**Erweiterte Literaturliste**

Vasen, H.F., et al., The International Collaborative Group on Hereditary Non-Polyposis Colorectal Cancer (ICG-HNPCC). Dis Colon Rectum, 1991. 34(5): p. 424-5.

Rodriguez-Bigas, M.A., et al., A National Cancer Institute Workshop on Hereditary Nonpolyposis Colorectal Cancer Syndrome: meeting highlights and Bethesda guidelines. J Natl Cancer Inst, 1997. 89(23): p. 1758-62.

Schulmann, K., et al., Small bowel cancer risk in Lynch syndrome. Gut, 2008. 57(11): p. 1629- 30.

Umar, A., et al., Revised Bethesda Guidelines for hereditary nonpolyposis colorectal cancer (Lynch syndrome) and microsatellite instability. J Natl Cancer Inst, 2004. 96(4): p. 261-8

Levine, A.J., et al., Cancer risks for the relatives of colorectal cancer cases with a methylated MLH1 promoter region: data from the Colorectal Cancer Family Registry. Cancer Prev Res (Phila), 2012. 5(2): p. 328-35

Foulkes, W.D., A tale of four syndromes: familial adenomatous polyposis, Gardner syndrome, attenuated APC and Turcot syndrome. QJM, 1995. 88(12): p. 853-63.

Lynch, H.T., et al., Attenuated familial adenomatous polyposis (AFAP). A phenotypically and genotypically distinctive variant of FAP. Cancer, 1995. 76(12): p. 2427-33.

Lynch, H.T. and T.C. Smyrk, Classification of familial adenomatous polyposis: a diagnostic nightmare. Am J Hum Genet, 1998. 62(6): p. 1288-9.

Soravia, C., et al., Genotype-phenotype correlations in attenuated adenomatous polyposis coli. Am J Hum Genet, 1998. 62: p. 1290-1301.

Hernegger, G.S., H.G. Moore, and J.G. Guillem, Attenuated familial adenomatous polyposis: an evolving and poorly understood entity. Dis Colon Rectum, 2002. 45(1): p. 127-34; discussion 134-6.

Knudsen, A.L., M.L. Bisgaard, and S. Bulow, Attenuated familial adenomatous polyposis (AFAP). A review of the literature. Fam Cancer, 2003. 2(1): p. 43-55.

Al-Tassan, N., et al., Inherited variants of MYH associated with somatic G:C-->T:A mutations in colorectal tumors. Nat Genet, 2002. 30(2): p. 227-32.

Cao, Y., et al., Challenge in the differentiation between attenuated familial adenomatous polyposis and hereditary nonpolyposis colorectal cancer: case report with review of the literature. Am J Gastroenterol, 2002. 97(7): p. 1822-7.

Aretz, S., M. Genuardi, and F.J. Hes, Clinical utility gene card for: MUTYH-associated polyposis (MAP), autosomal recessive colorectal adenomatous polyposis. 2012, Eur J Hum Genet.

Sampson, J.R. and N. Jones, MUTYH-associated polyposis. Best Pract Res Clin Gastroenterol, 2009. 23(2): p. 209-18.

Morreau, H., R. Riddel, and S. Aretz, MUTYH-associated polyposis., in WHO Classification of Tumours of the Digestive System, F.T. Bosman, et al., Editors. 2010, IARC/ WHO Press: Lyon. p. 156-159

Aretz, S., et al., MUTYH-associated polyposis: 70 of 71 patients with biallelic mutations present with an attenuated or atypical phenotype. Int J Cancer, 2006. 119(4): p. 807-14

Lubbe, S.J., et al., Clinical implications of the colorectal cancer risk associated with MUTYH mutation. J Clin Oncol, 2009. 27(24): p. 3975-80.

Vogt, S., et al., Expanded extracolonic tumor spectrum in MUTYH-associated polyposis. Gastroenterology, 2009. 137(6): p. 1976-85 e1-10

Aarnio, M., et al., Life-time risk of different cancers in hereditary non-polyposis colorectal cancer (HNPCC) syndrome. Int J Cancer, 1995. 64(6): p. 430-3.

