

DAFTAR PUSTAKA

Aaltonen LA, Peltomaki P, Leach FS, et al. 1993. Clues to the pathogenesis of familial colorectal cancer. *Science* 260: 812-6.

Aaltonen LA, Peltomaki P, Mecklin JP, et al. 1994. Replication errors in benign and malignant tumors from hereditary nonpolyposis colorectal cancer patients. *Cancer Res* 54: 1645.

Aaltonen LA, Salovaara R, Kristo P, et al. 1998. Incidence of hereditary nonpolyposis colorectal cancer and the feasibility of molecular screening for the disease. *N Engl J Med* 338: 1481.

Aarnio M, Sankila R, Pukkala E, et al. 1999. Cancer risk in mutation carriers of DNA mismatch-repair genes. *Int J Cancer* 81: 214.

Ahlquist DA, Sargent DJ, Loprinzi CL, et al. 2008. Stool DNA and occult blood testing for screen detection of colorectal neoplasia. *Ann Intern Med* 149: 441-50.

Akhurst RJ, Balmain A. 1999. Genetic events and the role of TGF beta in epithelial tumour progression. *J Pathol* 187: 82.

Aktas H, Cai H, Cooper GM. 1997. Ras links growth factor signaling to the cell cycle machinery via regulation of cyclin D1 and the Cdk inhibitor p27KIP1. *Mol Cell Biol* 17 :3850.

Al-Tassan N, Chmiel NH, Maynard J, et al. 2002. Inherited variants of MYH associated with somatic G:C→T:A mutations in colorectal tumors. *Nat Genet* 30: 227-32.

Amado RG, Wolf M, Peeters M, et al. 2008. Wild-type KRAS is required for panitumumab efficacy in patients with metastatic colorectal cancer. *J Clin Oncol* 26 :1626-34.

American Cancer Society: *Cancer Fact and Figures 2020*. Atlanta, Ga: American Cancer Society, 2020. January 17, 2020).

Andre T, Boni C, Mounedji-Boudiaf L, et al. 2004. Oxaliplatin, fluorouracil, and leucovorin as adjuvant treatment for colon cancer. *N Engl J Med* 350 :2343-51.

Aoki Y, Niihori T, Narumi Y, Kure S, Matsubara Y. 2008. The RAS/MAPK syndromes: novel roles of the RAS pathway in human genetic disorders. *Hum Mutat* 29:992-1006.

Aplin AE, Howe A, Alahari SK, and Juliano RL. 1998. Signal transduction and signal modulation by cell adhesion receptors: the role of integrins, cadherins,

immunoglobulin-cell adhesion molecules, and selectins. *Pharmacol. Rev.* 50: 197–263.

Arber N, Eagle CJ, Spicak J, et al. 2006. Celecoxib for the prevention of colorectal adenomatous polyps. *N Engl J Med* 355: 885-95.

Backlund MG, Mann JR, Holla VR, et al. 2005. 15-Hydroxyprostaglandin dehydrogenase is down-regulated in colorectal cancer. *J Biol Chem* 280:3217-23.

Baker SJ, Fearon ER, Nigro JM, et al. 1989. Chromosome 17 deletions and p53 gene mutations in colorectal carcinomas. *Science* 244:217-21.

Baker SJ, Markowitz S, Fearon ER, Willson JK, Vogelstein B. 1990. Suppression of human colorectal carcinoma cell growth by wild-type p53. *Science* 249:912-5.

Baker SJ, Preisinger AC, Jessup JM, et al. 1990. p53 Gene mutations occur in combination with 17p allelic deletions as late events in colorectal tumorigenesis. *Cancer Res* 50:7717-22.

Barault L, Charon-Barra C, Jooste V, et al. 2008. Hypermethylator phenotype in sporadic colon cancer: study on a population-based series of 582 cases. *Cancer Res* 68: 8541-6.

Barber TD, McManus K, Yuen KW, et al. 2008. Chromatoid cohesion defects may underlie chromosome instability in human colorectal cancers. *Proc Natl Acad Sci U S A* 105:3443-8.

Baron JA, Cole BF, Sandler RS, et al. 2003. A randomized trial of aspirin to prevent colorectal adenomas. *N Engl J Med* 348:891-9.

Baughman Jr FA, List CF, Williams JR, et al. 1969. The glioma-polyposis syndrome. *N Engl J Med* 281:1345.

Baylin SB, Belinsky SA, Herman JG. 2000. Aberrant methylation of gene promoters in cancer: concepts, misconcepts, and promise. *J Natl Cancer Inst* 92:1460.

Beacham CH, Shields HM, Raffensperger EC, Enterline HT. 1978. Juvenile and adenomatous gastrointestinal polyposis. *Am J Dig Dis* 23:1137

Belliveau P, Graham AM. 1984. Mesenteric desmoid tumor in Gardner's syndrome treated by sulindac. *Dis Colon Rectum* 27:53.

Bennett RL, French KS, Resta RG, Doyle DL. Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National

Society of Genetic Counselor. *J Genet Counsel* 2008; 17: 424-33. doi: 10.1007/s10897-008-9169-9.

Bergers G, Hanahan D, Coussens LM. 1998. Angiogenesis and apoptosis are cellular parameters of neoplastic progression in transgenic mouse models of tumorigenesis. *Int. J. Dev. Biol.* 42: 995–1002.

Bertagnolli MM, Eagle CJ, Zauber AG, et al. 2006. Celecoxib for the prevention of sporadic colorectal adenomas. *N Engl J Med* 355:873-84.

Bertagnolli MM, Niedzwiecki D, Compton CC, et al. 2009. Microsatellite instability predicts improved response to adjuvant therapy with irinotecan, fluorouracil, and leucovorin in stage III colon cancer: Cancer and Leukemia Group B Protocol 89803. *J Clin Oncol* 27: 1814-21.

Bertagnolli MM, Warren RS, Niedzwiecki D, et al. 2009. p27Kip1 in stage III colon cancer: implications for outcome following adjuvant chemotherapy in Cancer and Leukemia Group B protocol 89803. *Clin Cancer Res* 15:2116-22.

Bertario L, Russo A, Radice P, et al. 2000. Genotype and phenotype factors as determinants for rectal stump cancer in patients with familial adenomatous polyposis. *Hereditary Colorectal Tumors Registry. Ann Surg* 231:538.

Biesecker LG, Green RC. Diagnostic Clinical Genome and exome Sequencing. *N Engl J Med.* 2014 Jun 19;370(25):2418-25. doi: 10.1056/NEJMra1312543.

Bisgaard ML, Fenger K, Bulow S, et al. 1994. Familial adenomatous polyposis (FAP): frequency, penetrance, and mutation rate. *Hum Mutat* 3:121.

Bjork J, Akerbrant H, Iselius L, et al. 2001. Periampullary adenomas and adenocarcinomas in familial adenomatous polyposis: cumulative risks and APC gene mutations. *Gastroenterology* 121:1127.

Blair NP, Trempe CL. 1980. Hypertrophy of the retinal pigment epithelium associated with Gardner's syndrome. *Am J Ophthalmol* 90:661.

Boland CR, Koi M, Chang DK, Carethers JM. 2008. The biochemical basis of microsatellite instability and abnormal immunohistochemistry and clinical behavior in Lynch syndrome: from bench to bedside. *Fam Cancer* 7:41-52.

Boland CR, Thibodeau SN, Hamilton SR, et al. 1998. A National Cancer Institute workshop on microsatellite instability for cancer detection and familial predisposition: development of international criteria for the determination of microsatellite instability in colorectal cancer. *Cancer Res* 58:5248.

Boland CR. 1997. Genetic pathways to colorectal cancer. *Hosp Pract (Off Ed)* 32:79.

Boman BM, Huang E. 2008. Human colon cancer stem cells: a new paradigm in gastrointestinal oncology. *J Clin Oncol* 26:2828-38.

Bos JL, Fearon ER, Hamilton SR, et al. 1987. Prevalence of ras gene mutations in human colorectal cancers. *Nature* 327:293-7.

