

SCIENTIFIC PUBLICATIONS (Abstracts and similar not included)

Lauri A. Aaltonen, M.D., PhD

20 SELECTED SCIENTIFIC PUBLICATIONS

18 Original articles (of 262 total)

2. Peltomäki P, Aaltonen LA, Sistonen P, Pylkkänen L, Mecklin J-P, Järvinen H, Green J, Jass J, Weber J, Leach F, Petersen G, Hamilton S, de la Chapelle A & Vogelstein B: Genetic mapping of a locus predisposing to human colorectal cancer. *Science* 260, 810-812, 1993.
3. Aaltonen LA, Peltomäki P, Leach F, Sistonen P, Pylkkänen L, Mecklin J-P, Järvinen H, Powell S, Jen J, Hamilton S, Petersen G, Kinzler K, Vogelstein B & de la Chapelle A: Clues to the pathogenesis of familial colorectal cancer. *Science* 260, 812-816, 1993.
11. Hemminki A, Peltomäki P, Mecklin J-P, Järvinen H, Salovaara R, Nyström-Lahti M, de la Chapelle A & Aaltonen LA. Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. *Nature Genetics* 8, 405-410, 1994.
18. Shibata D, Navidi W, Salovaara R, Li Z-H & Aaltonen LA. Somatic microsatellite mutations as molecular tumor clocks. *Nature Medicine* 2, 676-681, 1996.
22. Hemminki A, Tomlinson I, Markie D, Järvinen H, Sistonen P, Björkqvist A-M, Knuutila S, Salovaara R, Bodmer W, Shibata D, de la Chapelle A & Aaltonen LA. Localization of a susceptibility locus for Peutz-Jeghers syndrome to 19p using comparative genomic hybridization and targeted linkage analysis. *Nature Genetics* 15, 87-90, 1997.
28. Hemminki A, Markie D, Tomlinson I, Avizienyte E, Roth S, Loukola A, Bignell G, Warren W, Aminoff M, Höglund P, Järvinen H, Kristo P, Pelin K, Ridanpää M, Salovaara R, Toro T, Bodmer W, Olschwang S, Olsen AS, Stratton MR, de la Chapelle A & Aaltonen LA. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. *Nature* 391, 184-187, 1998.
30. Aaltonen LA, Salovaara R, Kristo P, Canzian F, Hemminki A, Peltomäki P, Chadwick RB, Percesepe A, Kääriäinen H, Ahtola H, Eskelinen M, Härkönen N, Julkunen R, Kangas E, Ojala S, Tulikoura J, Valkamo E, Järvinen H, Mecklin J-P & de la Chapelle A. Incidence of hereditary nonpolyposis colorectal cancer, and molecular screening for the disease. *New England Journal of Medicine* 338, 1481-1487, 1998.
32. Howe JR, Roth S, Ringold JC, Summers RW, Järvinen HJ, Sistonen P, Tomlinson IPM, Houlston RS, Bevan S, Mitros FA, Stone EM & Aaltonen LA. Mutations in the SMAD4/DPC4 gene in juvenile polyposis. *Science* 280, 1086-1088, 1998.
78. Tomlinson IPM, Alam NA, Rowan AJ, Barclay E, Kelsell D, Leigh I, Gorman P, Lamlum H, Rahman S, Roylance RR & Olpin S - Bevan S, Barker K, Hearle N & Houlston RS – Kiuru M, Lehtonen R, Karhu A, Vilkki S, Laiho P, Eklund C, Vierimaa O, Aittomäki K, Hietala M, Sistonen P, Paetau A, Salovaara R, Herva R, Launonen V & Aaltonen LA. Germline mutations in the fumarate hydratase gene predispose

to dominantly inherited uterine fibroids, skin leiomyomata and renal cell cancer. *Nature Genetics* 30, 406-410, 2002.

135. Vierimaa O, Georgitsi M, Lehtonen R, Vahteristo P, Kokko A, Raitila A, Tuppurainen K, Ebeling TML, Salmela PI, Paschke R, Gündogdu S, de Menis E, Mäkinen M, Launonen V, Karhu A & Aaltonen LA. Pituitary adenoma predisposition caused by germline mutations in the AIP gene. *Science* 312, 1228-1230, 2006.

184. Tuupanen S, Turunen M, Lehtonen R, Hallikas O, Vanharanta S, Kivioja T, Björklund M, Wei G, Yan J, Niittymäki I, Mecklin J-P, Järvinen H, Ristimäki A, Di-Bernardo M, East P, Carvajal-Carmona L, Houlston RS, Tomlinson I, Palin K, Ukkonen E, Karhu A, Taipale J & Aaltonen LA. The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. *Nature Genetics* 41, 885-890, 2009.

212. Mäkinen N, Mehine M, Tolvanen J, Kaasinen E, Li Y, Lehtonen HJ, Gentile M, Yan J, Enge M, Taipale M, Aavikko M, Katainen R, Virolainen E, Böbling T, Koski TA, Launonen V, Sjöberg J, Taipale J, Vahteristo P & Aaltonen LA. MED12, the Mediator Complex Subunit 12 Gene, Is Mutated at High Frequency in Uterine Leiomyomas. *Science* 334, 252-255, 2011.

227. Saarinen S, Pukkala E, Vahteristo P, Mäkinen MJ, Franssila K & Aaltonen LA. High Familial Risk in Nodular Lymphocyte Predominant Hodgkin Lymphoma. *Journal of Clinical Oncology* 31, 938-943, 2013.

228. Sur IK, Hallikas O, Vähärautio A, Yan J, Turunen M, Enge M, Taipale M, Karhu A, Aaltonen LA & Taipale J, Mice Lacking a Myc Enhancer Element that Includes Human SNP rs6983267 Are Resistant to Intestinal Tumors. *Science* 338, 1360-1363, 2012.

236. Mehine M, Kaasinen E, Mäkinen N, Katainen R, Kämpjärvi K, Pitkänen E, Heinonen H-R, Bützow R, Kilpivaara O, Kuosmanen A, Ristolainen H, Gentile M, Sjöberg J, Vahteristo P & Aaltonen LA. Characterization of Uterine Leiomyomas by Whole Genome Sequencing. *New England Journal of Medicine* 369, 453-463, 2013.

237. Gylfe AE, Kondelin J, Turunen M, Ristolainen H, Katainen R, Pitkänen E, Kaasinen E, Rantanen V, Tanskanen T, Varjosalo M, Lehtonen H, Palin K, Taipale M, Taipale J, Renkonen-Sinisalo L, Järvinen H, Böhm J, Mecklin J-P, Ristimäki A, Kilpivaara O, Tuupanen S, Karhu A, Vahteristo P & Aaltonen LA. Identification of candidate oncogenes discovered in human colorectal cancers with microsatellite instability. *Gastroenterology* 145, 540-543, 2013.

256. Katainen R, Dave K, Pitkänen E, Palin K, Kivioja T, Välimäki N, Gylfe A, Ristolainen H, Hänninen UA, Cajuso T, Kondelin J, Tanskanen T, Mecklin J-P, Järvinen H, Renkonen-Sinisalo L, Lepistö A, Kaasinen E, Kilpivaara O, Tuupanen S, Enge M, Taipale J & Aaltonen LA. CTCF/cohesin binding sites are frequently mutated in cancer. *Nature Genetics* 47, 818-821, 2015.

258. Mehine M, Kaasinen E, Heinonen H-R, Mäkinen N, Kämpjärvi K, Sarvilinna N, Aavikko M, Vähärautio A, Pasanen A, Bützow R, Heikinheimo O, Sjöberg J, Pitkänen E, Vahteristo P & Aaltonen LA. Integrated data analysis reveals uterine leiomyoma subtypes with distinct driver pathways and biomarkers. *Proceedings of the National Academy of Sciences* 113, 1315-1320, 2016.

2 Reviews

8. Eng C, Kiuru M, Fernandez MJ & Aaltonen LA. Role for mitochondrial enzymes in inherited neoplasia and beyond. *Nature Reviews Cancer* 3, 193-202, 2003.

13. Kilpivaara O & Aaltonen LA. Diagnostic Cancer Genome Sequencing and the Contribution of Germline Variants. *Science* 339, 1559-1562, 2013.

COMPLETE LIST OF ORIGINAL ARTICLES

1. Peltomäki P, Sistonen P, Mecklin J-P, Pylkkänen L, Aaltonen L, Nordling S, Kere J, Järvinen H, Hamilton S, Petersen G, Kinzler K, Vogelstein B & de la Chapelle A: Evidence that the MCC-APC gene region in 5q21 is not the site for susceptibility to hereditary nonpolyposis colorectal carcinoma. *Cancer Research* 52, 4530-4533, 1992.

2. Peltomäki P, Aaltonen LA, Sistonen P, Pylkkänen L, Mecklin J-P, Järvinen H, Green J, Jass J, Weber J, Leach F, Petersen G, Hamilton S, de la Chapelle A & Vogelstein B: Genetic mapping of a locus predisposing to human colorectal cancer. *Science* 260, 810-812, 1993.

3. Aaltonen LA, Peltomäki P, Leach F, Sistonen P, Pylkkänen L, Mecklin J-P, Järvinen H, Powell S, Jen J, Hamilton S, Petersen G, Kinzler K, Vogelstein B & de la Chapelle A: Clues to the pathogenesis of familial colorectal cancer. *Science* 260, 812-816, 1993.