Jarvinen, H.J., et al., Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. Gastroenterology, 2000. 118(5): p. 829-34

Edelstein, D.L., et al., Rapid development of colorectal neoplasia in patients with Lynch syndrome. Clin Gastroenterol Hepatol, 2011. 9(4): p. 340-3

Engel, C., et al., Efficacy of annual colonoscopic surveillance in individuals with hereditary nonpolyposis colorectal cancer. Clin Gastroenterol Hepatol, 2010. 8(2): p. 174-82.

Vasen, H.F., et al., MSH2 mutation carriers are at higher risk of cancer than MLH1 mutation carriers: a study of hereditary nonpolyposis colorectal cancer families. J Clin Oncol, 2001. 19(20): p. 4074-80.

Aarnio, M., et al., Cancer risk in mutation carriers of DNA-mismatch-repair genes. Int J Cancer, 1999. 81(2): p. 214-8.

Vasen, H.F., et al., Guidelines for the clinical management of Lynch syndrome (hereditary nonpolyposis cancer). J Med Genet, 2007. 44(6): p. 353-62.

Schulmann, K., et al., Small bowel cancer risk in Lynch syndrome. Gut, 2008. 57(11): p. 1629- 30

Schulmann, K., et al., HNPCC-associated small bowel cancer: clinical and molecular characteristics. Gastroenterology, 2005. 128(3): p. 590-9.

de Vos tot Nederveen Cappel, W.H., et al., Surveillance for hereditary nonpolyposis colorectal cancer: a long-term study on 114 families. Dis Colon Rectum, 2002. 45(12): p. 1588-94.

Church, J.M., et al., Teenagers with familial adenomatous polyposis: what is their risk for colorectal cancer? Dis Colon Rectum, 2002. 45(7): p. 887-9.

King, J.E., et al., Care of patients and their families with familial adenomatous polyposis. Mayo Clin Proc, 2000. 75(1): p. 57-67.

Bertario, L., et al., Causes of death and postsurgical survival in familial adenomatous polyposis: results from the Italian Registry. Italian Registry of Familial Polyposis Writing Committee. Semin Surg Oncol, 1994. 10(3): p. 225-34.

Galle, T.S., K. Juel, and S. Bulow, Causes of death in familial adenomatous polyposis. Scand J Gastroenterol, 1999. 34(8): p. 808-12.

Heiskanen, I., T. Luostarinen, and H.J. Jarvinen, Impact of screening examinations on survival in familial adenomatous polyposis. Scand J Gastroenterol, 2000. 35(12): p. 1284-7.

Vasen, H.F., et al., Decision analysis in the surgical treatment of patients with familial adenomatous polyposis: a Dutch-Scandinavian collaborative study including 659 patients. Gut, 2001. 49(2): p. 231-5.

Bulow, S., Results of national registration of familial adenomatous polyposis. Gut, 2003. 52(5): p. 742-6

Bulow, S., Clinical features in familial polyposis coli. Results of the Danish Polyposis Register. Dis Colon Rectum, 1986. 29(2): p. 102-7.

De Cosse, J.J., et al., Rectal cancer risk in patients treated for familial adenomatous polyposis. The Leeds Castle Polyposis Group. Br J Surg, 1992. 79(12): p. 1372-5.

Nyam, D.C., et al., Ileal pouch-anal canal anastomosis for familial adenomatous polyposis: early and late results. Ann Surg, 1997. 226(4): p. 514-9; discussion 519-21.

Parc, Y.R., et al., Familial adenomatous polyposis: results after ileal pouch-anal anastomosis in teenagers. Dis Colon Rectum, 2000. 43(7): p. 893-8; discussion 898-902.

Van Duijvendijk, P., et al., Quality of life after total colectomy with ileorectal anastomosis or proctocolectomy and ileal pouch-anal anastomosis for familial adenomatous polyposis. Br J Surg, 2000. 87(5): p. 590-6.