Bougatef K, Ouerhani S, Moussa A, Kourda N, Coulet F, Colas C et al. 2008. Prevalence of mutation in APC, CTNNB1 and BRAF in Tunisian patients with sporadic colorectal cancer. *Cancer Genetic and Cytogenetic* 187: 12-8.

Brock JAK, Allen VM, Kieser K, Langlois S. Family History Screening: Use of Three Generation Pedigree in Clinical Practice. *J Obstet Gynaecol Can* 2010;32(7): 663-72. [https://doi.org/10.1016/S1701-2163\(16\)34570-4](https://doi.org/10.1016/S1701-2163(16)34570-4)

Broderick P, Carvajal-Carmona L, Pittman AM, et al. 2007. A genome-wide association study shows that common alleles of SMAD7 influence colorectal cancer risk. *Nat Genet* 39:1315-7.

Bronner CE, Baker SM, Morrison PT, et al. 1994. Mutation in the DNA mismatch repair gene homologue hMLH1 is associated with hereditary non-polyposis colon cancer. *Nature* 368:258-61.

Brosens LAA, Offerhaus GJA, Giardiello M. *Surg Clin North Am.* 2015;95(5); 1067-80). Doi: 10.1016/j.suc.2015.05.004.

Buckingham L. Molecular Detection of Inherited Diseases. In: Buckingham L, Flaws M. *Molecular Diagnostic. Fundamental , Methods, & Clinical Applications.* F.A. Davis Company 4th eds. Philadelphia 2011; 310-331. ISBN-13: 978-0803668294 ISBN-10: 0803668295 www.amazon.com DOI: <https://doi.org/10.29074/ascls.21.3.184>

Bulow S, Sondergaard JO, Witt I, et al. 1984. Mandibular osteomas in familial polyposis coli. *Dis Colon Rectum* 27:105.

Burdick D, Prior JT. 1982. Peutz-Jeghers syndrome: a clinicopathologic study of a large family with a 27-year follow-up. *Cancer* 50:2139.

Burn J, Chapman P, Delhanty J, et al. 1991. The UK Northern region genetic register for familial adenomatous polyposis coli: use of age of onset, congenital

hypertrophy of the retinal pigment epithelium, and DNA markers in risk calculations. *J Med Genet* 28:289.

Burt R et al. 1995. Genetic of Colon Cancer: Impact of inheritance on colon cancer risk. *Ann Rev Med* 46: 371-9.

Burt RW, Barthel JS, Dunn KB, David DS, Drelichman E, Ford JM et al. Colon Cancer Screening. *Journal of the National Comprehensive Cancer Network*. 2010;8(1): 8-56.

Burt RW. Colon Cancer Screening. *Gastroenterology* 2000;119:837-53. Doi:10.1053/gast.2000.16508 Burt RW. Colon Cancer Screening. *Gastroenterology* 2000;119:837-53. Doi:10.1053/gast.2000.16508

Bussey H. 1975. Familial polyposis coli. Baltimore: Johns Hopkins Press

Bussey HJ, Veale AM, Morson BC. 1978. Genetics of gastrointestinal polyposis. *Gastroenterology* 74:1325.

Byers T. 1997. American Cancer Society guidelines for screening and surveillance for early detection of colorectal polyps and cancer. *CA Cancer J Clin* 47: 154-60.

Cannon-Albright L, Skolnick M, Bishop T, Lee R, Burt R. 1988. Common inheritance of susceptibility to colonic adenomatous polyps and associated colorectal cancers. *N Engl J Med* 319:533-7.

Cha YI, DuBois RN. 2007. NSAIDs and cancer prevention: targets downstream of COX-2. *Annu Rev Med* 58:239-52.

Chan AT, Ogino S, Fuchs CS. 2007. Aspirin use and risk of colorectal cancer in relation to the expression of COX-2. *N Engl J Med* 356:2131-42.

Chen WD, Han ZJ, Skoletsy J, et al. 2005. Detection in fecal DNA of colon cancerspecific methylation of the nonexpressed vimentin gene. *J Natl Cancer Inst* 97:1124-32.

Chin L, Andersen JN, Futreal PA. Cancer genomics: from discovery science to personalized medicine. *Nature Medicine* 2011; 17: 297-303. doi: 10.1038/nm.2323.

Chung DC. 2000. The genetic basis of colorectal cancer: insights into critical pathways of tumorigenesis. *Gastroenterology* 119:854.

Compton C, Hawk ET, Grochow L, Lee F, Ritter M, Niederhuber JE. 2008. Colon cancer. In: Abeloff MD, Armitage J, Niederhuber JE, Kastan MB, McKenna GW, eds. *Abeloff's clinical oncology*. Philadelphia: Churchill Livingstone 1477-534.

Cordon-Cardo C, Prives C. 1999. At the crossroads of inflammation and tumorigenesis. *J. Exp Med* 190:1367–70.

Coussens LM, Raymond WW, Bergers G, Laig-Webster M, Behrendtsen O, Werb Z, Caughey GH, Hanahan D. 1999. Inflammatory mast cells up-regulate angiogenesis during squamous epithelial carcinogenesis. *Genes Dev* 13:1382–97.

Cunningham D, Humblet Y, Siena S, et al. 2004. Cetuximab monotherapy and cetuximab plus irinotecan in irinotecan-refractory metastatic colorectal cancer. *N Engl J Med* 351:337-45.

Dalerba P, Dylla SJ, Park IK, et al. 2007. Phenotypic characterization of human colorectal cancer stem cells. *Proc Natl Acad Sci USA* 104:10158-63.

Debinski HS, Love S, Spigelman AD, Phillips RK. 1996. Colorectal polyp counts and cancer risk in familial adenomatous polyposis. *Gastroenterology* 110:1028.

Debinski HS, Spigelman AD, Hatfield A, et al. 1995. Upper intestinal surveillance in familial adenomatous polyposis. *Eur J Cancer* 31A:1149.

Di Nicolantonio F, Martini M, Molinari F, et al. 2008. Wild-type BRAF is required for response to panitumumab or cetuximab in metastatic colorectal cancer. *J Clin Oncol* 26:5705-12.

Dionigi G, Bianchi V, Villa F, Rovera F, Boni L, Annoni M et al. 2007. Differences between familial and sporadic forms of colorectal cancer with DNA microsatellite instability. *Surgical Oncology* 16: 537-42.

Downward J. 1998. Mechanisms and consequences of activation of protein kinase B/Akt. *Curr. Opin. Cell Biol* 10:262–7.

Entius MM, Keller JJ, Westerman AM, et al. 2001. Molecular genetic alterations in hamartomatous polyps and carcinomas of patients with Peutz-Jeghers syndrome. *J Clin Pathol* 54:126.

Entius MM, Westerman AM, van Velthuysen ML, et al. 1999. Molecular and phenotypic markers of hamartomatous polyposis syndromes in the gastrointestinal tract. *HepatoGastroenterology* 46:661.

Eppert K, Scherer SW, Ozcelik H, et al. 1996. MADR2 maps to 18q21 and encodes a TGF β -regulated MAD-related protein that is functionally mutated in colorectal carcinoma. *Cell* 86:543-52.

Esplin ED, Snyder MP. *World J Clin Oncol.* 2014 10; 5(5): 1036-47. Doi: 10.5306/wjco.v5.i5.1036

Evan G. 1998. A Matter of Life and Cell Death. *Science* 281:1317-22.

Fearon ER, Bommer GT. 2008. Molecular biology of colorectal cancer. In: DeVita VT Jr, Lawrence TS, Rosenberg SA, eds. *De-Vita, Hellman, and Rosenberg's cancer: principles & practice of oncology*. Vol. 1. Philadelphia: Lippincott Williams & Wilkins 1218-31.

Fearon ER, Cho KR, Nigro JM, et al. 1990. Identification of a chromosome 18q gene that is altered in colorectal cancers. *Science* 247:49.

Fearon ER, Vogelstein B. 1990. A genetic model for colorectal tumorigenesis. *Cell* 61:759.

Fedi P, Tronick SR, and Aaronson SA. 1997. Growth factors. In: Holland JF, Bast RC, Morton DL, E. Frei DW. *Cancer Medicine*. Kufe, Weichselbaum RR (eds). Baltimore MD: Williams and Wilkins 41–64.