4. Peltomäki P, Lothe RA, Aaltonen LA, Pylkkänen L, Nyström-Lahti M, Seruca R, David L, Holm R, Ryberg D, Haugen A, Brøgger A, Børresen A-L & de la Chapelle A. Microsatellite instability is associated with tumors that characterize the hereditary nonpolyposis colorectal carcinoma syndrome. *Cancer Research* 53, 5853-5855, 1993.

5. Lothe RA, Peltomäki P, Meling GI, Aaltonen LA, Nyström-Lahti M, Pylkkänen L, Heimdal K, Andersen TI, Møller P, Rognum TO, Fosså SD, Haldorsen T, Langmark F, Brøgger A, de la Chapelle A & Børresen A-L. Genomic instability in colorectal cancer; relationship to clinicopathological variables and family history. *Cancer Research* 53, 5849-5852, 1993.

6. Leach FS, Nicolaidis NC, Papadopoulos N, Liu B, Jen J, Parsons R, Peltomäki P, Sistonen P, Aaltonen LA, Nyström-Lahti M, Guan X-Y, Fournier REK, Todd S, Lewis T, Leach RJ, Naylor SL, Weissenbach J, Mecklin J-P, Järvinen H, Petersen GM, Hamilton SR, Green J, Jass J, Watson P, Lynch HT, Trent JM, de la Chapelle A, Kinzler KW & Vogelstein B. Mutations of a mutS homolog in hereditary nonpolyposis colorectal cancer. *Cell* 75, 1215-1225, 1993.

7. Aaltonen LA, Sankila R, Mecklin J-P, Järvinen H, Pukkala E, Peltomäki P & de la Chapelle A: A novel approach to estimate the proportion of hereditary non-polyposis colorectal cancer of total colorectal cancer burden. *Cancer Detection and Prevention* 18, 57-63, 1994.

8. Aaltonen LA, Peltomäki P, Mecklin J-P, Järvinen H, Jass JR, Green JS, Lynch HT, Watson P, Tallqvist G, Juhola M, Sistonen P, Hamilton SR, Kinzler KW, Vogelstein B & de la Chapelle A. Replication errors in benign and malignant tumors from hereditary nonpolyposis colorectal cancer patients. *Cancer Research* 54, 1645-1648, 1994.
9. Nyström-Lahti M, Sistonen P, Mecklin J-P, Pylkkänen L, Aaltonen LA, Järvinen H, Weissenbach J, de la Chapelle A & Peltomäki P. Close linkage to chromosome 3p and conservation of ancestral founding haplotype in hereditary nonpolyposis colorectal cancer families. *Proceedings of the National Academy of Sciences* 91, 6054-6058, 1994.
10. Nyström-Lahti M, Parsons R, Sistonen P, Pylkkänen L, Aaltonen LA, Leach FS, Hamilton SR, Watson P, Bronson E, Fusaro R, Cavalieri J, Lynch J, Lanspa S, Smyrk T, Lynch P, Drouhard T, Kinzler KW, Vogelstein B, Lynch HT, de la Chapelle A & Peltomäki P. Mismatch repair genes on chromosomes 2p and 3p account for a major share of hereditary nonpolyposis colorectal cancer families evaluable by linkage. *American Journal of Human Genetics* 55, 659-665, 1994.
11. Hemminki A, Peltomäki P, Mecklin J-P, Järvinen H, Salovaara R, Nyström-Lahti M, de la Chapelle A & Aaltonen LA. Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. *Nature Genetics* 8, 405-410, 1994.
12. Nyström-Lahti M, Kristo P, Nicolaidis NC, Chang S-Y, Aaltonen LA, Moisio A-L, Järvinen HJ, Mecklin J-P, Kinzler KW, Vogelstein B, de la Chapelle A & Peltomäki P. Founding mutations and Alu-mediated recombination in hereditary colon cancer. *Nature Medicine* 1, 1203-1206, 1995.
13. Aarnio M, Mecklin J-P, Aaltonen LA, Nyström-Lahti M & Järvinen HJ. Life-time risk of different cancers in hereditary nonpolyposis colorectal cancer (HNPCC) syndrome. *International Journal of Cancer* 64, 430-433, 1995.
14. Sankila R, Aaltonen LA, Järvinen HJ & Mecklin J-P. Better survival rates in *MLH1*-associated hereditary colorectal cancer. *Gastroenterology* 110, 682-687, 1996.
15. Li Z-H, Salovaara R, Aaltonen LA & Shibata D. Telomerase activity is commonly detected in hereditary nonpolyposis colorectal cancers. *American Journal of Pathology* 148, 1075-1079, 1996.
16. Li Z-H, Aaltonen LA, Shu Q, Grizzle W & Shibata D. Effects of mutation and growth rates on patterns of microsatellite instability. *American Journal of Pathology* 148, 1757-1761, 1996.
17. Tarkkanen M, Aaltonen LA, Böbling T, Kivioja A, Karaharju E, Elomaa I, Knuutila S. No evidence of microsatellite instability in bone tumors. *British Journal of Cancer* 74, 453-455, 1996.
18. Shibata D, Navidi W, Salovaara R, Li Z-H & Aaltonen LA. Somatic microsatellite mutations as molecular tumor clocks. *Nature Medicine* 2, 676-681, 1996.
19. Wolf M, Aaltonen LA, Szymanska J, Tarkkanen M, Blomqvist C, Berner J-M, Myklebost O, Knuutila S. Complexity of 12q13-22 amplicon in liposarcoma - Microsatellite repeat analysis. *Genes, Chromosomes & Cancer* 18, 66-70, 1997.

20. Canzian F, Salovaara R, Hemminki A, Kristo P, Chadwick RB, Aaltonen LA & de la Chapelle A. Semiautomated assesment of loss of heterozygosity and replication error in tumors. **Cancer Research** 56, 3331-3337, 1996.
21. Wu Y, Nyström-Lahti M, Osinga J, Looman MWG, Peltomäki P, Aaltonen LA, de la Chapelle A, Hofstra RMW & Buys CHCM. MSH2 and MLH1 mutations in sporadic replication error positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. **Genes, Chromosomes & Cancer** 18, 269-278, 1997.
22. Hemminki A, Tomlinson I, Markie D, Järvinen H, Sistonen P, Björkqvist A-M, Knuutila S, Salovaara R, Bodmer W, Shibata D, de la Chapelle A & Aaltonen LA. Localization of a susceptibility locus for Peutz-Jeghers syndrome to 19p using comparative genomic hybridization and targeted linkage analysis. **Nature Genetics** 15, 87-90, 1997.
23. Aarnio M, Salovaara R, Aaltonen LA, Mecklin J-P & Järvinen HJ. Features of gastric cancer in hereditary nonpolyposis colorectal cancer syndrome. **International Journal of Cancer** 74, 551-555, 1997.
24. Olschwang S, Markie D, Seal S, Neale K, Phillips R, Cottrell S, Ellis I, Hodgson S, Zauber P, Spigelman A, Iwama T, Loff S, McKeown C, Marchese C, Sampson J, Davies S, Talbot I, Wyke J, Thomas G, Bodmer W, Hemminki A, Avizienyte E, de la Chapelle A, Aaltonen LA, Stratton M, Houlston R & Tomlinson I. Peutz-Jeghers disease: Most, but not all, families are compatible with linkage to 19p13.3. **Journal of Medical Genetics** 35, 42-44, 1998.
25. Hemminki A, Höglund P, Pukkala E, Salovaara R, Järvinen H, Norio R & Aaltonen LA. Intestinal cancer in patients with a germline mutation in the down-regulated in adenoma (DRA) gene. **Oncogene** 16, 681-684, 1998.
26. Roth S, Kristo P, Auranen A, Shayeghi M, Seal S, Collins N, Barfoot R, Rahman N, Klemi P-J, Grénman S, Sarantaus L, Nevanlinna H, Butzow R, Ashworth A, Stratton MR & Aaltonen LA. A missense mutation in the BRCA2 gene in three siblings with ovarian cancer. **British Journal of Cancer** 77, 1199-1202, 1998.
27. Marsh DJ, Roth S, Lunetta KL, Hemminki A, Dahia PLM, Sistonen P, Zheng Z, Caron S, van Orsouw NJ, Bodmer WF, Cottrell SE, Dunlop MG, Eccles D, Hodgson SV, Järvinen H, Kellokumpu I, Markie D, Neale K, Phillips R, Rozen P, Syngal S, Vijg J, Tomlinson IPM, Aaltonen LA & Eng C. Exclusion of PTEN and 10q22-24 as the susceptibility locus for juvenile polyposis syndrome (JPS). **Cancer Research** 57, 5017-5021, 1997.
28. Hemminki A, Markie D, Tomlinson I, Avizienyte E, Roth S, Loukola A, Bignell G, Warren W, Aminoff M, Höglund P, Järvinen H, Kristo P, Pelin K, Ridanpää M, Salovaara R, Toro T, Bodmer W, Olschwang S, Olsen AS, Stratton MR, de la Chapelle A & Aaltonen LA. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. **Nature** 391, 184-187, 1998.
29. Percesepe A, Kristo P, Aaltonen LA, Ponz de Leon M, de la Chapelle A & Peltomäki P. Mismatch repair genes and mononucleotide tracts as mutation targets in colorectal tumors with different degrees of microsatellite instability. **Oncogene** 17, 157-163, 1998.