Church, J., et al., Risk of rectal cancer in patients after colectomy and ileorectal anastomosis for familial adenomatous polyposis: a function of available surgical options. Dis Colon Rectum, 2003. 46(9): p. 1175-81.

Spigelman, A.D., et al., Upper gastrointestinal cancer in patients with familial adenomatous polyposis. Lancet, 1989. 2(8666): p. 783-5.

Bulow, S., et al., Duodenal adenomatosis in familial adenomatous polyposis. Gut, 2004. 53(3): p. 381-6.

Kadmon, M., A. Tandara, and C. Herfarth, Duodenal adenomatosis in familial adenomatous polyposis coli. A review of the literature and results from the Heidelberg Polyposis Register. Int J Colorectal Dis, 2001. 16(2): p. 63-75.

Norton, I.D., et al., Safety and outcome of endoscopic snare excision of the major duodenal papilla. Gastrointest Endosc, 2002. 56(2): p. 239-43.

Bulow, C. and S. Bulow, Is screening for thyroid carcinoma indicated in familial adenomatous polyposis? The Leeds Castle Polyposis Group. Int J Colorectal Dis, 1997. 12(4): p. 240-2.

Jarrar, A.M., et al., Screening for thyroid cancer in patients with familial adenomatous polyposis. Ann Surg, 2011. 253(3): p. 515-21.

Martayan, A., et al., Gene variants associated to malignant thyroid disease in familial adenomatous polyposis: a novel APC germline mutation. J Endocrinol Invest, 2010. 33(9): p. 603-6.

Herraiz, M., et al., Prevalence of thyroid cancer in familial adenomatous polyposis syndrome and the role of screening ultrasound examinations. Clin Gastroenterol Hepatol, 2007. 5(3): p. 367-73.

Hernegger, G.S., H.G. Moore, and J.G. Guillem, Attenuated familial adenomatous polyposis: an evolving and poorly understood entity. Dis Colon Rectum, 2002. 45(1): p. 127-34; discussion 134-6.

Leggett, B.A., et al., Severe upper gastrointestinal polyposis associated with sparse colonic polyposis in a familial adenomatous polyposis family with an APC mutation at codon 1520. Gut, 1997. 41(4): p. 518-21.

Zwick, A., et al., Gastric adenocarcinoma and dysplasia in fundic gland polyps of a patient with attenuated adenomatous polyposis coli. Gastroenterology, 1997. 113(2): p. 659-63.

Hofgartner, W.T., et al., Gastric adenocarcinoma associated with fundic gland polyps in a patient with attenuated familial adenomatous polyposis. Am J Gastroenterol, 1999. 94(8): p. 2275-81.

Nascimbeni, R., et al., Rectum-sparing surgery may be appropriate for biallelic MutYHassociated polyposis. Dis Colon Rectum, 2010. 53(12): p. 1670-5.

Vogt, S., et al., Expanded extracolonic tumor spectrum in MUTYH-associated polyposis. Gastroenterology, 2009. 137(6): p. 1976-85 e1-10.

Zbuk, K.M. and C. Eng, Hamartomatous polyposis syndromes. Nat Clin Pract Gastroenterol Hepatol, 2007. 4(9): p. 492-502.

Aretz, S., et al., High proportion of large genomic STK11 deletions in Peutz-Jeghers syndrome. Hum Mutat, 2005. 26(6): p. 513-9.

Hinds, R., et al., Complications of childhood Peutz-Jeghers syndrome: implications for pediatric screening. J Pediatr Gastroenterol Nutr, 2004. 39(2): p. 219-20.

Mehenni, H., et al., Cancer risks in LKB1 germline mutation carriers. Gut, 2006. 55(7): p. 984- 90.

Giardiello, F.M., et al., Very high risk of cancer in familial Peutz-Jeghers syndrome. Gastroenterology, 2000. 119(6): p. 1447-53.

Hearle, N., et al., Frequency and spectrum of cancers in the Peutz-Jeghers syndrome. Clin Cancer Res, 2006. 12(10): p. 3209-15.

