Med* 316: 780 – 789, 844- 852

[6] **Maron**, B.J. (1997) Hypertrophic cardiomyopathy. *Lancet* 350: 127-133

[7] **Lind** JM, Chiu C, Semsarian C. Genetic basis of hypertrophic cardiomyopathy. *Expert Rev Cardiovasc Ther.* 2006 Nov;4(6): 927-34. Review.

[8] **Kelly** DP, Strauss AW (1994) Inherited cardiomyopathies. *N Engl J Med* 330: 930-932

[9] **Spirito**, P. Ch., E. Seidman, W.J. McKenna, B. J. Maron: The management of hypertrophic cardiomyopathy. *N.Engl. J.Med.* 336 (1997) 775-785

[10] **Maron** B.J., Mc Kenna WJ, Danielson GK, et al. American College of Cardiology/European Society of Cardiology Clinical Expert Consensus Document on Hypertrophic Cardiomyopathy. A report of the American College of Cardiology Task Force on Clinical Expert Consensus Documents and the European Society of Cardiology Committee for Practice Guidelines Committee to Develop an Expert Consensus Document on Hypertrophic Cardiomyopathy. *J Am Coll Cardiol* 2003; 42: 1687-1713 and *Eur Heart J* 2003; 24: 1965-1991.

[12] **Pare** JA, Fraser RG, Pirozynski WJ, Shanks JA, Stubington D. Hereditary cardiovascular dysplasia. A form of familial cardiomyopathy. *Am J Med.* 1961 Jul;31:37-62.

[13] **Jarcho** JA, McKenna W, Pare JAP(1989); Mapping a gene for familial hypertrophic cardiomyopathy to chromosome 14q1.N.Engl. J. Med.321: 1372-1378

- [14] **Geisterfer- Lowrance** AA, Kass S, Tanigawa G et al.(1990) A molecular basis for familial hypertrophic cardiomyopathy: a beta cardiac myosin heavy chain gene missense mutation. *Cell* 62: 999-100
- [15] **Watkins** H, McKenna WJ, Thierfelder et al. (1995) Mutations in the genes for cardiac troponin T and α -tropomyosin in hypertrophic cardiomyopathy. *N Engl J Med* 332: 1058-1064
- [16] **Niimura** H, Bachinski LL, Sangwatanaroj S et al.(1998) Mutations in the gene for cardiac myosin-binding protein C and late-onset familial hypertrophic cardiomyopathy. *N Engl J Med* 338: 1248-1257
- [17] **Morgensen** J, Klausen IC, Pedersen AK, Egeblad H, Bross P, Kruse TA, Gregersen N, Hansen PS, Baandrup U, Borglum AD. Alpha-cardiac actin is a novel disease gene in familial hypertrophic cardiomyopathy. *J Clin Invest* 1999;103:39-43.
- [18] **Satoh** M, Takahashi M, Sakamoto T, Hiroe M, Marumo F, Kimura A. Structural analysis of the titin gene in hypertrophic cardiomyopathy: identification of a novel disease gene. *Biochem Biophys Res Commun* 1999;262:411-7.
- [19] **Huxley**, A.F.: Muscle structure and theories of contraction. *Prog Biophys Biophys Chem* 1957; 7: 255-318
- [20] **Seidmann**, J.G. und Seidman, C.: The genetic basis for cardiomyopathy from identification to mechanistic paradigms, *Cell* 2001;104: 557-67.
- [21] **Thierfelder**, MacRae C., Watkins H. et al (1993) A familial hypertrophic cardiomyopathy locus maps to chromosome 15q2. *Proc. Natl. Acad. Sci. USA* 90: 6270-6274
- [22] **Thierfelder**, Watkins H., MacRae C et al (1994) α -Tropomyosin and cardiac troponin T mutations cause familiar hypertrophic cardiomyopathy: A disease of the sarcomere. *Cell* 77: 701-712
- [23] **Zot** AS, Potter JD. Structural aspects of troponin-tropomyosin regulation of skeletal muscle contraction. *Annu Rev Biophys Chem.* 1987;16:535–559.
- [24] **Tobacman** LS. Thin filament-mediated regulation of cardiac contraction. *Annu Rev Physiol.* 1996;58:447–481.
- [25] **Royal Prince Alfred Hospital** Sydney, Australien:: FHC mutation database. <http://www.angis.org.au/Databases/Heart/> Stand: 01. September 2007.
- [26] **Watkins** H, Anan R, Coviello DA, et al. A de novo mutation in α -Tropomyosin that causes hypertrophic cardiomyopathy. *Circulation.* 1995;91:2302–2305.
- [27] **Nakajima-Taniguchi** C, Matsui H, et al. Novel missense mutation in alpha tropomyosin gene found in Japanese patients with hypertrophic

cardiomyopathy. *J Mol Cell Cardiol.* 1995;27:2053–2058.