30. Aaltonen LA, Salovaara R, Kristo P, Canzian F, Hemminki A, Peltomäki P, Chadwick RB, Percesepe A, Kääriäinen H, Ahtola H, Eskelinen M, Härkönen N, Julkunen R, Kangas E, Ojala S, Tulikoura J, Valkamo E, Järvinen H, Mecklin J-P & de la Chapelle A. Incidence of hereditary nonpolyposis colorectal cancer, and molecular screening for the disease. *New England Journal of Medicine* 338, 1481-1487, 1998.
31. Avizienyte E, Roth S, Loukola A, Hemminki A, Lothe RA, Stenwig AE, Fosså SD, Salovaara R & Aaltonen LA. Somatic mutations in LKB1 are rare in sporadic colorectal and testicular tumors. *Cancer Research* 58, 2087-2090, 1998.
32. Howe JR, Roth S, Ringold JC, Summers RW, Järvinen HJ, Sistonen P, Tomlinson IPM, Houlston RS, Bevan S, Mitros FA, Stone EM & Aaltonen LA. Mutations in the SMAD4/DPC4 gene in juvenile polyposis. *Science* 280, 1086-1088, 1998.
33. Tsao JL, Zhang J, Salovaara R, Järvinen HJ, Mecklin J-P, Aaltonen LA, Shibata D. Tracing cell fates in human colorectal tumors from somatic microsatellite mutations: evidence of adenomas with stem cell architecture. *American Journal of Pathology* 153, 1189-1200, 1998.
34. Ylikorkala A, Avizienyte E, Tomlinson IPM, Tiainen M, Roth S, Loukola A, Hemminki A, Johansson M, Sistonen P, Markie D, Neale K, Phillips R, Zauber P, Twama T, Sampson J, Järvinen H, Mäkelä TP & Aaltonen LA. Mutations and impaired function of LKB1 in familial and non-familial Peutz-Jeghers syndrome and a sporadic testicular cancer. *Human Molecular Genetics* 8, 45-51, 1999.
35. Prior TW, Chadwick RB, Papp AC, Arcot AN, Isa AM, Pearl DK, Stemmerman G, Percesepe A, Loukola A, Aaltonen LA & de la Chapelle A. The I1307K polymorphism of the APC gene in colorectal cancer. *Gastroenterology* 116, 58-63, 1999.
36. Aarnio M, Sankila R, Pukkala E, Salovaara R, Aaltonen LA, de la Chapelle A, Peltomäki P, Mecklin J-P & Järvinen HJ. Cancer risk in mutation carriers of DNA mismatch repair genes. *International Journal of Cancer* 81, 214-218, 1999.
37. Avizienyte E, Loukola A, Roth S, Hemminki A, Salovaara R, Arola J, Bützow R, Husgafvel-Pursiainen K, Tarkkanen M, Kokkola A, Järvinen H & Aaltonen LA. LKB1 somatic mutations in sporadic tumors. *American Journal of Pathology* 154, 677-681, 1999.
38. Wolf M, Tarkkanen M, Hulsebos T, Larramendy ML, Forus A, Myklebost O, Aaltonen LA, Elomaa I & Knuutila S: Characterization of the 17p amplicon in human sarcomas: microsatellite marker analysis. *International Journal of Cancer* 82, 329-333, 1999.
39. Wang Z-J, Churchman M, Avizienyte E, McKeown C, Davies S, Evans DG, Ferguson A, Ellis I, Xu W-H, Yan Z-Y, Aaltonen LA, & Tomlinson IPM. Germline mutations of the LKB1 (STK11) gene in Peutz-Jeghers patients. *Journal of Medical Genetics* 36, 365-368, 1999.
40. Tsao JL, Tavare S, Salovaara R, Jass JR, Aaltonen LA & Shibata D. Colorectal adenoma and cancer divergence: evidence of multilineage progression. *American Journal of Pathology* 154, 1815-1824, 1999.

41. Roth S, Sistonen P, Hemminki A, Salovaara R, Loukola A, Johansson M, Avizienyte E, Cleary KA, Lynch P, Amos C, Kristo P, Mecklin J-P, Kellokumpu I, Järvinen H & Aaltonen LA. *Smad* genes in juvenile polyposis. ***Genes, Chromosomes and Cancer*** 26, 54-61, 1999.
42. Sarraf P, Mueller E, Smith WM, Wright HM, Kum JB, Aaltonen LA, de la Chapelle A, Spiegelman BM & Eng C. Loss of function mutations in PPAR γ associated with human colon cancer. ***Molecular Cell*** 3, 799-804, 1999.
43. Chen RW, Avizienyte E, Roth S, Leivo I, Mäkitie AA, Aaltonen L-M & Aaltonen LA. *PTEN* and *LKB1* genes in laryngeal tumors. ***Journal of Medical Genetics*** 36, 943-944, 1999.
44. Loukola A, Salovaara R, Kristo P, Moisio A-L, Kääriäinen H, Ahtola H, Eskelinen M, Härkönen N, Julkunen R, Kangas E, Ojala S, Tulikoura J, Valkamo E, Järvinen H, Mecklin J-P, de la Chapelle A & Aaltonen LA. Microsatellite instability in adenomas as a marker for hereditary nonpolyposis colorectal cancer. ***American Journal of Pathology*** 155, 1849-1853, 1999.
45. Loukola A, de la Chapelle A & Aaltonen LA. Strategies for screening for hereditary nonpolyposis colorectal cancer. ***Journal of Medical Genetics*** 36, 819-822, 1999.
46. Riccio A, Aaltonen LA, Godwin AK, Loukola A, Percesepe A, Salovaara R, Masciullo V, Genuardi M, Paravatou-Petsotas M, Bassi DE, Ruggeri BA, Klein-Szanto AJ, Testa JR, Neri G & Bellacosa A. The DNA repair gene *MBD4* (*MEDI*) is mutated in human carcinomas with microsatellite instability. ***Nature Genetics*** 23, 266-268, 1999
47. Esteller M, Avizienyte E, Corn PG, Lothe RA, Baylin SB, Aaltonen LA & Herman JG. Epigenetic inactivation of *LKB1* in primary tumors associated with the Peutz-Jeghers syndrome ***Oncogene*** 19, 164-168, 2000
48. Roth S, Johansson M, Loukola A, Peltomäki P, Järvinen H, Mecklin J-P & Aaltonen LA. Mutation analysis of *SMAD2*, *SMAD3* and *SMAD4* genes in hereditary nonpolyposis colorectal cancer. ***Journal of Medical Genetics*** 37, 298-300, 2000.
49. Kuismanen SA, Holmberg MT, Salovaara R, Schweizer P, Aaltonen LA, de la Chapelle A, Nystrom-Lahti M, Peltomäki P. Epigenetic phenotypes distinguish microsatellite stable and -unstable colorectal cancers. ***Proceedings of the National Academy of Sciences*** 96, 12661-12666, 1999.
50. Launonen V, Avizienyte E, Loukola A, Laiho P, Salovaara R, Järvinen H, Mecklin J-P, Oku A, Shimane M, Kim HC, Kim JC, Nezu J-i, & Aaltonen LA. No Evidence of Peutz-Jeghers Syndrome Gene *LKB1* involvement in Left-Sided Colorectal Carcinomas. ***Cancer Research*** 60, 546-8, 2000.
51. Tsao J-L, Yatabe Y, Salovaara R, Järvinen HJ, Mecklin J-P, Aaltonen LA, Tavare' S & Shibata D. Genetic reconstruction of individual colorectal tumor histories. ***Proceedings of the National Academy of Sciences*** 97, 1236-41, 2000.