Fishel R, Lescoe MK, Rao MR, et al. 1993. The human mutator gene homolog MSH2 and its association with hereditary nonpolyposis colon cancer. *Cell* 75:1027-38. [Erratum, *Cell* 1994;77:167.]

Fletcher JA, Pinkus JL, Lage JM, et al. 1992. Clonal 6p21 rearrangement is restricted to the mesenchymal component of an endometrial polyp. *Genes Chromosomes Cancer* 5:260.

Foley TR, McGarrity TJ, Abt AB. 1988. Peutz-Jeghers syndrome: a clinicopathologic survey of the "Harrisburg family" with a 49-year follow-up. *Gastroenterology* 95:1535.

Frayling IM, Beck NE, Ilyas M, et al. 1998. The APC variants I1307K and E1317Q are associated with colorectal tumors, but not always with a family history. *Proc Natl Acad Sci USA* 95:10722.

Giancotti FG, and Ruoslahti E. 1999. Integrin signaling. *Science* 285:1028–1032.

Giardiello FM, Brensinger JD, Tersmette AC, et al. 2000. Very high risk of cancer in familial Peutz-Jeghers syndrome. *Gastroenterology* 119:1447.

Goel A, Arnold CN, Boland CR. 2001. Multistep progression of colorectal cancer in the setting of microsatellite instability: new details and novel insights. *Gastroenterology* 121:1497.

Goss KH, Groden J. 2000. Biology of the adenomatous polyposis coli tumor suppressor. *J Clin Oncol* 18:1967-79.

Grady WM, Markowitz S. 2002. Colorectal cancer: genetic alterations. In: Kelsen D, Daly J, Kern S, Levin B, Tepper J, eds. *Gastrointestinal oncology: principles and practice*. Philadelphia: Lippincott Williams & Wilkins 685-702.

Grady WM, Markowitz SD. 2008. TGF- β signaling pathway and tumor suppression. In: Derynck R, Miyazano K, eds. *The TGF- β family*. Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press 889-938.

Grady WM, Myeroff LL, Swinler SE, et al. 1999. Mutational inactivation of transforming growth factor β receptor type II in microsatellite stable colon cancers. *Cancer Res* 59:320-4.

Green SE, Bradburn DM, Varma JS, Burn J. 1998. Hereditary non-polyposis colorectal cancer. *Int J Colorectal Dis* 13:3.

Groden J, Thliveris A, Samowitz W, et al. 1991. Identification and characterization of the familial adenomatous polyposis coli gene. *Cell* 66:589.

Grotsky HW, Rickert RR, Smith WD, Newsome JF. 1982. Familial juvenile polyposis coli: a clinical and pathologic study of a large kindred. *Gastroenterology* 82:494.

Gruber SB, Entius MM, Petersen GM, et al. 1998. Pathogenesis of adenocarcinoma in Peutz-Jeghers syndrome. *Cancer Res* 58:5267.

Guan RJ, Fu Y, Holt PR, Pardee AB. 1999. Association of K-ras mutations with p16 methylation in human colon cancer. *Gastroenterology* 116:1063.

Gurbuz AK, Giardiello FM, Petersen GM, et al. 1994. Desmoid tumours in familial adenomatous polyposis. *Gut* 35:377.

Guttmacher AE, Collins FS, Carmona RH. Family History-More Important Than Ever. *N Engl J Med*. 2016; 351: 2333-6. www.nejm.org
doi:10.1056/NEJMs042979

Hahn WC, Counter CM, Lundberg AS, Beijersbergen, Lbrooks RMW, and Weinberg RA. 1999. Creation of human tumor cells with defined genetic elements. *Nature* 440: 464–468.

Hamilton SR, Liu B, Parsons RE, et al. 1995. The molecular basis of Turcot's syndrome. *N Engl J Med* 332:839.

Hamosh A, McInnes RR, Nussbaum RL, Willard HF, Lisi EC, Sobreira N. Lynch syndrome (DNA Mismatch Repair Gene Mutation, MM 120435. *Clinical Case Studies Illustrating Genetic principles*. In: *Genetics in Medicine*. 8thEds. Thomson

& Thomson. Elsevier 2016; 145-6. www.unife.it › materiale-didattico › t-t_cases_copy www.elsevier.com › books › nussbaum

Hampel H, Frankel WL, Martin E, et al. 2005. Screening for the Lynch syndrome (hereditary nonpolyposis colorectal cancer). *N Engl J Med* 352:1851-60.

Hanahan D, Folkman J. 1996. Patterns and emerging mechanisms of the angiogenic switch during tumorigenesis. *Cell* 86:353–64.

Hanahan D, Weinberg RA. 2000. The Hallmark of Cancer. *Cell* 100: 57-70.

Hatta M, Surachmanto EE, Islam AA, Wahid S. Expression of mRNA IL-17F and sIL-17F in atopic asthma patients. *BMC Res Notes*. 2017;10(1):202. Published 2017 Jun 12. doi:10.1186/s13104-017-2517-93.

He YQ, Sheng JQ, Ling XL et al. 2012. Estradiol regulates miR-135b and MMR expressions via estrogen receptor- β in colorectal cells. *Experimental and Molecular Medicine* 44 (12): 723 – 32.

Heldin CH, Miyazono K, ten Dijke P. 1997. TGF- β signaling from cell membrane to nucleus through SMAD proteins. *Nature* 390:465.

Hemminki A, Markie D, Tomlinson I, et al. 1998. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. *Nature* 391:184.

Hemminki K, Mutanen P. 2001. Genetic epidemiology of multistage carcinogenesis. *Mutat Res* 473:11-21.

Herman JG, Umar A, Polyak K, et al. 1998. Incidence and functional consequences of hMLH1 promoter hypermethylation in colorectal carcinoma. *Proc Natl Acad Sci U S A* 95:6870-5.

Herman JG, Umar A, Polyak K, et al. 1998. Incidence and functional consequences of hMLH1 promoter hypermethylation in colorectal carcinoma. *Proc Natl Acad Sci USA* 95:6870.

Houlston R, Bevan S, Williams A, et al. 1998. Mutations in DPC4 (SMAD4) cause juvenile polyposis syndrome, but only account for a minority of cases. *HumMol Genet* 7:1907.

Houlston R, Crabtree M, Phillips R, Tomlinson I. 2001. Explaining differences in the severity of familial adenomatous polyposis and the search for modifier genes. *Gut* 48:1

Houlston RS, Webb E, Broderick P, et al. 2008. Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer. *Nat Genet* 40:1426-35.

Huang SC, Chen CR, Lavine JE, et al. 2000. Genetic heterogeneity in familial juvenile polyposis. *Cancer Res* 60:6882.

Hudson JD, Shoaibi MA, Maestro R, Carnero A, Hannon GJ, Beach DH. 1999. A proinflammatory cytokine inhibits p53 tumor suppressor activity. *J Exp Med* 190:1375-82.

Hundt S, Haug U, Brenner H. 2009. Comparative evaluation of immunochemical fecal occult blood tests for colorectal adenoma detection. *Ann Intern Med* 150:162-9.

Hunter T. 1997. Oncoprotein networks. *Cell* 88:333-46.

Hurwitz H, Fehrenbacher L, Novotny W, et al. 2004. Bevacizumab plus irinotecan, fluorouracil, and leucovorin for metastatic colorectal cancer. *N Engl J Med* 350:2335-42.

Imperiale TF, Ransohoff DF, Itzkowitz SH, Turnbull BA, Ross ME. 2004. Fecal DNA versus fecal occult blood for colorectal-cancer screening in an average risk population. *N Engl J Med* 351:2704-14.

Ionov Y, Peinado M, Malkhosyan S, Shibata D, Perucho M. 1993. Ubiquitous somatic mutations in simple repeated sequences reveal a new mechanism for colonic carcinogenesis. *Nature* 363:558-61.

Issa JP. 2004. CpG island methylator phenotype in cancer. *Nat Rev Cancer* 4:988-93.

Itzkowitz S, Brand R, Jandorf L, et al. 2008. A simplified, noninvasive stool DNA test for colorectal cancer detection. *Am J Gastroenterol* 103:2862-70.