[28] **Yamauchi-Takahara** K, Nakajima-Taniguchi C, Matsui H, et al. Clinical implications of hypertrophic cardiomyopathy associated with mutations in the alpha-tropomyosin gene. *Heart.* 1996;76:63–65.

[29] **Coviello** DA, Maron BJ, Spirito P, et al. Clinical features of hypertrophic cardiomyopathy caused by mutation of a "hot spot" in the α -tropomyosin gene. *J Am Coll Cardiol.* 1997;29:635–640.

[30] **Watkins**, H.; Conner, D.; Thierfelder, L.; Jarcho, J. A.; MacRae, C.; McKenna, W. J.; Maron, B. J.; Seidman, J. G. und Seidman, C. E. (1995): Mutations in the cardiac myosin binding protein-C gene on chromosome 11 cause familial hypertrophic cardiomyopathy, *Nat Genet* (11) [4], Seite 434-7.

[31] **Bonne** G, Carrier L, Richard P, Hainique P, Schwartz K (1998) Familial hypertrophic cardiomyopathy. From mutations to functional defects. *Circ. Res.* 83: 580-593

[32] **Vossberg** HP, Uhl K, Mehl S, Moolman J (1998) A newly created splice donor site in the MYBP-C gene is responsible for inherited hypertrophic cardiomyopathy. *J Muscle Res Cell Motil*

[33] **CardioGenomics Program** for Genomic Applications Project 3 Boston, USA:: Mutation database. <http://genetics.med.harvard.edu/~seidman/cg3>". Stand: 01. September 2007.

[34] **Niimura** H, Bachinski LL, Sangwatanaroj S et al.(1998) Mutations in the gene for cardiac myosin-binding protein C and late-onset familial hypertrophic cardiomyopathy. *N Engl J Med* 338: 1248-1257

[35] **Anderson**, P. A.; Greig, A.; Mark, T. M.; Malouf, N. N.; Oakeley, A. E.; Ungerleider, R. M.; Allen, P. D. und Kay, B. K. (1995): Molecular basis of human cardiac troponin T isoforms expressed in the developing, adult, and failing heart, *Circ Res* (76) [4], Seite 681-6.

[36] **Mesnard**, L.; Logeart, D.; Taviaux, S.; Diriong, S.; Mercadier, J. J. und Samson, F. (1995): Human cardiac troponin T: cloning and expression of new isoforms in the normal and failing heart, *Circ Res* (76) [4], Seite 687-92.

[37] **Zot**, A. S. und Potter, J. D. (1987): Structural aspects of troponin-tropomyosin regulation of skeletal muscle contraction, *Annu Rev Biophys Chem* (16), Seite 535-59.

[38] **Jin**, J. P.; Huang, Q. Q.; Yeh, H. I. und Lin, J. J. (1992): Complete nucleotide sequence and structural organization of rat cardiac troponin T gene. A single gene generates embryonic and adult isoforms via developmentally regulated alternative splicing, *J Mol Biol* (227) [4], Seite 1269-76.