52. Järvinen HJ, Aarnio M, Mustonen H, Aktan-Collan K, Aaltonen LA, Peltomäki P, de la Chapelle A & Mecklin J-P. Controlled 15-year trial on screening for colorectal cancer in hereditary nonpolyposis colorectal cancer syndrome. ***Gastroenterology*** 118, 829-834, 2000.
53. Salovaara R, Loukola A, Kristo P, Kääriäinen H, Ahtola H, Eskelinen M, Härkönen N, Julkunen R, Kangas E, Ojala S, Tulikoura J, Valkamo E, Järvinen H, Mecklin J-P, Aaltonen L-A, & de la Chapelle A. Population-based molecular detection of hereditary nonpolyposis colorectal cancer. ***Journal of Clinical Oncology*** 18, 2193-2200, 2000.
54. Wheeler JMD, Loukola A, Aaltonen LA, Mortensen NJ & Bodmer WF. The role of hypermethylation of the *hMLH1* promoter region in HNPCC versus MSI+ sporadic colorectal cancers. ***Journal of Medical Genetics*** 37, 588-592, 2000.
55. Loukola A, Vilkki S, Singh J, Launonen V & Aaltonen LA. Germline and somatic mutation analysis of *MLH3* in MSI-positive colorectal cancer ***American Journal of Pathology*** 157, 347-352, 2000.
56. Avizienyte E, Launonen V, Salovaara R, Kiviluoto T & Aaltonen LA. *E-cadherin* is not frequently mutated in hereditary gastric cancer. ***Journal of Medical Genetics*** 38, 49-52, 2001.
57. Hemminki A, Mecklin J-P, Järvinen HJ, Aaltonen LA & Joensuu H. Microsatellite instability is a favorable prognostic indicator in colorectal cancer patients receiving adjuvant 5-fluorouracil based chemotherapy. ***Gastroenterology*** 119, 921-928, 2000.
58. Roth S, Laiho P, Salovaara R, Launonen V & Aaltonen LA. No *SMAD4* hypermethylation in colorectal cancer. ***British Journal of Cancer*** 83, 1015-1019, 2000.
59. Aaltonen L-M, Chen RW, Roth S, Mäkitie A, Rihkanen H, Vaheri A, & Aaltonen LA. Role of *TP53* polymorphism in human papillomavirus-associated premalignant laryngeal neoplasm. ***Journal of Medical Genetics*** 38, 327, 2001.
60. Zhu Y, Loukola A, Monni O, Kuokkanen K, Franssila K, Elonen E, Vilpo J, Joensuu H, Kere J, Aaltonen LA & Knuutila S. *PPP2R1B* gene in chronic lymphocytic leukemias and mantle cell lymphomas. ***Leukemia Lymphoma*** 41, 177-183, 2001.
61. Dunlop M, Farrington S, Nicholl I, Aaltonen LA, Petersen G, Porteous M & Carothers A. Population carrier frequency of *hMSH2* and *hMLH1* mutations ***British Journal of Cancer*** 83, 1643-1645, 2000.
62. Launonen V, Vierimaa O, Kiuru M, Isola J, Roth S, Pukkala E, Sistonen P, Herva R & Aaltonen LA. Inherited susceptibility to uterine leiomyomas and renal cell cancer. ***Proceedings of the National Academy of Sciences*** 98, 3387-3392, 2001.
63. Lassus H, Laitinen MPE, Anttonen M, Heikinheimo M, Aaltonen LA, Ritvos O & Butzow R. Comparison of serous and mucinous ovarian carcinomas: distinct pattern of allelic loss at distal 8p and expression of transcription factor GATA-4. ***Laboratory Investigation*** 81, 517-526, 2001.

64. Loukola A, Eklin K, Laiho P, Salovaara R, Kristo P, Järvinen H, Mecklin J-P, Launonen V & Aaltonen LA. Microsatellite Marker Analysis in Screening for Hereditary Nonpolyposis Colorectal Cancer (HNPCC). *Cancer Research* 61, 4545-4549, 2001.
65. Woodford-Richens KL, Halford S, Rowan A, Bevan S, Aaltonen LA, Wasan H, Bicknell D, Bodmer WF, Houlston RS & Tomlinson IPM. *CDX2* mutations do not account for Juvenile Polyposis or Peutz-Jeghers syndrome and occur infrequently in sporadic colorectal cancers. *British Journal of Cancer* 84, 1314-1316, 2001.
66. Lassus H, Salovaara R, Aaltonen LA & Butzow R. Allelic analysis of serous ovarian carcinoma reveals two putative tumor suppressor loci at 18q22-q23 distal to *SMAD4*, *SMAD2* and *DCC*. *American Journal of Pathology* 159, 35-42, 2001.
67. Vilkki S, Tsao J-L, Loukola A, Pöyhönen M, Vierimaa O, Herva R, Aaltonen LA & Shibata D. Extensive somatic microsatellite mutations in normal human tissue. *Cancer Research* 61, 4541-4544, 2001.
68. Kiuru M, Launonen V, Hietala M, Aittomäki K, Vierimaa O, Salovaara R, Arola J, Pukkala E, Sistonen P, Herva R, & Aaltonen LA. Familial cutaneous leiomyomatosis is a two-hit condition associated with renal cell cancer of characteristic histopathology. *American Journal of Pathology* 159, 825-829, 2001.
69. Woodford-Richens KL, Rowan AJ, Poulson R, Bevan S, Salovaara R, Aaltonen LA, Houlston RS, Wright NA & Tomlinson IPM. Comprehensive analysis of *SMAD4* mutations and protein expression in juvenile polyposis: evidence for a distinct genetic pathway and polyp morphology in *SMAD4* mutation carriers. *American Journal of Pathology* 159, 1293-300, 2001.
70. Vahteristo P, Tamminen A, Karvinen P, Eerola H, Eklund C, Aaltonen LA, Blomqvist C, Aittomäki K & Nevanlinna H. *p53*, *CHK2*, and *CHK1* genes in Finnish families with Li-Fraumeni syndrome: further evidence of *CHK2* in inherited cancer predisposition. *Cancer Research* 61, 5718-5722, 2001.
71. Salashsor S, Huo H, Diep CB, Loukola A, Zhang H, Liu T, Chen J, Iselius L, Rubio C, Lothe RA, Aaltonen LA, Sun X-F, Lindmark G & Lindblom A. A germline E-cadherin mutation in a family with gastric and colon cancer. *International Journal of Molecular Medicine* 8, 439-443, 2001.
72. Zhou X-P, Woodford-Richens K, Lehtonen R, Kurose K, Aldred M, Hampel H, Launonen V, Virta S, Pilarski R, Salovaara R, Bodmer WF, Conrad BA, Dunlop M, Hodgson SV, Iwama T, Järvinen H, Kellokumpu I, Kim JC, Leggett B, Markie D, Mecklin J-P, Neale K, Phillips R, Piris J, Rozen P, Houlston RS, Aaltonen LA, Tomlinson IPM & Eng C. Germline mutations in *BMPRIA/ALK3*, encoding a member of the TGF β receptor-SMAD superfamily, cause a subset of cases with juvenile polyposis syndrome and Cowden/Bannayan-Riley-Ruvalcaba syndrome. *American Journal of Human Genetics* 69, 704-711, 2001.
73. Nakagawa H, Nuovo GJ, Zervos EE, Martin EW Jr, Salovaara R, Aaltonen LA & de la Chapelle A. Age-related hypermethylation of *MLH1* 5' region in normal colonic mucosa is associated with microsatellite-unstable colorectal cancer development. *Cancer Research* 61, 6991-6995, 2001.

74. Esteller M, Fraga MF, Guo M, Garcia-Foncillas J, Hedenfalk I, Godwin AK, Trojan J, Vaurs-Barrière C, Bignon Y-J, Ramus S, Benitez J, Caldes T, Akiyama Y, Yuasa Y, Launonen V, Canal MJ, Rodriguez R, Capella G, Peinado MA, Borg A, Aaltonen LA, Ponder BA, Baylin SB & Herman JG. DNA methylation patterns in hereditary human cancers mimic sporadic tumorigenesis. *Human Molecular Genetics* 10, 3001-3007, 2001.
75. Salovaara R, Roth S, Loukola A, Launonen V, Sistonen P, Avizienyte E, Kristo P, Järvinen H, Souchelnytskyi S, Sarlomo-Rikala M & Aaltonen LA. Frequent loss of SMAD4/DPC4 protein in colorectal cancers. *Gut* 51, 56-59, 2002.
76. Laiho P, Launonen V, Lahermo P, Esteller M, Guo M, Herman JG, Mecklin J-P, Järvinen H, Sistonen P, Kim K-M, Shibata D, Houlston RS & Aaltonen LA. Low-level microsatellite instability in most colorectal carcinomas. *Cancer Research* 62, 1166-1170, 2002.
77. Kim K-M, Salovaara R, Mecklin J-P, Järvinen H-J, Aaltonen LA & Shibata D. Poly-A deletions in hereditary nonpolyposis colorectal cancer: Mutations before a gatekeeper. *American Journal of Pathology* 160, 1503-1506, 2002.
78. Tomlinson IPM, Alam NA, Rowan AJ, Barclay E, Kelsell D, Leigh I, Gorman P, Lamlum H, Rahman S, Roylance RR & Olpin S - Bevan S, Barker K, Hearle N & Houlston RS – Kiuru M, Lehtonen R, Karhu A, Vilkki S, Laiho P, Eklund C, Vierimaa O, Aittomäki K, Hietala M, Sistonen P, Paetau A, Salovaara R, Herva R, Launonen V & Aaltonen LA. Germline mutations in the fumarate hydratase gene predispose to dominantly inherited uterine fibroids, skin leiomyomata and renal cell cancer. *Nature Genetics* 30, 406-410, 2002.
79. Birkenkamp-Demtroder K, Christensen LL, Olesen SH, Frederiksen CM, Laiho P, Aaltonen LA, Laurberg S, Sorensen FB, Hagemann R & Orntoft T. Gene expression in colorectal cancer. *Cancer Research* 62, 4352-4363, 2002.
80. Howe JR, Shellnut J, Wagner B, Ringold JC, Sayed MG, Ahmed AF, Lynch PM, Amos CI, Sistonen P & Aaltonen LA. Common deletion of *SMAD4* in juvenile polyposis is a mutational hotspot. *American Journal of Human Genetics* 70, 1357-1362, 2002.
81. Oliveira C, Bordin MC, Grehan N, Huntsman D, Suriano G, Machado JC, Kiviluoto T, Aaltonen L, Jackson CE, Seruca R, Caldas C. Screening E-cadherin in gastric cancer families reveals germline mutations only in hereditary diffuse gastric cancer kindred. *Human Mutation* 19, 510-517, 2002.
82. Kiuru M, Lehtonen R, Arola J, Salovaara R, Järvinen H, Aittomäki K, Sjöberg J, Visakorpi T, Knuutila S, Isola J, Delahunt B, Herva R, Launonen V, Karhu A & Aaltonen LA. Few *FH* mutations in sporadic counterparts of tumor types observed in hereditary leiomyomatosis and renal cell cancer (HLRCC) families. *Cancer Research* 62, 4554-4557, 2002.
83. Renkonen-Sinisalo L, Sipponen P, Aarnio M, Julkunen R, Aaltonen LA, Järvinen HJ & Mecklin J-P. No support for endoscopic surveillance for gastric cancer in hereditary non-polyposis colorectal cancer. *Scandinavian Journal of Gastroenterology* 37, 574-577, 2002.