Itzkowitz SH, Jandorf L, Brand R, et al. 2007. Improved fecal DNA test for colorectal cancer screening. *Clin Gastroenterol Hepatol* 5:111-7.

Jacoby RF, Schlack S, Cole CE, et al. 1997. A juvenile polyposis tumor suppressor locus at 10q22 is deleted from nonepithelial cells in the lamina propria. *Gastroenterology* 112:1398.

Jacoby RF, Schlack S, Sekhon G, Laxova R. 1997. Del(10) (q22.3q24.1) associated with juvenile polyposis. *Am J Med Genet* 70:361.

Jang YS, Steinhagen RM, Heimann TM. 1997. Colorectal cancer in familial adenomatous polyposis. *Dis Colon Rectum* 40:312.

Jarvinen HJ, Aarnio M, Mustonen H, et al. 2000. Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology* 118:829-34.

Jasperson KW, Tuohy TM, Neklason DW, Burt RW. Hereditary and Familial Colon Cancer. *Gastroenterology* 2010; 138(6): 2044-2058. Doi:10.1053/j.gastro.2010.01.054.

Jass JR, Smyrk TC, Stewart SM, et al. 1994. Pathology of hereditary non-polyposis colorectal cancer. *Anticancer Res* 14:1631.

Jass JR, Stewart SM. 1992. Evolution of hereditary non-polyposis colorectal cancer. *Gut* 33:783.

Jass JR, Walsh MD, Barker M, Simms LA, Young J, Leggett BA. 2002. Distinction between familial and sporadic forms of colorectal cancer showing DNA microsatellite instability. *European Journal of Cancer* 39: 858-66.

Jass JR. 2000. Pathology of hereditary nonpolyposis colorectal cancer. *Ann N Y Acad Sci* 910:62.

Jass JR. 2007. Classification of colorectal cancer based on correlation of clinical, morphological and molecular features. *Histopathology* 50:113-30.

Jemal A, Siegel R, Ward E, et al. 2008. Cancer statistics. *CA Cancer J Clin* 58:71-96.

Jen J, Powell SM, Papadopoulos N, et al. 1994. Molecular determinants of dysplasia in colorectal lesions. *Cancer Res* 54:5523.

Jenne DE, Reimann H, Nezu J, et al. 1998. Peutz-Jeghers syndrome is caused by mutations in a novel serine threonine kinase. *Nat Genet* 18:38.

Jhawer M, Goel S, Wilson AJ, et al. 2008. PIK3CA mutation/PTEN expression status predicts response of colon cancer cells to the epidermal growth factor receptor inhibitor cetuximab. *Cancer Res* 68: 1953-61.

Johnson CM, Wei C, Ensor JE, Smolenski DJ, Amos CI, Levin B, Berry DA. Meta-analysis of Colorectal Cancer Risk Factors. *Cancer Causes control. NIH-PA Author Manuscript* 2013; 24(6):1207-1222. Doi:10.1007/s10552-013-0201.5.

Johnson JP. 1991. Cell adhesion molecules of the immunoglobulin supergene family and their role in malignant transformation and progression to metastatic disease. *Cancer Metastasis Rev* 10:11-22.

Jones IT, Jagelman DG, Fazio VW, et al. 1986. Desmoid tumors in familial polyposis coli. *Ann Surg* 204:94.

Jones S, Chen WD, Parmigiani G, et al. 2008. Comparative lesion sequencing provides insights into tumor evolution. *Proc Natl Acad Sci U S A* 105:4283-8.

Jones S, Emmerson P, Maynard J, et al. 2002. Biallelic germline mutations in MYH predispose to multiple colorectal adenoma and somatic G:C→T:A mutations. *Hum Mol Genet* 11:2961-7.

Kamory E, Olasz J, Czuka O. 2008. Somatic APC Inactivation Mechanisms in Sporadic Colorectal Cancer Cases in Hungary. *Pathol.Oncol.Res* 14:51-6.

Kane MF, Loda M, Gaida GM, et al. 1997. Methylation of the hMLH1 promoter correlates with lack of expression of hMLH1 in sporadic colon tumors and mismatch repair-defective human tumor cell lines. *Cancer Res* 57:808-11.

Karapetis CS, Khambata-Ford S, Jonker DJ, et al. 2008. K-ras mutations and benefit from cetuximab in advanced colorectal cancer. *N Engl J Med* 359:1757-65.

Kastrinos F, Syngal S. 2007. Recently identified colon cancer predispositions: MYH and MSH6 mutations. *Semin Oncol* 34:418-24.

Kiel KD, Suit HD. 1984. Radiation therapy in the treatment of aggressive fibromatoses (desmoid tumors). *Cancer* 54:2051

Kilpivaara O, Aaltonen LA. Diagnostic Cancer Genome Sequencing and the Contribution of Germline Variants. *Science*. 2013 Mar 29;339(6127):1559-62. doi: 10.1126/science.1233899.

Kim ER, Kim YH. Clinical Application of Genetics in Management of Colorectal Cancer. *Intestinal research* 2014;12(3): 184-193. Doi: <https://doi.org/10.5217/ir.2014.12.3.184>.)

Kim GP, Colangelo LH, Wieand HS, et al. 2007. Prognostic and predictive roles of high-degree microsatellite instability in colon cancer: a National Cancer Institute-National Surgical Adjuvant Breast and Bowel Project Collaborative Study. *J Clin Oncol* 25:767-72.

Kim IJ, Ku JL, Yoon KA, et al. 2000. Germline mutations of the dpc4 gene in Korean juvenile polyposis patients. *Int J Cancer* 86:529.

Kinzbrunner B, Ritter S, Domingo J, Rosenthal CJ. 1983. Remission of rapidly growing desmoid tumors after tamoxifen therapy. *Cancer* 52:2201.

Kinzler KW, Nilbert MC, Su LK, et al. 1991. Identification of FAP locus genes from chromosome 5q21. *Science* 253:661.

Kinzler KW, Vogelstein B. 1998. Landscaping the cancer terrain. *Science* 280:1036.

Kinzler KW, Vogelstein B. 2002. Colorectal tumors. In: Vogelstein B, Kinzler KW, eds. *The genetic basis of human cancer*. 2nd ed. New York: McGraw-Hill 583-612.

Kinzler KW, Vogelstein B. 1996. Lessons from hereditary colorectal cancer. *Cell* 87: 159–170.

Kinzler KW, Vogelstein B. 1998. Landscaping the cancer terrain. *Science* 280: 1036-7.

Klein WA, Miller HH, Anderson M, DeCosse JJ. 1987. The use of indomethacin, sulindac, and tamoxifen for the treatment of desmoid tumors associated with familial polyposis. *Cancer* 60:2863.

Knudson Jr AG. 1977. Genetics and etiology of human cancer. *Adv Hum Genet* 8:1.

Ko LJ, Prives C. 1996. p53: puzzle and paradigm. *Genes Dev* 10:1054.

Kolodner R. 1996. Biochemistry and genetics of eukaryotic mismatch repair. *Genes Dev* 10:1433.

Kolodner RD, Tytell JD, Schmeits JL, et al. 1999. Germ-line msh6 mutations in colorectal cancer families. *Cancer Res* 59:5068-74.

Kondo Y, Issa JP. 2004. Epigenetic changes in colorectal cancer. *Cancer Metastasis Rev* 23:29-9.

Korinek V, Barker N, Morin PJ, et al. 1997. Constitutive transcriptional activation by a beta-catenin-Tcf complex in APC^{-/-} colon carcinoma. *Science* 275:1784-7.

Kresno SB. 2011. Makna Tes Genetik dalam Upaya Mencegah Kanker. Dalam: Kresna SB. *Ilmu Dasar Onkologi*. Badan Penerbit FKUI 2ed 375-98. A Yu H, Li H, Cui Y et al. The mRNA level of MLH1 in peripheral blood is biomarker for the diagnosis of hereditary nonpolyposis colorectal cancer.

Laken SJ, Petersen GM, Gruber SB, et al. 1997. Familial colorectal cancer in Ashkenazim due to a hypermutable tract in APC. *Nat Genet* 17:79.