- [39] **Moolman** JC, Corfield VA, Posen B, Ngumbela K, Seidman CE, Brink PA, Watkins H (1997) Sudden death due to Troponin T mutations. *J Am Coll Cardiol* 29: 549-555
- [40] **Anan** R, Shono H, Kisanuki A, Arima S, Nakao S, Tanaka H. Patients with familial hypertrophic cardiomyopathy caused by a Phe110Ile missense mutation in the cardiac troponin T gene have variable cardiac morphologies and a favorable prognosis. *Circulation*. 1998 Aug 4;98(5):391-7.
- [41] **Saez**, L. J.; Gianola, K. M.; McNally, E. M.; Feghali, R.; Eddy, R.; Shows, T. B. und Leinwand, L. A. (1987): Human cardiac myosin heavy chain genes and their linkage in the genome, *Nucleic Acids Res* (15) [13], Seite 5443-59.
- [42] **Matsuoka**, R.; Yoshida, M. C.; Kanda, N.; Kimura, M.; Ozasa, H. und Takao, A. (1989): Human cardiac myosin heavy chain gene mapped within chromosome region 14q11.2---q13, *Am J Med Genet* (32) [2], Seite 279-84.
- [43] **Jaenicke**, T.; Diederich, K. W.; Haas, W.; Schleich, J.; Lichter, P.; Pfordt, M.; Bach, A. und Vosberg, H. P. (1990): The complete sequence of the human beta-myosin heavy chain gene and a comparative analysis of its product, *Genomics* (8) [2], Seite 194-206.
- [44] **Bonne**, G.; Carrier, L.; Richard, P.; Hainque, B. und Schwartz, K. (1998): Familial hypertrophic cardiomyopathy: from mutations to functional defects, *Circ Res* (83) [6], Seite 580-93.
- [45] **Davies** MJ, McKenna WJ (1995) Hypertrophic cardiomyopathy; Pathology and pathogenesis. *Histopathology* 26; 493-500
- [46] **Maron**, BJ. Roberts WC (1979) Quantitative analysis of cardiac muscle cell disorganisation in the ventricular septum of patients with hypertrophic cardiomyopathy: *Circulation* 59: 689 - 706
- [47] **Maron** BJ, Wolfson JK, Ebstein SE, Roberts WC (1986) Intramural (« small vessel ») coronary artery disease in hypertrophic cardiomyopathy. *J Am Coll Cardiol* 8: 545
- [48] **McKenna** WJ, Krickler DM, Goodwin JF (1984) Arrhythmias in dilated and hypertrophic cardiomyopathy. *Med. Clin. North. Am.* 68: 983-1000
- [49] **Kuhn** H, Gietzen F, Beer G. (2003) Kardiomyopathien. In: Classen M, Dierkesmann R, Heimpel H, Kock K-M, Meyer J, Müller O-A, Specker Ch, Theiss W (Hrsg) *Rationale Diagnostik und Therapie in der Inneren Medizin, Leitlinien*. Urban und Fischer 13D
- [50] **Spirito**, P. Ch., E. Seidman, W.J. McKenna, B. J. Maron: The management of hypertrophic cardiomyopathy. *N.Engl. J. Med.* 336 (1997) 775-785
- [51] **Paz** R, Jortner R, Tunick PA, Scalarovsky S, Eilat B, Perez JL, Kronzon (1996) The effect of the ingestion of ethanol on obstruction of the left

ventricular outflow tract in hypertrophic cardiomyopathy. *N Engl. J. Med* 335: 938-941

[52] **Bonow** RO, Frederick TM, Bacharach SL, et al. Atrial systole and left ventricular filling in patients with hypertrophic cardiomyopathy. *Am J Cardiol* 1983; 51:1386-1391.

[53] **Seggewiss** H, Gleichmann U, Faber L, et al. Percutaneous transluminal septal myocardial ablation in hypertrophic obstructive cardiomyopathy. Acute results and 3 month follow up in 25 patients. *J Am Coll Cardiol* 1998; 31: 252-258.

[54] **McIntosh** CL, Maron BJ, Current operative treatment of obstructive hypertrophic cardiomyopathy. *Circulation* 1988; 78: 487-495

[55] **Seiler** C, Hess OM, Schoenbeck M, et al. Long term follow up of medical versus surgical therapy for hypertrophic cardiomyopathy: A retrospective study. *J Am Coll Cardiol* 1991; 17: 634-642

[56] **Cannan** CR, Reeder GS, Bailey KR (1995) Natural history of hypertrophic cardiomyopathy: A population based study, 1976 through 1990. *Circulation* 92: 2488-2495

[57] **Spirito**, P. Ch, Chiarella F., Caratino I., (1989) Clinical course and prognosis of hypertrophic cardiomyopathy in an outpatients population. *N. Engl. J. Med.* 320: 749-755

[58] **McKenna** WJ, Deanfield J., Faruqui A. (1981) Prognosis in hypertrophic cardiomyopathy: role of age and clinical, electrocardiographic and haemodynamic features. *Am J. Cardiol.* 47: 532-538

[59] **Orita** M, Iwahana H, Kanazawa H, Hayashi K, Sekiya T. Detection of polymorphisms of human DNA by gel electrophoresis as single-strand conformation polymorphisms. *Proc Natl Acad Sci U S A* 1989; 86:2766-70.