84. Rossi DJ, Ylikorkala A, Korsisaari N, Salovaara R, Luukko K, Launonen V, Henkemeyer M, Ristimäki A, Aaltonen LA & Mäkelä TP. A mouse model for Peutz-Jeghers syndrome reveals induction of Cyclooxygenase-2 in Peutz-Jeghers polyposis. *Proceedings of the National Academy of Sciences* 99, 2327-12332, 2002.
85. Vilkki S, Launonen V, Karhu A, Sistonen P, Västriik I & Aaltonen LA. Screening for microsatellite instability target genes in colorectal cancers. *Journal of Medical Genetics* 39, 785-789 2002.
86. Zhou X-P, Loukola A, Salovaara R, Nystrom-Lahti M, Peltomaki P, de la Chapelle A, Aaltonen LA & Eng C. PTEN Mutational Spectra, Expression Levels, and Subcellular Localization in Microsatellite Stable and Unstable Colorectal Cancers. *American Journal of Pathology* 161, 439-447, 2002.
87. Lehtonen R, Kiuru M, Rökman A, Ikonen T, Cunningham JM, Schaid DJ, Matikainen M, Nupponen NN, Karhu A, Kallioniemi O-P, Thibodeau SN, Schleutker J & Aaltonen LA. No fumarate hydratase (FH) mutations in hereditary prostate cancer. *Journal of Medical Genetics* 40, e19, 2003.
88. Jagmohan-Changur S, Poikonen T, Vilkki S, Launonen V, Wikman F, Orntoft TF, Møller P, Vasen H, Tops C, Kolodner RD, Mecklin J-P, Järvinen H, Bevan S, Houlston RS, Aaltonen LA, Fodde R, Wijnen J & Karhu A. EXO1 Variants Occur Commonly in Normal Population: Evidence Against a Role in Hereditary Nonpolyposis Colorectal Cancer. *Cancer Research* 63, 154-158, 2003.
89. Laiho P, Hienonen T, Karhu A, Lipton L, Aalto Y, Thomas H, Birkenkamp-Demtroder K, Hodgson S, Salovaara R, Mecklin J-P, Järvinen H, Knuutila S, Halford S, Orntoft TF, Tomlinson I, Launonen V, Houlston R & Aaltonen LA. Genome-wide allelotyping of 104 Finnish colorectal cancers reveals an excess of allelic imbalance in chromosome 20q in familial cases. *Oncogene* 22, 2206-2214, 2003.
90. Sieber OM, Lipton L, Crabtree M, Heinimann K, Fidalgo P, Phillips RKS, Bisgaard M-L, Orntoft TF, Aaltonen LA, Hodgson SV, Thomas JW & Tomlinson IPM. The multiple colorectal adenoma phenotype, familial adenomatous polyposis and germline mutations in MYH. *New England Journal of Medicine* 348, 791-799, 2003.
91. Laiho P, Hienonen T, Mecklin J-P, Järvinen H, Karhu A, Launonen V & Aaltonen LA. Mutation and LOH analysis of ACO2 in Colorectal Cancer; no evidence of biallelic genetic inactivation. *Journal of Medical Genetics* 40, e73, 2003.
92. Hienonen T, Laiho P, Salovaara R, Mecklin J-P, Järvinen H, Sistonen P, Peltomäki P, Lehtonen R, Nupponen NN, Launonen V, Karhu A & Aaltonen LA. Little evidence for involvement of MLH3 in colorectal cancer predisposition. *International Journal of Cancer* in 106, 292-296, 2003.
93. Kilpivaara O, Laiho P, Aaltonen LA & Nevanlinna H. CHEK2 1100delC and Colorectal Cancer. *Journal of Medical Genetics* 40, e110, 2003.
94. Enholm S, Hienonen T, Suomalainen A, Tomlinson I, Lipton L, Kärjä V, Eskelinen M, Mecklin J-P, Karhu A, Järvinen HJ & Aaltonen LA. Proportion and phenotype of MYH associated colorectal neoplasia in a population based series of Finnish colorectal cancer patients. *American Journal of Pathology* 163, 827-832, 2003.

95. Hemminki K, Aaltonen L, Li X. Subsequent primary malignancies after endometrial carcinoma and ovarian carcinoma. *Cancer* 97, 2432-2439, 2003.
96. Alam NA, Gorman P, Jaeger EEM, Kelsell D, Leigh IM, Ratnavel R, Murdoch ME, Houlston RS, Aaltonen LA, Roylance RR & Tomlinson IPM. Germline deletions of EXO1 do not cause colorectal tumors and lesions that are null for EXO1 do not have microsatellite instability. *Cancer Genetics and Cytogenetics* 147, 121-127, 2003.
97. Lehtonen R, Kiuru M, Vanharanta S, Sjöberg J, Aaltonen L-M, Aittomäki K, Arola J, Butzow R, Eng C, Husgafvel-Pursiainen K, Isola J, Järvinen H, Koivisto P, Mecklin J-P, Peltomäki P, Salovaara R, Wasenius V-M, Karhu A, Launonen V, Nupponen NN & Aaltonen LA. Biallelic inactivation of fumarate hydratase (FH) occurs in non-syndromic uterine leiomyomas but is rare in other tumors. *American Journal of Pathology* 164, 17-22, 2004.
98. Vanharanta S, Buchta M, McWhinney SR, Virta S, Peszkowska M, Morrison CD, Lehtonen R, Januszewicz A, Järvinen H, Juhola M, Mecklin J-P, Pukkala E, Herva R, Kiuru M, Nupponen NN, Aaltonen LA, Neumann HPH & Eng C. Early onset renal cell carcinoma as novel extra-paraganglial component of *SDHB*-associated heritable paraganglioma. *American Journal of Human Genetics* 74, 153-159, 2004.
99. Lipton L, Halford SE, Johnson V, Novelli MR, Jones A, Cummings C, Sieber O, Bisgaard ML, Hodgson SV, Aaltonen LA, Thomas HJV & Tomlinson IPM. Carcinogenesis in *MYH*-associated polyposis follows a distinct genetic pathway. *Cancer Research* 63, 7595-7599, 2003.
100. Calabrese P, Tsao J-L, Yatabe Y, Salovaara R, Mecklin J-P, Järvinen H, Aaltonen LA, Tavaré S & Shibata D. Colorectal pre-tumor progression before and after loss of DNA mismatch repair *American Journal of Pathology* 164, 1447-1453, 2004.
101. Howe JR, Sayed MG, Ahmed F, Ringold J, Larsen-Haidle J, Merg A, Mitros F, Vocarro C, Petersen G, Giardiello F, Tinley S, Aaltonen LA & Lynch H. The prevalence of *MADH4* and *BMPRI1A* mutations in juvenile polyposis, and absence of *BMPRI2*, *BMPRI1B*, and *ACVRI* mutations *Journal of Medical Genetics* 41, 484-491, 2004.
102. Domingo E, Laiho P, Ollikainen M, Pinto M, Wang L, French AJ, Westra J, Frebourg T, Espín E, Armengol M, Hamelin R, Yamamoto H, Hofstra RM, Seruca R, Lindblom A, Peltomäki PT, Thibodeau SN, Aaltonen LA & Schwartz S Jr. *BRAF* screening as a low-cost effective strategy for simplifying HNPCC genetic testing. *Journal of Medical Genetics* 41, 664-668, 2004.
103. Oliveira C, Westra JL, Arango D, Ollikainen M, Domingo E, Ferreira A, Velho S, Niessen R, Lagerstedt K, Alhopuro P, Laiho P, Veiga I, Teixeira M, Ligtenberg M, Kleibeuker JH, Sijmons RH, Plukker JT, Imai K, Lage P, Hamelin R, Albuquerque C, Schwartz S, Lindblom A, Peltomäki P, Yamamoto H, Aaltonen LA, Seruca R & Hofstra R. Distinct patterns of *KRAS* mutations in colorectal carcinomas according to germline mismatch repair defects and hMLH1 methylation status. *Human Molecular Genetics* 13, 2303-2311, 2004.

