

Literaturverzeichnis:

1. Andrade FH, Merriam AP, Guo W et al.: Paradoxical absence of M lines and downregulation of creatine kinase in mouse extraocular muscle. *J Appl Physiol.* 2003; 95: S.692-699.
2. Arad M, Seidman JG, Seidman CE: Phenotypic Diversity in Hypertrophic Cardiomyopathy. *Hum Mol Gen.* 2002; 11: S.2499-2506.
3. Blair E, Redwood C, Ashrafian H et al.: Mutations in the γ 2-subunit of AMP-activated protein kinase cause familial hypertrophic cardiomyopathy: evidence for the central role of energy compromise in disease pathogenesis. *Human Molecular Genetics.* 2001; 10: S.1215-1220.
- Mon4. Braunwald E: Heart Disease; A Textbook of Cardiovascular Medicine. 5 USA, W.B. Saunders Company, 1997 0-7216-5666-8.
5. Condie A, Eeles R, Borresen AL, Coles C, Cooper C, Prosser J: 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.* 1993; 2: S.58-66.
6. Cuda G, Fananapazir L, Epstein ND, Sellers JR: The in vitro motility activity of beta-cardiac myosin depends on the nature of the beta-myosin heavy chain gene mutation in hypertrophic cardiomyopathy. *J Muscle Res Cell Motil.* 1997; 18: S.275-283.
7. Fatkin D, Graham RM: Molecular Mechanisms of inherited Cardiomyopathies. *Physiol Rev.* 2002; 82: S.945-980.
8. Franz WM, Müller OJ, Katus HA: Cardiomyopathies: from genetics to the prospect of treatment. *The Lancet.* 2001; 358: S.1627-1636.
9. Geier C, Oezcelik C, Perrot A, Bit-Avragim N, Scheffold T, Osterziel KJ: Muscle LIM Protein: a novel disease gene for hypertrophic cardiomyopathy?. *Circulation.* 2001; 104: S.II-521.
10. Geier C, Perrot A, Özcelik C et al.: Mutations in the Human Muscle LIM Protein Gene in Families With Hypertrophic Cardiomyopathy. *Circulation.* 2003; 107: S.1390-1395.
11. Geisterfer-Lowrance AAT, Kass S, Tanigawa G et al.: A molecular basis for familial hypertrophic cardiomyopathy: a beta cardiac myosin heavy chain mutation. *Cell.* 1990; 62: S.999-1006.

12. Hayashi K, Yandell DW: How sensitive is PCR-SSCP. *Hum Mutat.* 1993; 2: S.338-346.
13. Herrmann S, Schmidt-Petersen K, Pfeifer J et al.: A polymorphism in the endothelin-A receptor gene predicts survival in patients with idiopathic dilated cardiomyopathy. *Eur Heart J.* 2001; 22: S.1948-53.
14. Himmel M, van der Ven PFN, Stöcklein W, Fürst DO et al.: The Limits of Promiscuity: Isoform-Specific Dimerization of Filamins. *Biochemistry.* 2003; 42: S.430-439.
15. Ho CY, Lever HM, DeSanctis R, Farver CF, Seidman JG, Seidman CE: Homozygous mutation in cardiac Troponin T. Implications for hypertrophic cardiomyopathy. *Circulation.* 2000; 102: S.1950-1955.
16. Hoffmann B, Schmidt-Traub H, Perrot A, Osterziel KJ, Gessner R: First Mutation in cardiac troponin C. *Hum Mutat.* 2001; 17: S.524.
17. Hunter JJ, Chien KR: Signaling Pathways for Cardiac Hypertrophy and Failure. *N Engl J Med.* 1999; 341: S.1276-1283.
18. Huxley AF: Muscle structure and theories of contraction. *Prog Biophys Biophys Chem.* 1957; 7: S.255-318.
19. Huxley HE: The Mechanism of Muscular Contraction. *Science.* 1969; 164: S.1356-1366.
20. J.G. Seidman, Christine Seidman: The Genetic Basis for Cardiomyopathy: From Mutation Identification to Mechanistic Paradigms. *Cell.* 2001; 104: S.557-567.
21. Kimura A, Harada H, Park JE et al.: Mutations in the cardiac troponin I gene associated with hypertrophic cardiomyopathy. *Nature genetics.* 1997; 16: S.379-382.
22. Lahiri DK, Nürnberg JI, Jr.: A rapid, nonenzymatic method for the preparation of HMW DNA from blood for RFLP-studies. *Nucleic Acids Res.* 1991; 19: S.5444.
23. Lahiri DK, Nürnberg JI, Jr.: A rapid non-enzymatic method for the preparation of HMW DNA from blood for RFLP studies. *Nucleic Acids Res.* 1991; 19: S.5444.
24. Lankford EB, Epstein ND, Fananapazir L, Sweeney HL: Abnormal contractile properties of muscle fibers expressing β-myosin heavy chain gene mutations in patients with hypertrophic cardiomyopathy. *J Clin Invest.* 1995; 95: S.1409-1414.
25. Lewis R.: SNPs as windows on Evolution. *The Scientist.* 2002; Jan.:
26. Li H, Linke WA, Oberhauser AF et al.: Reverse engineering of the giant muscle protein Titin. *Nature.* 2002; 418: S.998-1002.