Lamlum H, Ilyas M, Rowan A, et al. 1999. The type of somatic mutation at APC in familial adenomatous polyposis is determined by the site of the germline mutation: a new facet to Knudson's "two-hit" hypothesis. *Nat Med* 5:1071.

Lanari A. 1983. Effect of progesterone on desmoid tumors (aggressive fibromatosis). *N Engl J Med* 309:1523.

Leach FS, Nicolaides NC, Papadopoulos N, et al. 1993. Mutations of a mutS homolog in hereditary nonpolyposis colorectal cancer. *Cell* 75:1215-25.

Leary RJ, Lin JC, Cummins J, et al. 2008. Integrated analysis of homozygous deletions, focal amplifications, and sequence alterations in breast and colorectal cancers. *Proc Natl Acad Sci U S A* 105: 16224-9.

Leppert M, Burt R, Hughes JP, et al. 1990. Genetic analysis of an inherited predisposition to colon cancer in a family with a variable number of adenomatous polyps. *N Engl J Med* 322:904.

LeSher AR, Castronuovo Jr JJ, Filippone Jr AL. 1989. Hepatoblastoma in a patient with familial polyposis coli. *Surgery* 105:668.

Levin B, Lieberman DA, McFarland B, et al. 2008. Screening and surveillance for the early detection of colorectal cancer and adenomatous polyps, 2008: a joint guideline from the American Cancer Society, the US Multi-Society Task Force on Colorectal Cancer, and the American College of Radiology. *CA Cancer J Clin* 58: 130-60.

Levy DB, Smith KJ, Beazer-Barclay Y, et al. 1994. Inactivation of both APC alleles in human and mouse tumors. *Cancer Res* 54:5953.

Li M, Chen WD, Papadopoulos N, et al. 2009. Sensitive digital quantification of DNA methylation in clinical samples. *Nat Biotechnol* 27:858-63.

Li M, Wang IX, Li Y, Bruzel A, Richards AL, Toung JM, , Cheung VG. Widespread RNA and DNA Sequence Differences in the Human Transcriptome. *Science* 2011; 333: 53-8. . doi: 10.1126/science.1207018

Liaw D, Marsh DJ, Li J, et al. 1997. Germline mutations of the PTEN gene in Cowden disease, an inherited breast and thyroid cancer syndrome. *Nat Genet* 16:64-7.

Libutti SK, Saltz LB, Tepper JE. 2008. Colon cancer. In: DeVita VT Jr, Lawrence TS, Rosenberg SA, eds. *DeVita, Hellman, and Rosenberg's cancer: principles and practice of oncology*. Vol. 1. Philadelphia: Lippincott Williams & Wilkins 1232-84.

Lichtenstein P, Holm NV, Verkasalo PK, et al. 2000. Environmental and heritable factors in the causation of cancer: analyses of cohorts of twins from Sweden, Denmark, and Finland. *N Engl J Med* 343: 78-85.

Lievre A, Bachet JB, Boige V, et al. 2008. KRAS mutations as an independent prognostic factor in patients with advanced colorectal cancer treated with cetuximab. *J Clin Oncol* 26:374-9.

Lindor NM, Rabe K, Petersen GM, et al. 2005. Lower cancer incidence in Amsterdam-I criteria families without mismatch repair deficiency: familial colorectal cancer type X. *JAMA* 293:1979-85.

Liu B, Parsons R, Papadopoulos N, et al. 1996. Analysis of mismatch repair genes in hereditary non-polyposis colorectal cancer patients. *Nat Med* 2:169.

Liu Q, Tan YQ. Advances in Identification of Susceptibility Gene Defects of Hereditary Colorectal Cancer. *J Cancer* 2019; 10(3):643-653. Doi;10.7150/jca.28542)

Lothe RA, Peltomaki P, Meling GI, et al. 1993. Genomic instability in colorectal cancer: relationship to clinicopathological variables and family history. *Cancer Res* 53:5849.

Lukashev ME, Werb Z. 1998. ECM signaling: orchestrating cell behaviour and misbehaviour. *Trends Cell Biol* 8; 437–441.

Luongo C, Moser AR, Gledhill S, Dove WF. 1994. Loss of *Apc* in intestinal adenomas from Min mice. *Cancer Res* 54:5947.

Lynch HT, Fitzgibbons Jr R, Chong S, et al. 1994. Use of doxorubicin and dacarbazine for the management of unresectable intra-abdominal desmoid tumors in Gardner's syndrome. *Dis Colon Rectum* 37:260.

Lynch HT, Kaul K. 2000. Microsatellite instability, clinical implications, and new methodologies. *J Natl Cancer Inst* 92:511.

Lynch HT, Lynch JF, Lynch PM, Attard T. 2008. Hereditary colorectal cancer syndromes: molecular genetics, genetic counseling, diagnosis and management. *Fam Cancer* 7:27-39.

Lynch HT, Lynch JF. 1994. 25 years of HNPCC. *Anticancer Res* 14:1617.

Lynch HT, Shaw MW, Magnuson CW, et al. 1966. Hereditary factors in cancer: study of two large Midwestern kindreds. *Arch Intern Med* 117:206.

Lynch HT, Smyrk T, Lynch J, et al. 1995. Update on the differential diagnosis, surveillance and management of hereditary non-polyposis colorectal cancer. *Eur J Cancer* 31A:1039.

Lynch HT, Smyrk T, Lynch J. 1997. An update of HNPCC (Lynch syndrome). *Cancer Genet Cytogenet* 93:84.

Lynch HT, Smyrk TC, Lanspa SJ, et al. 1993. Upper gastrointestinal manifestations in families with hereditary flat adenoma syndrome. *Cancer* 71:2709.

Lynch HT, Smyrk TC, Watson P, et al. 1993. Genetics, natural history, tumor spectrum, and pathology of hereditary nonpolyposis colorectal cancer: an updated review. *Gastroenterology* 104:1535.

Lynch HT, Watson P, Krieglner M, et al. 1988. Differential diagnosis of hereditary nonpolyposis colorectal cancer (Lynch syndrome I and Lynch syndrome II). *Dis Colon Rectum* 31:372.

Malkin D, Li FP, Strong LC, et al. 1990. Germ line p53 mutations in a familial syndrome of breast cancer, sarcomas, and other neoplasms. *Science* 250:1233-8. [Erratum, 1993;259:878.]

Mark P. de Caestecker, Piek E, Roberts AB. 2000. Role of Transforming Growth Factor- β Signaling in Cancer. *J Natl Cancer Inst* 92 (17): 1388-1402.

Markie D, Huson S, Maher E, et al. 1996. A pericentric inversion of chromosome six in a patient with Peutz-Jeghers' syndrome and the use of FISH to localise the breakpoints on a genetic map. *Hum Genet* 98:125.

Markowitz S, Wang J, Meyeroff L, Parsons R, Sun L, Lutterbaugh J, Fan R, Zborowska E, Kinzler K, Vogelstein B. 1995. Inactivation of the type II TGF- β receptor in colon cancer-cells with microsatellite instability. *Science* 268:1336-8.

Markowitz SD, Bertagnolli MM. 2009. Molecular Basis of Colorectal Cancer. *N Engl J Med* 361:2449-60

Markowitz SD, Dawson DM, Willis J, Willson JK. 2002. Focus on colon cancer. *Cancer Cell* 1:233-6.

Markowitz SD. 2007. Aspirin and colon cancer— targeting prevention? *N Engl J Med* 356:2195-8.

Marsh DJ, Dahia PL, Zheng Z, et al. 1997. Germline mutations in PTEN are present in Bannayan-Zonana syndrome. *Nat Genet* 16:333-4.

Mecklin JP, Sipponen P, Jarvinen HJ. 1986. Histopathology of colorectal carcinomas and adenomas in cancer family syndrome. *Dis Colon Rectum* 29:849.

Medema RH, and Bos JL. 1993. The role of p21-ras in receptor tyrosine kinase signaling. *Crit. Rev. Oncog* 4:615-661.

Meyerhardt JA, Mayer RJ. 2005. Systemic therapy for colorectal cancer. *N Engl J Med* 352:476-87.