104. Alhopuro P, Ahvenainen T, Mecklin J-P, Juhola M, Järvinen HJ, Karhu A, & Aaltonen LA. NOD2 3020insC alone is not sufficient for colorectal cancer predisposition. *Cancer Research* 64, 7245-7247, 2004.
105. Birkenkamp-Demtröder K, Olesen SH, Sørensen FB, Laurberg S, Laiho P, Aaltonen LA & Ørntoft TF. Differential gene expression in colon cancer of the caecum versus the sigmoid and rectosigmoid. *Gut* 54, 374-84, 2005.
106. Kiuru M, Lehtonen R, Eerola H, Aittomäki K, Blomqvist C, Nevanlinna H, Aaltonen LA & Launonen V. No germline *FH* mutations in familial breast cancer patients. *European Journal of Human Genetics* 13, 506-509, 2005.
107. Hienonen T, Sammalkorpi H, Isohanni P, Versteeg R, Karikoski R & Aaltonen LA. A 17p11.2 germline deletion in a patient with Smith-Magenis syndrome and neuroblastoma. *Journal of Medical Genetics* 42, Online e3, 2005.
108. Alazzouzi H, Alhopuro P, Salovaara R, Sammalkorpi H, Järvinen H, Mecklin J-P, Schwartz S Jr, Aaltonen LA & Arango D. SMAD4 as a prognostic marker in colorectal cancer. *Clinical Cancer Research* 11, 2606-2611, 2005.
109. Rasinperä H, Forsblom C, Enattah N, Halonen P, Salo K, Victorzon M, Mecklin J-P, Järvinen H, Enholm S, Sellick G, Alazzouzi H, Houlston R, Robinson J, Groop P-H, Tomlinson I, Schwartz Jr S, Aaltonen LA & Järvelä I, and The FinnDiane Study Group. The C/C-13910 genotype of adult-type hypolactasia is associated with an increased risk of colorectal cancer in the Finnish population. *Gut* 54, 643-647, 2005.
110. Vahteristo P, Ojala S, Tamminen A, Tommiska J, Sammalkorpi H, Kiuru-Kuhlefelt S, Eerola H, Aaltonen LA, Aittomäki K & Nevanlinna H. No MSH6 germline mutations in breast cancer families with colorectal and/or endometrial cancer. *Journal of Medical Genetics* 42, Online e22, 2005.
111. Alhopuro P, Katajisto P, Lehtonen R, Ylisaukko-oja SK, Näätsaari L, Karhu A, Westerman AM, Wilson JHP, de Rooij FWM, Vogel T, Moeslein G, Tomlinson IP, Aaltonen LA, Mäkelä TP & Launonen V. Mutation analysis of three genes encoding novel LKB1 interacting proteins, BRG1, STRADalpha, and MO25alpha, in Peutz-Jeghers Syndrome. *British Journal of Cancer* 92, 1126-1129, 2005.
112. Eschrich S, Yang I, Bloom G, Kwong KY, Boulware D, Cantor A, Coppola D, Kruhoffer M, Aaltonen L, Orntoft TF, Quackenbush J & Yeatman TJ. Molecular staging for survival prediction of colorectal cancer patients. *Journal of Clinical Oncology* 15, 3526-3535, 2005
113. Domingo E, Niessen RC, Oliveira C, Alhopuro P, Moutinho C, Espín E, Armengol M, Sijmons RH, Kleibeuker JH, Seruca R, Aaltonen LA, Imai K, Yamamoto H, Schwartz S Jr, Hofstra RMW. BRAF-V600E is not involved in the colorectal tumorigenesis of HNPCC in patients with functional MLH1 and MSH2 genes. *Oncogene* 24, 3995-3998, 2005.
114. Alhopuro P, Ylisaukko-oja SK, Koskinen WJ, Bono P, Arola J, Järvinen HJ, Mecklin J-P, Atula T, Kontio R, Mäkitie AA, Suominen S, Leivo I, Vahteristo P, Aaltonen L-M & Aaltonen LA. The *MDM2*

promoter polymorphism SNP309T>G and the risk of uterine leiomyosarcoma, colorectal cancer, and squamous cell carcinoma of the head and neck. *Journal of Medical Genetics* 42, 694-698, 2005.

115. Hienonen T, Sammalkorpi H, Enholm S, Alhopuro P, Barber TD, Lehtonen R, Nupponen NN, Lehtonen H, Salovaara R, Mecklin J-P, Järvinen H, Koistinen R, Arango D, Launonen V, Vogelstein B, Karhu A & Aaltonen LA. Mutations in two short non-coding mononucleotide repeats in most microsatellite unstable colorectal cancers. *Cancer Research* 65, 4607-4613, 2005.

116. Vanharanta S, Wortham NC, Laiho P, Sjöberg J, Aittomäki K, Arola J, Tomlinson IP, Karhu A, Arango D & Aaltonen LA. 7q deletion mapping and expression profiling in uterine fibroids. *Oncogene* 24, 6545-6554, 2005.

117. Hampel H, Stephens JA, Pukkala E, Sankila R, Aaltonen LA, Mecklin J-P & de la Chapelle A. Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer syndrome: Later age of onset. *Gastroenterology* 129, 415-421, 2005

118. Hienonen T, Salovaara R, Mecklin J-P, Järvinen H, Karhu A & Aaltonen LA. Preferential amplification of *AURKA* 91A (Ile31) in familial colorectal cancers. *International Journal of Cancer* 118, 505-508, 2006.

119. Alhopuro P, Parker AR, Lehtonen R, Enholm S, Järvinen H, Mecklin J-P, Karhu A, Eshleman JR & Aaltonen LA. A novel functionally deficient *MYH* variant in individuals with colorectal adenomatous polyposis. *Human Mutation* online report, 26, 393, 2005.

120. Alhopuro P, Alazzouzi H, Sammalkorpi H, Davalos V, Salovaara R, Hemminki A, Järvinen H, Mecklin J-P, Schwartz S Jr, Aaltonen LA & Arango D. SMAD4 levels and response to 5-fluorouracil in colorectal cancer. *Clinical Cancer Research* 11, 6311-6316, 2005.

121. Arango D, Laiho P, Kokko A, Alhopuro P, Sammalkorpi H, Salovaara R, Nicorici D, Hautaniemi S, Alazzouzi H, Mecklin J-P, Järvinen H, Hemminki A, Astola J, Schwartz S Jr & Aaltonen LA. Gene expression profiling predicts recurrence in Dukes C colorectal cancer. *Gastroenterology* 129, 874-884, 2005.

122. Kokko A, Ylisaukko-oja SSK, Kiuru M, Takatalo MS, Salmikangas P, Tuimala J, Arango D, Karhu A, Aaltonen LA & Jäntti J. Modeling tumor predisposing *FH* mutations in yeast; effects on fumarase activity, growth phenotype and gene expression profile. *International Journal of Cancer* 118, 1340-1345, 2006.

123. Sotamaa K, Liyanarachchi S, Mecklin J-P, Järvinen H, Aaltonen LA, Peltomäki P & de la Chapelle A. *p53* codon 72 and *MDM2* SNP 309 Polymorphisms and Age of Colorectal Cancer Onset in Lynch Syndrome. *Clinical Cancer Research* 11, 6840-6844, 2005

124. Kruhoffer M, Jensen JL, Laiho P, Dyrskjot L, Salovaara R, Arango D, Birkenkamp-Demtroder K, Sorensen FB, Christensen LL, Buhl L, Mecklin JP, Jarvinen H, Thykjaer T, Wikman FP, Bech-Knudsen F, Juhola M, Nupponen NN, Laurberg S, Andersen CL, Aaltonen LA & Orntoft TF. Gene expression signatures for colorectal cancer microsatellite status and HNPCC. *British Journal of Cancer* 92, 2240-2248, 2005.

125. Lehtonen HJ, Kiuru M, Ylisaukko-oja SK, Salovaara R, Herva R, Koivisto PA, Vierimaa O, Aittomäki K, Pukkala E, Launonen V & Aaltonen LA. Increased risk of cancer in patients with fumarate hydratase germline mutation. *Journal of Medical Genetics* 43, 523-526, 2006.
126. Alazzouzi H, Davalos V, Kokko A, Domingo E, Woerner SM, Wilson AJ, Konrad L, Espín E, Armengol M, Imai K, Yamamoto H, Mariadason JM, Gebert JF, Aaltonen LA, Schwartz Jr. S & Arango D. Mechanisms of inactivation of the receptor tyrosine kinase EPHB2 in colorectal tumors. *Cancer Research* 65, 10170-3, 2005.
127. Calabrese P, Mecklin J-P, Järvinen HJ, Aaltonen LA, Travare S & Shibata D. Number of mutations to different types of colorectal cancer. *BMC Cancer* 5, 126, 2005.
128. Sweet K, Willis J, Zhou X-P, Gallione C, Sawada T, Alhopuro P, Khoo SK, Patocs A, Martin C, Bridgeman S, Heinz J, Pilarski R, Lehtonen R, Prior T, Frebourg T, Teh BT, Marchuk DA, Aaltonen LA & Eng C. Molecular classification of patients with unexplained hamartomatous and hyperplastic polyposis. *Journal of the American Medical Association* 294, 2465-2473, 2005.
129. Kilpivaara O, Alhopuro P, Vahteristo P, Aaltonen LA & Nevanlinna H. *CHEK2* I157T associates with familial and sporadic colorectal cancer. *Journal of Medical Genetics* 43, e34, 2006.
130. Vanharanta S, Pollard PJ, Lehtonen HJ, Laiho P, Sjöberg J, Leminen A, Aittomäki K, Arola J, Kruhoffer M, Orntoft TF, Tomlinson IP, Kiuru M, Arango D & Aaltonen LA. Distinct expression profile in fumarate hydratase-deficient uterine fibroids. *Human Molecular Genetics* 15, 97-103, 2006
131. Ylisaukko-oja S, Kiuru M, Lehtonen HJ, Lehtonen R, Pukkala E, Arola J, Launonen V & Aaltonen LA. Analysis of *Fumarate Hydratase* Mutations in a Population Based Series of Early Onset Uterine Leiomyosarcoma Patients. *International Journal of Cancer* 119, 283-287, 2006.
132. Ropero S, Fraga MF, Ballestar E, Hamelin R, Yamamoto H, Boix-Chornet M, Caballero R, Alaminos M, Setien F, Paz MF, Herranz M, Palacios J, Arango D, Orntoft T, Aaltonen LA, Schwartz S Jr & Esteller M. A truncating mutation of *HDAC2* in human cancers confers resistance to histone deacetylase inhibition. *Nature Genetics* 38, 566-569, 2006.
133. Ylisaukko-oja SK, Cybulski C, Lehtonen R, Kiuru M, Matyjasik J, Szymańska A, Szymańska-Pasternak J, Dyrskjot L, Butzow R, Orntoft TF, Launonen V, Lubiński J & Aaltonen LA. Germline *fumarate hydratase* mutations in patients with ovarian mucinous cystadenoma. *European Journal of Human Genetics* 14, 880-883, 2006.
134. Volikos E, Robinson J, Aittomäki K, Mecklin JP, Jarvinen H, Westerman AM, de Rooji FW, Vogel T, Moeslein G, Launonen V, Tomlinson IP, Silver AR & Aaltonen LA. *LKB1* exonic and whole gene deletions are a common cause of Peutz-Jeghers syndrome. *Journal of Medical Genetics* 43, e18, 2006.
135. Vierimaa O, Georgitsi M, Lehtonen R, Vahteristo P, Kokko A, Raitila A, Tuppurainen K, Ebeling TML, Salmela PI, Paschke R, Gündogdu S, de Menis E, Mäkinen M, Launonen V, Karhu A & Aaltonen LA. Pituitary adenoma predisposition caused by germline mutations in the *AIP* gene. *Science* 312, 1228-1230, 2006.