[62] **Johns** DR, Hurko O. (nach Johns et al. 1989) Preferential amplification and molecular characterization of junction sequences of a pathogenetic deletion in human mitochondrial DNA. *Genomics.* 1989 Oct;5(3):623-8.

[63] **Mullis** KB, Faloona FA.; specific synthesis of DNA in vitro via a polymerase-catalyzed chain reaction. *Methods Enzymol.* 1987;155:335-50.

[64] **Orita** M, Iwahana H, Kanazawa H, Hayashi K, Sekiya T. Detection of polymorphisms of human DNA by gel electrophoresis as single-strand conformation polymorphisms. *Proc Natl Acad Sci U S A* 1989; 86:2766-70.

[65] **Grompe**, M.: The rapid detection of unknown mutations in nucleic acids, *Nat Genet* (5) 1993; 2: 111-117.

[66] **Cooper**, D. N., Schmidtke, J. : DNA restriction fragment length polymorphisms and heterozygosity in the human genome, *Hum Genet* (66) 1984; 1: 1-16.

[67] **Max Heiman**, Yale University New Haven, USA.: Webcutter 2.0.
<http://www.firstmarket.com/cutter/cut2.html>

[68] **Maron** BJ, Gottdiener JS, Epstein SE. Patterns and significance of distribution of left ventricular hypertrophy in hypertrophic cardiomyopathy. A wide angle, two dimensional echocardiographic study of 125 patients. *Am J Cardiol*. 1981 Sep;48(3):418-28.

[69] **McKenna** WJ, Franklin RC, Nihoyannopoulos P, Robinson KC, Deanfield JE. Arrhythmia and prognosis in infants, children and adolescents with hypertrophic cardiomyopathy. *Am Coll Cardiol*. 1988 Jan;11(1):147-53

[70] **Freeman** K, Nakao K, Leinwand LA. Low sequence variation in the gene encoding the human beta-myosin heavy chain. *Genomics*. 2001 Aug;76(1-3):73-80. .

[71] **Regitz-Zagrosek** V, Erdmann J, Wellnhofer E, Raible J, Fleck E. Novel mutation in the alpha-tropomyosin gene and transition from hypertrophic to hypocontractile dilated cardiomyopathy. *Circulation* 2000;102:112-6.

[73] **Franz** WM, Müller OJ, Katus HA. Cardiomyopathies: from genetics to the prospect of treatment. *Lancet*. 2001 Nov 10;358(9293):1627-37. Review.

[74] **Erdmann** J, Hegemann N, Weidemann A, Kallisch H, Hummel M, Hetzer R, Fleck E, Regitz-Zagrosek V. Screening the human bradykinin B2 receptor gene in patients with cardiovascular diseases: identification of a functional mutation in the promoter and a new coding variant (T21M). *Am J Med Genet*. 1998 Dec 28;80(5):521-5.

[75] **Yamauchi-Takahara** K., Nakajima- Taniguch C, Matsui H et al. (1996) Clinical implications of hypertrophic cardiomyopathy associates with mutations in the a-tropomyosin gene. *Heart* 76: 63-65

[76] **Erdmann** J, Daehmlow S, Wischke S, Senyuva M, Werner U, Raible J, Tanis N, Dyachenko S, Hummel M, Hetzer R, Regitz-Zagrosek V. Mutation Spectrum in a Large Cohort of Unrelated Consecutive Patients with Hypertrophic Cardiomyopathy. *Clin. Gen* 2003 Oct;64(4):339-349.

[77] **Cotton** RG, Scriver CR. Proof of disease causing mutation. *Hum Mutat* 1998;12:1-3.