Miyaki M, Konishi M, Kikuchi-Yanoshita R, et al. 1994. Characteristics of somatic mutation of the adenomatous polyposis coli gene in colorectal tumors. *Cancer Res* 54:3011.

Miyaki M, Konishi M, Tanaka K, et al. 1997. Germline mutation of MSH6 as the cause of hereditary nonpolyposis colorectal cancer. *Nat Genet* 17:271-2.

Miyaki M, Lijima T, Kimura J, et al. 1999. Frequent mutation of β -catenin and APC genes in primary colorectal tumors from patient with HNPCC. *Cancer Research* 59: 4506-9.

Miyaki M, Tanaka K, Kikuchi-Yanoshita R, et al. 1995. Familial polyposis: recent advances. *Crit Rev Oncol Hematol* 19:1.

Miyakura Y, Sugano K, Konishi F, et al. 2001. Extensive methylation of hMLH1 promoter region predominates in proximal colon cancer with microsatellite instability. *Gastroenterology* 121:1300.

Miyoshi Y, Nagase H, Ando H, et al. 1992. Somatic mutations of the APC gene in colorectal tumors: mutation cluster region in the APC gene. *Hum Mol Genet* 1:229.

Morin PJ, Sparks AB, Korinek V, et al. 1997. Activation of beta-catenin-Tcf signaling in colon cancer by mutations in beta-catenin or APC. *Science* 275:1787-90.

Munden PM, Sobol WM, Weingeist TA. 1991. Ocular findings in Turcot's syndrome (gliomapolypsis). *Ophthalmology* 98:111.

Myung SJ, Rerko RM, Yan M, et al. 2006. 15-Hydroxyprostaglandin dehydrogenase is an in vivo suppressor of colon tumorigenesis. *Proc Natl Acad Sci U S A* 103:12098-102.

Nagase H, Miyoshi Y, Horii A, et al. 1992. Correlation between the location of germline mutations in the APC gene and the number of colorectal polyps in familial adenomatous polyposis patients. *Cancer Res* 52:4055.

Nakata H, Wang SL, Chung DC, et al. 1998. Oncogenic ras induces gastrin gene expression in colon cancer. *Gastroenterology* 115:1144.

Naylor EW, Lebenthal E. 1980. Gardner's syndrome: recent developments in research and management. *Dig Dis Sci* 25:945.

Neklason DW, Kerber RA, Nilson DB, et al. 2008. Common familial colorectal cancer linked to chromosome 7q31: a genomewide analysis. *Cancer Res* 68:8993-7.

Nishisho I, Nakamura Y, Miyoshi Y, et al. 1991. Mutations of chromosome 5q21 genes in FAP and colorectal cancer patients. *Science* 253:665.

Nosho K, Irahara N, Shima K, et al. 2008. Comprehensive biostatistical analysis of CpG island methylator phenotype in colorectal cancer using a large population based sample. *PLoS One* 3(11):e3698.

Nussbaum RL, McInnes RR, Willard H, Hamosh A. Cancer Genetics and Genomic. In: *Genetics in Medicine*. 8theds. Thomson & Thomson eBook. Philadelphia/Elsevier 2016: 309-32. ISBN 978-1-4377-0696-3 www.academia.edu

Nussbaum RL, McInnes RR, Willard H, Hamosh A. Patterns of Single-Gene Inheritance. In: *Genetics in Medicine*. 8theds. Thomson & Thomson eBook. Philadelphia/Elsevier 2016: 107-32. ISBN 978-1-4377-0696-3 www.academia.edu

Nussbaum RL, McInnes RR, Willard H, Hamosh A. Risk Assessment and Genetic Counseling. In: *Genetics in Medicine*. 8theds. Thomson & Thomson eBook. Philadelphia/Elsevier 2016: 333-48. ISBN 978-1-4377-0696-3 www.academia.edu

NW FACULTY. Developmental BioEngineering (DBE). RESEARCH. PCR PRIMER INFORMATION. <https://www.utwente.nl/en/tnw/dbe/research/pcp-primers/> (2019)

O'Brien CA, Pollett A, Gallinger S, Dick JE. 2007. A human colon cancer cell capable of initiating tumour growth in immunodeficient mice. *Nature* 445:106-10.

O'Brien MJ. 2007. Hyperplastic and serrated polyps of the colorectum. *Gastroenterol Clin North Am* 36:947-68.

Ogino S, Nosho K, Kirkner GJ, et al. 2009. CpG island methylator phenotype, microsatellite instability, BRAF mutation and clinical outcome in colon cancer. *Gut* 58:90-6.

Olumi AF, Grossfeld GD, Hayward SW, Carroll PR, Tlsty TD, and Cunha GR. 1999. Carcinoma-associated fibroblasts direct tumor progression of initiated human prostatic epithelium. *Cancer Res*. 59:5002–11.

Papadopoulos N, Nicolaides NC, Wei YF, et al. 1994. Mutations of a mutL homolog in hereditary colon cancer. *Science* 263: 1625-9.

Papaemmanuil E, Carvajal-Carmona L, Sellick GS, et al. 2008. Deciphering the genetics of hereditary non-syndromic colorectal cancer. *Eur J Hum Genet* 16:1477-86.

Parsons DW, Jones S, Zhang X, et al. 2008. An integrated genomic analysis of human glioblastoma multiforme. *Science* 321:1807-12.

Parsons DW, Wang TL, Samuels Y, et al. 2005. Colorectal cancer: mutations in a signalling pathway. *Nature* 436:792.

Parsons R, Myeroff LL, Liu B, et al. 1995. Microsatellite instability and mutations of the transforming growth factor β type II receptor gene in colorectal cancer. *Cancer Res* 55:5548-50.

Petersen GM, Slack J, Nakamura Y. 1991. Screening guidelines and premorbid diagnosis of familial adenomatous polyposis using linkage. *Gastroenterology* 100:1658.

Plail RO, Bussey HJ, Glazer G, Thomson JP. 1987. Adenomatous polyposis: an association with carcinoma of the thyroid. *Br J Surg* 74:377.

Powell SM, Zilz N, Beazer-Barclay Y, et al. 1992. APC mutations occur early during colorectal tumorigenesis. *Nature* 359:235.

Prives C. 1998. Signaling to p53: breaking the MDM2-p53 circuit. *Cell* 95:5.

Pronk GJ, Bos JL. 1994. The role of p21ras in receptor tyrosine kinase signaling. *Biochim Biophys Acta* 1198:131.

Rajagopalan H, Bardelli A, Lengauer C, Kinzler KW, Vogelstein B, Velculescu VE. 2002. Tumorigenesis: RAF/RAS oncogenes and mismatch-repair status. *Nature* 418: 934.

Rajagopalan H, Jallepalli PV, Rago C, et al. 2004. Inactivation of hCDC4 can cause chromosomal instability. *Nature* 428:77-81.

Ribic CM, Sargent DJ, Moore MJ, et al. 2003. Tumor microsatellite-instability status as a predictor of benefit from fluorouracilbased adjuvant chemotherapy for colon cancer. *N Engl J Med* 349:247-57.

Ricci-Vitiani L, Lombardi DG, Pilozzi E, et al. 2007. Identification and expansion of human colon-cancer-initiating cells. *Nature* 445:111-5.

Riggins GJ, Thiagalingam S, Rozenblum E, et al. 1996. MAD-related genes in the human. *Nat Genet* 13:347-9.

Robbin DH, Itzkowitz SH. 2002. The molecular and genetic basis of colon cancer. *Med Clin N Am* 86:1467-95.

Robson ME, Storm CD, Weitzel J, Wollins DS, Offit K. 2010. Update: Genetic and Genomic Testing for Cancer Susceptibility. American Society of Clinical Oncology Policy Statement. *J Clin Oncol* 28: 5.

Rodriguez-Bigas MA, Boland CR, Hamilton SR, et al. 1997. A National Cancer Institute workshop on hereditary nonpolyposis colorectal cancer syndrome: meeting highlights and Bethesda guidelines. *J Natl Cancer Inst* 89:1758.

Rowan AJ, Lamlum H, Ilyas M, Wheeler J, Straub J, Papadopoulos A. 2000. APC mutations in sporadic colorectal tumors: A mutational "hot spot" and interdependence of the "two hits". *PNAS* 97 : 3352-7.