136. Laiho P, Kokko A, Vanharanta S, Salovaara R, Sammalkorpi H, Järvinen H, Mecklin J-P, Karttunen TJ, Tuppurainen K, Davalos V, Schwartz Jr. S, Arango D, Mäkinen MJ & Aaltonen LA. Serrated Carcinomas Form a Subclass of Colorectal Cancer with Distinct Molecular Basis. *Oncogene* 26, 312–320, 2006.
137. Kokko A, Laiho P, Lehtonen R, Korja S, Carvajal-Carmona LG, Järvinen H, Mecklin J-P, Eng C, Schleutker J, Tomlinson IPM, Vahteristo P & Aaltonen LA. *EPHB2* germline variants in patients with colorectal cancer or hyperplastic polyposis. *BMC Cancer* 6, 145, 2006
138. Bond GL, Menin C, Bertorelle R, Alhopuro P, Aaltonen LA & Levine AJ. MDM2 SNP309 Accelerates Colorectal Tumor Formation in Women. *Journal of Medical Genetics* 43, 950-952, 2006.
139. Alazzouzzi H, Suriano G, Guerra A, Plaja A, Espin E, Armengol M, Alhopuro P, Velho S, Shinomura Y, Gonzalez-Aguilera JJ, Yamamoto H, Aaltonen LA, Moreno V, Capella G, Peinado MA, Seruca R, Arango D & Schwartz S Jr. Tumor selection advantage of non-dominant negative P53 mutations in homozygous MDM2-SNP309 colorectal cancer cells. *Journal of Medical Genetics* 44, 75-80, 2007.
140. Karppinen S-M, Barkardottir RB, Backenhorn K, Sydenham T, Syrjäkoski K, Schleutker J, Ikonen T, Pylkäs K, Rapakko K, Erkkö H, Johannesdottir G, Gerdes A-M, Thomassen M, Agnarsson BA, Grip M, Kallioniemi A, Kere J, Aaltonen LA, Arason A, Møller P, Kruse TA, Borg Å & Winqvist R. Nordic collaborative study of the *BARD1* Cys557Ser allele in 3956 cancer cases: enrichment in familial *BRCA1/BRCA2* mutation negative breast cancer but not in other malignancies. *Journal of Medical Genetics* 43, 856-862, 2006.
141. Davalos V, Dopeso H, Castano J, Wilson AJ, Vilardell F, Romero-Gimenez J, Espin E, Armengol M, Capella G, Mariadason JM, Aaltonen LA, Schwartz S Jr & Arango D. *EPHB4* and survival in colorectal cancer patients. *Cancer Research* 66, 8943-8948, 2006.
142. Tuupanen S, Karhu A, Järvinen H, Mecklin JP, Launonen V & Aaltonen LA. No evidence for dual role of loss of heterozygosity in hereditary non-polyposis colorectal cancer. *Oncogene* 26, 2513-2517, 2007.
143. Aaltonen LA, Johns L, Järvinen H, Mecklin J-P, Houlston R. Explaining the excess familial risk of colorectal cancer associated with mismatch repair deficient and stable tumors. *Clinical Cancer Research* 13, 356-361, 2007.
144. Lehtonen HJ, Blanco I, Piulats JM, Herva R, Launonen V & Aaltonen LA. Conventional renal cancer in a patient with fumarate hydratase mutation. *Human Pathology* 38, 793-796, 2007.
145. Georgitsi M, Karhu A, Winqvist R, Visakorpi T, Waltering K, Vahteristo P, Launonen V & Aaltonen LA. Mutation analysis of *aryl hydrocarbon receptor interacting protein (AIP)* gene in colorectal, breast, and prostate cancers. *British Journal of Cancer* 96, 352-356, 2007.
146. Vanharanta S, Wortham NC, Langdorf C, El-Bahwary M, van der Spuy Z, Sjöberg J, Lehtinen R, Karhu A, Tomlinson IPM, Kiuru M, Arango D & Aaltonen LA. Definition of a minimal region of

deletion of chromosome 7 in uterine leiomyomas by tiling-path microarray CGH and mutation analysis of known genes in this region. **Genes, Chromosomes and Cancer** 46, 451-458, 2007.

147. Georgitsi M, Raitila A, Karhu A, Tuppurainen K, Mäkinen MJ, Vierimaa O, Paschke R, Saeger W, van der Luijt RB, Sane T, Robledo M, De Menis E, Weil RJ, Wasik A, Zielinski G, Lucewicz O, Lubinski J, Launonen V, Vahteristo P & Aaltonen LA. Molecular diagnosis of pituitary adenoma predisposition, caused by *aryl hydrocarbon receptor interacting protein* gene mutations **Proceedings of the National Academy of Sciences** 104, 4101-4105, 2007

148. Tuupanen S, Alhopuro P, Mecklin J-P, Järvinen H & Aaltonen LA. No evidence for association of *NOD2* R702W AND G908R with colorectal cancer. **International Journal of Cancer** 121, 76-79, 2007

149. Erkkö H, Xia B, Nikkilä J, Schleutker J, Syrjäkoski K, Mannermaa A, Kallioniemi A, Pylkäs K, Karppinen S-M, Rapakko K, Miron A, Mattila H, Bell DW, Haber DA, Grip M, Reiman M, Jukkola-Vuorinen A, Mustonen A, Kere J, Aaltonen LA, Kosma V-M, Kataja V, Soini Y, Drapkin RI, Livingston DM & Winqvist R. A recurrent mutation in *PALB2* in Finnish cancer families. **Nature** 446, 316 – 319, 2007.

150. Andersen CL, Schepeler T, Thorsen K, Birkenkamp-Demtröder K, Mansilla F, Aaltonen LA, Laurberg S & Orntoft T. Clusterin expression in normal mucosa and colorectal cancer. **Molecular and Cellular Proteomics** 6, 1039-1048, 2007.

151. Lehtonen HJ, Mäkinen MJ, Kiuru M, Laiho P, Herva R, van Minderhout I, Hogendoorn PCW, Cornelisse C, Devilee P, Launonen V & Aaltonen LA. Increased HIF1 in SDH and FH deficient tumors does not cause microsatellite instability. **International Journal of Cancer** 121, 1386-1389, 2007.

152. Sammalkorpi H, Alhopuro P, Lehtonen R, Tuimala J, Mecklin J-P, Järvinen HJ, Jiricny J, Karhu A, & Aaltonen LA. Background Mutation Frequency in Microsatellite-Unstable Colorectal Cancer. **Cancer Research** 67, 5691-5698, 2007.

153. Mansilla F, Birkenkamp-Demtröder K, Kruhoffer M, Sorensen FB, Andersen CL, Laiho P, Aaltonen LA, Verspaget HW & Orntoft TF. Differential expression of DHHC9 in microsatellite stable and instable human colorectal cancer subgroups. **British Journal of Cancer** 96, 1896-1903, 2007.

154. Raitila A, Georgitsi M, Karhu A, Tuppurainen K, Mäkinen MJ, Birkenkamp-Demtröder K, Salmenkivi K, Orntoft TF, Arola J, Launonen V, Vahteristo P & Aaltonen LA. No evidence of somatic AIP mutations in sporadic endocrine neoplasia. **Endocrine-Related Cancer** 14, 901-906, 2007.