27. Lynn RW, Taylor EW: Mechanism of adenosine triphosphate hydrolysis by actomyosin. *Biochemistry*. 1971; 10: S.4617-4624.
28. Marian AJ, Roberts R: Molecular genetic basis of hypertrophic cardiomyopathy: genetic markers for sudden cardiac death. *J Cardiovasc Electrophysiol*. 1998; 9: S.88-99.
29. Marian AJ, Roberts R: The molecular genetic basis for cardiomyopathy. *J Mol Cell Cardiol*. 2001; 33: S.655-670.
30. Marian AJ, Yu QT, Mann DL, Graham FL, Roberts R.: Expression of a mutation causing hypertrophic cardiomyopathy disrupts sarcomere assembly in adult feline cardiac myocytes. *Circulation Research*. 1995; 77: S.98-106.
31. Maron B. J.: Hypertrophic Cardiomyopathy. *Lancet*. 1997; 350: S.127-33.
32. Maron BJ: Hypertrophic Cardiomyopathy - A Systematic Review. *JAMA*. 2002; 287: S.1308-1320.
33. Maron BJ: Hypertrophic Cardiomyopathy. *Circulation*. 2002; 106: S.2419-2421.
34. Maron BJ, Gardin JM, Flack JM, Gidding SS, Kurosaki TT, Bild DE: Prevalence of hypertrophic cardiomyopathy in a general population of young adults. Echocardiographic analysis of 4111 subjects in the CARDIA study. *Circulation*. 1995; 92: S.785-789.
35. 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; 48: S.418-428.
36. Martin ER, Lai EH, Gilbert JR et al.: SNPing away at complex diseases: analysis of single-nucleotide polymorphisms around APOE in Alzheimers disease. *Am J Hum Genet*. 2000; 67: S.383-394.
37. McKenna WJ: Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of Cardiomyopathies. *Circulation*. 1996; 93: S.841.
38. McKenna WJ, Spirito P, Desnos M, Dubourg O, Komajda M: Experience from clinical genetics in hypertrophic cardiomyopathy: proposal for new diagnostic criteria in adult members of affected families. *Heart*. 1997; 77: S.130-132.
39. Nimura H, Bachinski LL, Sangwanaroj S et al.: Mutations in the gene for cardiac myosin-binding protein c and late-onset familial hypertrophic cardiomyopathy. *N Engl J Med*. 1998; 338: S.1248-1257.