Rubinfeld B, Souza B, Albert I, et al. 1993. Association of the APC gene product with beta-catenin. *Science* 262:1731.

Sachatello CR, Pickren JW, Grace Jr JT. 1970. Generalized juvenile gastrointestinal polyposis: a hereditary syndrome. *Gastroenterology* 58:699.

Salovaara R, Loukola A, Kristo P, et al. 2000. Population-based molecular detection of hereditary nonpolyposis colorectal cancer. *J Clin Oncol* 18:2193.

Saltz LB, Meropol NJ, Loehrer PJ, Needle MN, Kopit J, Mayer RJ. 2004. Phase II trial of cetuximab in patients with refractory colorectal cancer that expresses the epidermal growth factor receptor. *J Clin Oncol* 22:1201-8.

Samowitz WS, Curtin K, Lin HH, et al. 2001. The colon cancer burden of genetically defined hereditary nonpolyposis colon cancer. *Gastroenterology* 121:830.

Samowitz WS, Curtin K, Ma KN, et al. 2001. Microsatellite instability in sporadic colon cancer is associated with an improved prognosis at the population level. *Cancer Epidemiol Biomarkers Prev* 10:917-23.

Samuels Y, Wang Z, Bardelli A, et al. 2004. High frequency of mutations of the PIK3CA gene in human cancers. *Science* 304:554.

Sandler RS, Halabi S, Baron JA, et al. 2003. A randomized trial of aspirin to prevent colorectal adenomas in patients with previous colorectal cancer. *N Engl J Med* 348:883-90. [Erratum, *N Engl J Med* 2003; 348:1939.]

Schutte M, Hruban R, Hedrick L, Cho K, Nadasdy G, Weinstein C, Bova G, Isaacs W, Cairns P, Nawroz H. 1996. DPC4 gene in various tumor types. *Cancer Res* 56:2527-30.

Shen L, Toyota M, Kondo Y, et al. 2007. Integrated genetic and epigenetic analysis identifies three different subclasses of colon cancer. *Proc Natl Acad Sci U S A* 104:18654-9.

Shibata D, Peinado MA, Ionov Y, et al. 1994. Genomic instability in repeated sequences is an early somatic event in colorectal tumorigenesis that persists after transformation. *Nat Genet* 6:273.

Shibata D, Reale MA, Lavin P, et al. 1996. The DCC protein and prognosis in colorectal cancer. *N Engl J Med* 335:1727.

Shih IM, Yu J, He TC, et al. 2000. The beta-catenin binding domain of adenomatous polyposis coli is sufficient for tumor suppression. *Cancer Res* 60:1671.

Shitoh K, Konishi F, Miyaki M, et al. 2000. Pathogenesis of non-familial colorectal carcinoma with high microsatellite instability. *J Clin Pathol* 53: 841-5.

Siegel RL, Miller KD, Fedewa SA, et al.: Colorectal cancer statistics, 2017. *CA Cancer J Clin* 67(3): 177-193, 2017. (PUBMED Abstract <https://doi.org/10.3322/caac.21395>).

Siena S, Sartore-Bianchi A, Di Nicolantonio F, Balfour J, Bardelli A. 2009. Biomarkers predicting clinical outcome of epidermal growth factor receptor-targeted therapy in metastatic colorectal cancer. *J Natl Cancer Inst* 101:1308-24.

Sirait RH, Hatta M, Ramli M, Islam AA, Arief SK. Systemic lidocaine inhibits high mobility group box 1 messenger ribonucleic acid expression and protein in BALB/c mice after closed fracture musculoskeletal injury. *Saudi J Anaesth.* 2018;12:395-8. DOI: 10.4103/sja.SJA_685_17

Sjoblom T, Jones S, Wood LD, et al. 2006. The consensus coding sequences of human breast and colorectal cancers. *Science* 314:268-74.

Smith KJ, Johnson KA, Bryan TM, et al. 1993. The APC gene product in normal and tumor cells. *Proc Natl Acad Sci USA* 90:2846.

Soussi T, editor. 2002. Institut Curie. Available at: <http://p53.curie.fr>. Accessed October.

Spirio L, Olschwang S, Groden J, et al. 1993. Alleles of the APC gene: an attenuated form of familial polyposis. *Cell* 75:951.

Steinbach G, Lynch PM, Phillips RKS, et al. 2000. The effect of celecoxib, a cyclooxygenase-2 inhibitor, in familial adenomatous polyposis. *N Engl J Med* 342:1946-52.

Stevenson JK, Reid BJ. 1986. Unfamiliar aspects of familial polyposis coli. *Am J Surg* 152:81.

Stigliano V, Sanchez-Mete L, Martayan A, Anti M. Early onset colorectal cancer; A sporadic or inherited disease(?). *World J Gastroenterol.* 2014;20(35):12420-30. Doi 10.3748/wjg.v20.i35.12420

Su LK, Johnson KA, Smith KJ, et al. 1993. Association between wild type and mutant APC gene products. *Cancer Res* 53:2728.

Sudoyo AW. 2005. Kanker Kolorektal Usia Muda etnik Jawa, Sunda, Makasar, dan Minang di Indonesia. *Kajian Klinikopatologi dan Imunohistokimia Instabilitas Mikrosatelit. Disertasi Program Doktor Ilmu Kedokteran FKUI. Jakarta* 118.

Tambaip T, Karo BR, Hatta M, Dwiyanti R, Natzir R, Massi MN, Islam AA, Djawad K, Immunomodulatory effect of orally red fruit (*Pandanus conoideus*) extract on the expression of CC chemokine receptor 5 mRNA in HIV patients with antiretroviral therapy. 2018. *Res. J. Immunol.*, 11: 15-21. DOI: 10.3923/rji.2018.15.21.

Tenesa A, Farrington SM, Prendergast JG, et al. 2008. Genome-wide association scan identifies a colorectal cancer susceptibility locus on 11q23 and replicates risk loci at 8q24 and 18q21. *Nat Genet* 40:631-7.

Thiagalingam S, Lengauer C, Leach FS, et al. 1996. Evaluation of candidate tumour suppressor genes on chromosome 18 in colorectal cancers. *Nat Genet* 13: 343-6.

Thibodeau SN, Bren G, Schaid D. 1993. Microsatellite instability in cancer of the proximal colon. *Science* 260:816-9.

Tie-Jun Liang TJ, Wang HX, Zheng YY, Cao YQ, Wu X et al. APC hypermethylation for early diagnosis of colorectal cancer: a meta-analysis and literature review. *Oncotarget* 2017; 8 :46468-79. <https://doi.org/10.18632/oncotarget.17576>

Tomlinson I, Webb E, Carvajal-Carmona L, et al. 2007. A genome-wide association scan of tag SNPs identifies a susceptibility variant for colorectal cancer at 8q24.21. *Nat Genet* 39:984-8.

Tomlinson IP, Webb E, Carvajal-Carmona L, et al. 2008. A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. *Nat Genet* 40:623-30.

Toyota M, Ahuja N, Ohe-Toyota M, Herman J, Baylin SB, Issa JP. 1999. CpG island methylator phenotype in colorectal cancer. *Proc Natl Acad Sci U S A* 96: 8681-6.

Traboulsi EI, Krush AJ, Gardner EJ, et al. 1987. Prevalence and importance of pigmented ocular fundus lesions in Gardner's syndrome. *N Engl J Med* 316:661.

Tsukada K, Church JM, Jagelman DG, et al. 1991. Systemic cytotoxic chemotherapy and radiation therapy for desmoid in familial adenomatous polyposis. *Dis Colon Rectum* 34:1090.