[78] **Shaffer** EM, Rocchini AP, Spicer RL, et al. Effects of verapamil on left ventricular diastolic fillings in children with hypertrophic cardiomyopathy. *Am J cardiol* 1988; 61: 413-417

[79] **Maron** BJ. Hypertrophic cardiomyopathy in childhood. *Pediatr Clin Am N* 2004; 51: 1305

[80] **Kappenberger** L, Linde V, Daubert C, et al. Pacing in hypertrophic obstructive cardiomyopathy: A randomized crossover study. *Eur Heart J* 1997; 18: 1249-1256

[81] **Maron** BJ, Nishimura RA, McKenna WJ, et al. Assessment of permanent dual-chamber pacing as a treatment for drug-refractory symptomatic patients with obstructive hypertrophic cardiomyopathy: A randomized, double blind crossover study (M-PATHY). *Circulation* 1999;99:2927-2933

[82] **Maron** BJ, Shen W-K, Link ms, et al. Efficacy of implantable cardioverter-defibrillators for the prevention of sudden death in patients with hypertrophic cardiomyopathy. *N Engl J Med* 2000; 342; 342:365-373.

[83] **Seiler** C, Hess OM, Schoenbeck M, et al. Long term follow up of medical versus surgical therapy for hypertrophic cardiomyopathy: A retrospective study. *J Am Coll Cardiol* 1991; 17: 634-642

[84] **Spinardi**,L.;Mazars,R. et Theillet,C.: Protocols for an improved detection of point mutations by SSCP, *Nucleic Acids Res* 1991;19; 14: 4009

[85] **Condie**, A.; Eeles, R.; Borresen, A. L.; Coles, C.; Cooper, C. et al.: Detection of point mutations in the p53 gene: comparison of single-strand conformation polymorphism, constant denaturant gel electrophoresis, and hydroxylamine and osmium tetroxide techniques, *Hum Mutat* (2),1993; 1: 58-66.

[86] **Glavac**, D. et Dean, M.: Optimization of the single-strand conformation polymorphism (SSCP) technique for detection of point mutations, *Hum Mutat* (2) 1993; 5; 404-14.

[87] **Hayashi**, K. , Yandell, D. W.: How sensitive is PCR-SSCP?;*Hum Mutat* (2) 1993; 5: 338-346.

[88] **Sanger**, F.; Nicklen, S. et Coulson, A. R.: DNA sequencing with chain-terminating inhibitors, *Proc Natl Acad Sci U S A* (74) 1984; [12]: 5463-5467.

[89] **Hübl** PD. Dr. W., A-1130 Wien, med4you.at

[93] Gerull B, Osterziel KJ, Witt C. Dietz R.Thierfelder L : A rapid protocol for cardiac troponin T gene mutation detection in familial hypertrophic cardiomyopathy..*Hum Mutat.* 1998;11(2):179-82.

[94] **Seidman** CE, Seidman JG. Gene mutations that cause familial hypertrophic cardiomyopathy. In: Haber E, ed. *Molecular cardiovascular medicine*. New York: Scientific American, 1995:193-2 The New England Journal of Medicine Mar 13, 1997; 336 (11), pp 775-785

[95] **Echo in Context**, Duke University Medical Center 2002

[96] **Jae** H. Lim, Calvin E Oyer; Digital Pathology, Brown Medical School, Providence, RI 02912

[97] **Holinski-Feder**.E. Medizinisch Genetisches Zentrum Bayerstr.53, 80335 München

[98] **Wigle** ED, Rakowski H, Kimball B, Williams WG, Hypertrophic cardiomyopathy, clinical spectrum and treatment. *Circulation* 1995;92:1680-1692.

[99] **Charron** P, Dubourg O, Desnos M et al. Diagnostic value of electrocardiography and electrocardiography for familial hypertrophic cardiomyopathy in genotyped children. *Eur Heart J* 1998; 19: 1377-1382

[100] **Niimura** H, Patton KK, McKenna WJ, et al. sacromere protein gene mutations in hypertrophic cardiomyopathy of the elderly. *Circulation* 2002; 105:446-451.

[101] **Wigle** ED, Sasson Z, Henderson MA, et al. Hypertrophic cardiomyopathy: the importance of the site and the extent of hypertrophy –a review. *Prog Cardiovasc Dis* 1985; 28: 1-83.