40. Noguchi J, Yanagisawa M, Imamura M et al.: Complete primary structure and tissue expression of chicken pectoralis M-Protein. *J Biol Chem.* 1992; **267**: S.20302-20310.
41. Obermann WMJ, Gautel M, Weber K, Fürst DO: Molecular structure of the sarcomeric M-Band. *The EMBO Journal.* 1997; **16**: S.211-220.
42. Olson TM, Doan TP, Kishimoto NY, Whitby FG, Ackerman MJ, Fananapazir L: Inherited and de novo mutations in the cardiac actin gene cause hypertrophic cardiomyopathy. *J Mol Cell Cardiol.* 2000; **32**: S.1687-1694.
43. Roberts R, Sigwart U: New Concepts in Hypertrophic Cardiomyopathies, Part I. *Circulation.* 2001; **104**: S.2113-2116.
44. Rottbauer W, Gautel M, Zehelein J et al.: Novel splice donor site mutation in the cardiac myosin-binding protein-c gene in familial hypertrophic cardiomyopathy Characterization of cardiac transcript and protein. *J Clin Invest.* 1997; **100**: S.475-482.
45. Satoh M, Takahashi M, Sakamoto T et al.: Structural analysis of the Titin Gene in Hypertrophic Cardiomyopathy. *Biochem and Biophys Res Comm.* 1999; **262**: S.411-417.
46. Schultheiss T, Lin ZX, Lu MH et al.: Differential distribution of subsets of myofibrillar proteins in cardiac nonstriated and striated myofibrils. *J Cell Biol.* 1990; **110**: S.1159-1172.
47. Solaro RJ, Van Eyk J: Altered interactions among thin filament proteins modulate cardiac function. *J Mol Cell Cardiol.* 1996; **28**: S.217-230.
48. Steiner F, Weber K, Fürst DO: M Band Proteins myomesin and skelemin are encoded by the same gene: analysis of its organization and expression. *Genomics.* 1999; **56**: S.78-89.
49. The International SNP Map Working Group: A map of human genome sequence variation containing 1,42 million single nucleotide polymorphisms. *Nature.* 2001; **409**: S.928-933.
50. Thierfelder L , Watkins H, McRae C et al.: α -Tropomyosin and cardiac Troponin T mutations cause familial hypertrophic cardiomyopathy. *Cell.* 1994; **77**: S.710-712.
51. Tsikhovrebova L, Trinick J: Titin: Properties and Family Relationships. *Nature Reviews.* 2003; **4**: S.679-689.

52. van der Ven PFM, Wiesner S, Salmikangas P et al.: Indications for a Novel Muscular Dystrophy Pathway: γ -Filamin, the Muscle-specific Filamin Isoform, Interacts with Myotilin. J Cell Biol. 2000; 151: S.235-247.
53. Watanabe K, Muhle-Goll C, Kellermayer MSZ, Labeit S, Granzier H: Different molecular mechanics displayed by titins constitutively and differentially expressed tandem Ig segments. J Struct Biol. 2002; 137: S.248-258.
54. Watkins, McKenna, Thierfelder et al.: Mutations in the genes for cardiac troponin T and α -tropomyosin in hypertrophic cardiomyopathy. N Engl J Med. 1995; 332: S.1058-1064.

Abkürzungen

β -MHC	β -Myosin heavy chain
ADP+P	Adenosindiphosphat und Phosphor
AMP	Adenosinmonophosphat
Aqua. Dest.	Destilliertes Wasser
Arg	Arginin
Asn	Asparagin
ATP	Adenosintriphosphat
ATPase	Adenosintriphosphat spaltendes Enzym
Bp	Basenpaare
Ca^{2+}	Calcium
cAMP	Cyclo-Adenosinmonophosphat
cMyBP-C	Cardiac Myosin Binding Protein C
CTP	Cytosintriphosphat
Cys	Cystein
DCM	Dilatative Kardiomyopathie
ddNTPs	Didesoxynukleotidtriphosphate
Del	Deletion
DNA	Desoxyribonukleinsäure
dNTPs	Desoxynukleotidtriphosphate
EKG	Elektrokardiogramm
Et al.	Et alii (lat.: und andere)

FA	Formamid
gDNA	genomische DNA
Gln	Glutamin
Glu	Glutamat
Gly	Glycin
GTP	Guanintriphosphat
HCL	Salzsäure
HCM	Hypertrophe Kardiomyopathie
Kbp	Kilobasenpaare
Leu	Leucin
Lys	Lysin
MDE	Mutation Detection Enhancment
Met	Methionin
MgCl ₂	Magnesiumchlorid
MM	Millimol
My1-13	Bezeichnung der Myomesin-Domänen
NaCl	Natriumchlorid
PAA	Polyacrylamid
PCR	Polymerasekettenreaktion
Phe	Phenylalanin
Phe	Phenylalanin
SDS	Sodiumdisulfat
Ser	Serin
SNPs	Single Nucleotide Polymorphisms
SP-reiche Domäne	Serin/Prolin-reiche Domäne
SSCP	Single Strand Conformation Polymorphism
Trp	Tryptophan
Trp	Tryptophan
TTP	Thymintriphosphat
U/min	Umdrehungen pro Minute
Val	Valin