Turnpenny PD, Ellard S. Patterns of Inheritance. In: Turnpenny PD, Ellard S. *Emery's Elements of Medical Genetics 15thEds.* Elsevier 2017: 66-82 www.elsevier.com › turnpenny › 978-0-7020-6685-6

Turnpenny PD, Ellard S. Risk Calculation. . In: Turnpenny PD, Ellard S. *Emery's Elements of Medical Genetics 15thEds.* Elsevier 2017: 94-101 www.elsevier.com › turnpenny › 978-0-7020-6685-6

Turnpenny PD, Ellard S. The Cellular and Molecular Basis of Inheritance. In: Turnpenny PD, Ellard S. *Emery's Elements of Medical Genetics 15thEds.* Elsevier 2017: 9-23. ISBN: 978-0-7020-6685-6 www.elsevier.com

Turnpenny PD, Ellard S. The Genetics of Cancer and Cancer Genetics. In: Turnpenny PD, Ellard S. *Emery's Elements of Medical Genetics 15thEds.* Elsevier 2017: 177-99 www.elsevier.com › turnpenny › 978-0-7020-6685-6

Utsunomiya J, Nakamura T. 1975. The occult osteomatous changes in the mandible in patients with familial polyposis coli. *Br J Surg* 62:45.

Van Cutsem E, Peeters M, Siena S, et al. 2007. Open-label phase III trial of panitumumab plus best supportive care compared with best supportive care alone in patients with chemotherapy-refractory metastatic colorectal cancer. *J Clin Oncol* 25: 1658-64.

Van de Water NS, Jeevaratnam P, Browett PJ, et al. 1994. Direct mutational analysis in a family with hereditary non-polyposis colorectal cancer. *Aust N Z J Med* 24:682.

Vasen HF, Mecklin JP, Khan PM, Lynch HT. 1991. The International Collaborative Group on Hereditary Non-Polyposis Colorectal Cancer (ICG-HNPCC). *Dis Colon Rectum* 34:424.

Vasen HF, Sanders EA, Taal BG, et al. 1996. The risk of brain tumours in hereditary nonpolyposis colorectal cancer (HNPCC). *Int J Cancer* 65:422.

Vasen HF, Watson P, Mecklin JP, Lynch HT. 1999. New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch syndrome) proposed by the International Collaborative group on HNPCC. *Gastroenterology* 116:1453.

Vasen HF, Wijnen JT, Menko FH, et al. 1996. Cancer risk in families with hereditary nonpolyposis colorectal cancer diagnosed by mutation analysis. *Gastroenterology* 110:1020.

Vazquez A, Bond EE, Levine AJ, Bond GL. 2008. The genetics of the p53 pathway, apoptosis and cancer therapy. *Nat Rev Drug Discov* 7:979-87.

Veigl ML, Kasturi L, Olechnowicz J, et al. 1998. Biallelic inactivation of hMLH1 by epigenetic gene silencing, a novel mechanism causing human MSI cancers. *Proc Natl Acad Sci U S A* 95:8698-702.

Vogelstein B, Fearon ER, Hamilton SR, et al. 1988. Genetic alterations during colorectal-tumor development. *N Engl J Med* 319:525.

Vogelstein B, Papadopoulos N, Velculescu VE, Zhou S, Diaz LA, Kinzler KW. Cancer Genome Landscapes. *Science* 2013; 339: 1546-59. doi: 10.1126/science.1235122

Wallis YL, Macdonald F, Hulten M, et al. 1994. Genotype-phenotype correlation between position of constitutional APC gene mutation and CHRPE expression in familial adenomatous polyposis. *Hum Genet* 94:543.

Walsh N, Qizilbash A, Banerjee R, Waugh GA. 1987. Biliary neoplasia in Gardner's syndrome. *Arch Pathol Lab Med* 111:76.

Warthin AS. 1925. The further study of a cancer family. *J Cancer Res* 9:279.

Watanabe A, Nagashima H, Motoi M, Ogawa K. 1979. Familial juvenile polyposis of the stomach. *Gastroenterology* 77:148.

Watanabe T, Wu T-T, Catalano PJ, et al. 2001. Molecular predictors of survival after adjuvant chemotherapy for colon cancer. *N Engl J Med* 344:1196-206.

Watson P, Lynch HT. 1993. Extracolonic cancer in hereditary nonpolyposis colorectal cancer. *Cancer* 71:677.

Watson P, Vasen HF, Mecklin JP, et al. 1994. The risk of endometrial cancer in hereditary nonpolyposis colorectal cancer. *Am J Med* 96:516.

Weisenberger DJ, Siegmund KD, Campan M, et al. 2006. CpG island methylator phenotype underlies sporadic microsatellite instability and is tightly associated with BRAF mutation in colorectal cancer. *Nat Genet* 38:787-93.

Wheeler JM, Warren BF, Mortensen NJ, et al. 2002. An insight into the genetic pathway of adenocarcinoma of the small intestine. *Gut* 50:218.

Wiesner GL, Daley D, Lewis S, et al. 2003. A subset of familial colorectal neoplasia kindreds linked to chromosome 9q22.2-31.2. *Proc Natl Acad Sci U S A* 100: 12961-5.

Wijnen J, de Leeuw W, Vasen H, et al. 1999. Familial endometrial cancer in female carriers of MSH6 germline mutations. *Nat Genet* 23:142.

Wong R, Cunningham D. 2008. Using predictive biomarkers to select patients with advanced colorectal cancer for treatment with epidermal growth factor receptor antibodies. *J Clin Oncol* 26:5668-70. [Erratum, *J Clin Oncol* 2009;27:3070.]

Wood LD, Parsons DW, Jones S, et al. 2007. The genomic landscapes of human breast and colorectal cancers. *Science* 318: 1108-13.

Wu TT, Rezai B, Rashid A, et al. 1999. Genetic alterations and epithelial dysplasia in juvenile polyposis syndrome and sporadic juvenile polyps.

Wu Y, Berends MJ, Mensink RG, et al. 1999. Association of hereditary nonpolyposis colorectal cancer-related tumors displaying low microsatellite instability with MSH6 germline mutations. *Am J Hum Genet* 65:1291.

Wu Y, Berends MJ, Sijmons RH, et al. 2001. A role for MLH3 in hereditary nonpolyposis colorectal cancer. *Nat Genet* 29:137.

Wu Y, Nystrom-Lahti M, Osinga J, et al. 1997. MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. *Genes Chromosomes Cancer* 18:269.

Yamamoto H, Adachi Y, Taniguchi H, Kunimoto H, Nosho K, Suzuki H, Shinomura Y. 2012. Interrelationship between microsatellite instability and microRNA in gastrointestinal cancer. *World J Gastroenterol* 14 (22): 2745-55.

Yan M, Rerko RM, Platzer P, et al. 2004. 15-Hydroxyprostaglandin dehydrogenase, a COX-2 oncogene antagonist, is a TGF- β -induced suppressor of human gastrointestinal cancers. *Proc Natl Acad Sci USA* 101:17468-73.

Yang Y, Muzny DM, Reid JG, Brainbridge MN, Willis A, Ward PA, et al. Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorder. *N Engl J Med*. 2013 Oct 17;369(16):1502-11. doi: 10.1056/NEJMoa1306555. Epub 2013 Oct 2

Yashiro M, Carethers JM, Laghi L, et al. 2001. Genetic pathways in the evolution of morphologically distinct colorectal neoplasms. *Cancer Res* 61:2676.

Yin Y, Shen WH. 2008. PTEN: a new guardian of the genome. *Oncogene* 27: 5443-53.

Yu H, Li H, Cui Y et al. 2016. The mRNA level of MLH1 in peripheral blood is biomaker for the diagnosis of hereditary nonpolyposis colorectal cancer. *Am J Cancer Res* 6(5): 1135 – 40.

Zanke BW, Greenwood CM, Rangrej J, et al. 2007. Genome-wide association scan identifies a colorectal cancer susceptibility locus on chromosome 8q24. *Nat Genet* 39:989-94.

Zhiheng Zhou, Haibai Liu, Caixia Wang, Qian Lu, Qin Hai Huang, Chanjiao Zheng, Yixiong Lei. referensi Long non-coding RNAs as novel expression signatures modulate DNA damage and repair in cadmium toxicology. *Sci Rep.* 2015; 5: 15293. Published online 2015 Oct 16. doi: 10.1038/srep15293. PMCID: PMC460788.

Zou H, Harrington JJ, Shire AM, et al. 2007. Highly methylated genes in colorectal neoplasia: implications for screening. *Cancer Epidemiol Biomarkers Prev* 16:2686-96.