Bonadona V, Bonaiti B, Olschwang S, et al. Cancer risks associated with germline mutations in MLH1, MSH2, and MSH6 genes in Lynch syndrome. JAMA 2011;305:2304-2310.

Møller P, Seppälä TT, Bernstein I, et al. Cancer risk and survival in path\_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut 2018;67:1306-1316.

Ryan NAJ, Morris J, Green K, et al. Association of mismatch repair mutation with age at cancer onset in Lynch syndrome: Implications for stratified surveillance strategies. JAMA Oncol 2017;3:1702-1706.

Møller P, Seppälä T, Bernstein I, et al. Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. Gut 2017;66:464-472

Barrow E, Robinson L, Alduaij W, et al. Cumulative lifetime incidence of extracolonic cancers in Lynch syndrome: a report of 121 families with proven mutations. Clin Genet 2009;75(2):141-149.

Dominguez-Valentin M, Sampson J, Seppälä T, et al. Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genet Med 2020;22:15-25.

Schulmann K, Brasch FE, Kunstmann E, Engel C, Pagenstecher C, Vogelsang H, Kruger S, Vogel T, Knaebel HP, RuschoffJ, Hahn SA, Knebel-Doeberitz MV, Moeslein G, Meltzer SJ, Schackert HK, Tympner C, Mangold E, Schmiegel W. HNPCC-associated small bowel cancer: clinical and molecular characteristics. Gastroenterology. 2005;128:590–9. PubMed PMID: 15765394.

Watson P, Vasen HF, Mecklin JP, Bernstein I, Aarnio M, Järvinen HJ, Myrhøj T, Sunde L, Wijnen JT, Lynch HT. The risk of extra-colonic, extra-endometrial cancer in the Lynch syndrome. Int J Cancer. 2008;123:444–9

Rothwell PM, Fowkes FG, Belch JF, Ogawa H, Warlow CP, Meade TW. Effect of daily aspirin on long-term randomized trials. Lancet. 2011;377:31–41.

Nieuwenhuis MH, Casparie M, Mathus-Vliegen LM, Dekkers OM, Hogendoorn PC, Vasen HF. A nation-wide study comparing sporadic and familial adenomatous polyposis-related desmoid-type fibromatoses. Int J Cancer. 2011a;129:256–61

Sinha A, Tekkis PP, Gibbons DC, Phillips RK, Clark SK. Risk factors predicting desmoid occurrence in patients with familial adenomatous polyposis: a meta-analysis. Colorectal Dis. 2011;13:1222–9.

Neklason DW, Stevens J, Boucher KM, Kerber RA, Matsunami N, Barlow J, Mineau G, Leppert MF, Burt RW. American founder mutation for attenuated familial adenomatous polyposis. Clin Gastroenterol Hepatol. 2008;6:46–52.

Giardiello FM, Brensinger JD, Petersen GM, Luce MC, Hylind LM, Bacon JA, Booker SV, Parker RD, Hamilton SR. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. N Engl J Med. 1997;336:823–7.

Sieber OM, Lipton L, Crabtree M, Heinimann K, Fidalgo P, Phillips RK, Bisgaard ML, Orntoft TF, Aaltonen LA, Hodgson SV, Thomas HJ, Tomlinson IP. Multiple colorectal adenomas, classic adenomatous polyposis, and germ- line mutations in MYH. N Engl J Med. 2003;348:791–9.

Aretz S, Uhlhaas S, Goergens H, Siberg K, Vogel M, Pagenstecher C, Mangold E, Caspari R, Propping P, Friedl W. MUTYH-associated polyposis: 70 of 71 patients with biallelic mutations present with an attenuated or atypical phenotype. Int J Cancer. 2006;119:807–14

Syngal S, Brand RE, Church JM, Giardiello FM, Hampel HL, Burt RW, et al. ACG clinical guideline: Genetic testing and management of hereditary gastrointestinal cancer syndromes. American College of Gastroenterology. Available [online](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4695986/?report=reader). 2015. Accessed 4-30-20.