155. Georgitsi M, Raitila A, Karhu A, van der Luijt RB, Aalfs CM, Sane T, Vierimaa O, Mäkinen MJ, Tuppurainen K, Paschke R, Gimm O, Koch CA, Gündogdu S, Lucassen A, Tischkowitz M, Izatt L, Aylwin S, Bano G, Hodgson S, De Menis E, Launonen V, Vahteristo P & Aaltonen LA. Germline *CDKN1B/p27^{Kip1}* mutation in multiple endocrine neoplasia. **Journal of Endocrinology and Metabolism** 92, 3321-3325, 2007.

156. Laakso M, Tuupanen S, Karhu A, Lehtonen R, Karhu A, Aaltonen LA & Hautaniemi S. Computational identification of candidate loci for recessively inherited mutations using high throughput SNP arrays. **Bioinformatics** 23, 1952-1961, 2007.

157. Niemelä AM, Hynninen P, Mecklin J-P, Kuopio T, Kokko A, Aaltonen L, Parkkila A-K, Pastorekova S, Pastorek J, Waheed A, Sly WS, Orntoft TF, Kruhoffer M, Haapasalo H, Parkkila S & Kivelä A. Carbonic anhydrase IX is highly expressed in hereditary non-polyposis colorectal cancer. ***Cancer Epidemiology, Biomarkers & Prevention*** 16, 1760-1766, 2007.
158. Vahteristo P, Kokko A, Saksela O, Aittomäki K & Aaltonen LA. Blood derived gene expression profiling in unraveling recessive disease susceptibility. ***Journal of Medical Genetics*** 44, 718-720, 2007.
159. Mecklin JP, Aarnio M, Läärä E, Kairaluoma MV, Pylvänäinen K, Peltomäki P, Aaltonen LA & Järvinen HJ. Development of colorectal tumours in regular colonoscopic surveillance in Lynch syndrome. ***Gastroenterology*** 133, 1093-1098, 2007.
160. Aktan-Collan K, Haukkala A, Pylvänäinen K, Järvinen HJ, Aaltonen LA, Peltomäki PT, Rantanen E, Kääriäinen H, & Mecklin JP Direct contact in inviting high-risk members of hereditary colon cancer families to genetic counselling and DNA-testing. ***Journal of Medical Genetics*** 44, 732-738, 2007.
161. Yang Q, Zhang R, Horikawa I, Fujita K, Afshar Y, Kokko A, Laiho P, Aaltonen LA & Harris CC. Functional Diversity of Human Protection of Telomeres 1 Isoforms in Telomere Protection and Cellular Senescence. ***Cancer Research*** 67, 11677-11686, 2007.
162. Lehtonen HJ, Ylisaukko-Oja SK, Kiuru M, Karhu A, Lehtonen R, Vanharanta S, Jalanko A, Aaltonen LA & Launonen V. Stress-induced expression of a novel variant of human fumarate hydratase (FH). ***Gene Expression*** 14, 59-69, 2007.
163. Birkenkamp-Demtröder K, Mansilla F, Sorensen FB, Kruhoffer M, Cabezon T, Christensen LL, Aaltonen LA, Verspaget HW & Orntoft TF. Phosphoprotein Keratin 23 accumulates in MSS but not MSI colon cancers in vivo and impacts viability and proliferation in vitro. ***Molecular Oncology*** 1, 181-195, 2007
164. Tuupanen S, Niittymäki I, Nousiainen K, Vanharanta S, Mecklin J-P, Nuorva K, Järvinen H, Hautaniemi S, Karhu A & Aaltonen LA. Allelic imbalance at rs6983267 suggests selection of the risk allele in somatic colorectal tumor evolution. ***Cancer Research*** 68, 14-17, 2008.
165. Ahvenainen T, Lehtonen HJ, Lehtonen R, Vahteristo P, Aittomäki K, Baynam G, Dommering C, Eng C, Gruber S, Grönberg H, Harvima R, Herva R, Hietala M, Kujala M, Kääriäinen H, Sunde L, Vierimaa O, Pollard PJ, Tomlinson IPM, Björck E, Aaltonen LA & Launonen V. Mutation screening of *fumarate hydratase* by MLPA: detection of exonic deletion in a patient with leiomyomatosis and renal cell cancer ***Cancer Genetics and Cytogenetics*** 183, 83-88, 2008.
166. Siltanen S, Syrjäkoski K, Fagerholm R, Ikonen T, Lipman P, Mallot J, Holli K, Tammela TLJ, Järvinen HJ, Mecklin J-P, Aittomäki K, Blomqvist C, Bailey-Wilson JE, Nevanlinna H, Aaltonen LA, Schleutker J & Vahteristo P. *ARLTS* germline variants and the risk for breast, prostate, and colorectal cancer. ***European Journal of Human Genetics*** 16, 983-991, 2008
167. Tomlinson IPM, Webb E, Carvajal-Carmona L, Broderick P, Howarth K, Pittman AM, Spain S, Lubbe S, Walther A, Sullivan K, Jaeger E, Fielding S, Rowan A, Vijayakrishnan J, Domingo E, Chandler

I, Kemp Z, Qureshi M, Farrington SM, Tenesa A, Prendergast JGD, Barnetson RA, Penegar S, Barclay E, Wood W, Martin L, Gorman M, Thomas H, Peto J, Bishop DT, Gray R, Maher ER, Lucassen A, Kerr D, Evans DGR, The CORGI Consortium, Schafmayer C, Buch S, Völzke H, Hampe J, Schreiber S, John U, Koessler T, Pharoah P, van Wezel T, Morreau H, Wijnen JT, Hopper JL, Southey MC, Giles GG, Severi G, Castellví-Bel S, Ruiz-Ponte C, Carracedo A, Castells A, The EPICOLON Consortium, Försti A, Hemminki K, Vodicka P, Naccarati A, Lipton L, Ho JWC, Cheng KK, Sham PC, Luk J, Agúndez JAG, Ladero JM, de la Hoya M, Caldés T, Niittymäki I, Tuupanen S, Karhu A, Aaltonen L, Cazier J-B, Campbell H, Dunlop MG & Houlston RS. A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. *Nature Genetics* 40, 623-630, 2008.

168. Alhopuro P, Phichith D, Tuupanen S, Sammalkorpi H, Nybondas M, Saharinen J, Robinson JP, Yang Z, Chen L-Q, Orntoft T, Mecklin J-P, Järvinen H, Eng C, Moeslein G, Shibata D, Houlston RS, Lucassen A, Tomlinson IPM, Launonen V, Ristimäki A, Arango D, Karhu A, Sweeney HL & Aaltonen LA. Unregulated smooth muscle myosin in human intestinal neoplasia. *Proceedings of the National Academy of Sciences* 105, 5513-5518, 2008.

169. Georgitsi M, De Menis E, Cannavò S, Mäkinen MJ, Tuppurainen K, Pauletto P, Curtò L, Weil RJ, Paschke R, Lubinski J, Vahteristo P, Karhu A & Aaltonen LA. Aryl hydrocarbon receptor interacting protein (AIP) gene mutation analysis in children and adolescents with sporadic pituitary adenomas. *Clinical Endocrinology* 69, 621-627, 2008.

170. Petrova TV, Nykänen A, Norrmén C, Ivanov KI, Andersson LC, Haglund C, Puolakkainen P, Wempe F, von Melchner H, Gradwohl G, Vanharanta S, Aaltonen LA, Saharinen J, Gentile M, Clarke A, Taipale J, Oliver G & Alitalo K. Transcription factor PROX1 induces colon cancer progression by promoting the transition from benign to highly dysplastic phenotype. *Cancer Cell* 13, 407-419, 2008.

171. Sammalkorpi S, Alhopuro P, Niittymäki I, Orntoft TF, Hokland P, Karhu A & Aaltonen LA. Mutation analysis of *MYH11* in acute myeloid leukemia. *Leukemia and Lymphoma* 49, 1829-1831, 2008.

172. Georgitsi M, Heliövaara E, Paschke R, Kumar AVK, Tischkowitz M, Vierimaa O, Salmela P, Sane T, De Menis E, Cannavò S, Gündogdu S, Lucassen A, Izatt L, Aylwin S, Bano G, Hodgson S, Koch CA, Karhu A & Aaltonen LA. Large genomic deletions in *AIP* in pituitary adenoma predisposition *Journal of Clinical Endocrinology and Metabolism* 93, 4146-4151, 2008.

173. Alberici P, Gaspar C, Franken P, Gorski MM, de Vries I, Scott RJ, Ristimäki A, Aaltonen LA & Fodde R. Smad4 haploinsufficiency: a matter of dosage. *PathoGenetics* 1:doi:10.1186/1755-8417-1-2, 2008.

174. Pittman, A.M., Webb, E., Carvajal-Carmona, L., Howarth, K., Di-Bernardo, M., Broderick, P., Spain, S., Walther, A., Price, A., Sullivan, K., Twiss, P., Fielding, S., Rowan, A., Jaeger, E., Vijaykrishnan, J., Chandler, I., Penegar, S., Qureshi, M., Lubbe, S., Domingo, E., Kemp, Z., Barclay, E., Wood, W., Martin, L., Gorman, M., Thomas, H., Peto, J., Bishop, T., Gray, R., Maher, E.R., Lucassen, A., Kerr, D., Evans, G.R., The CORGI Consortium, van Wezel, T., Morreau, H., Wijnen, J.T., Hopper, J.L., Southey, M.C., Giles, G.G., Severi, G., Castellvi-Bel, S., Ruiz-Ponte, C., Carracedo, A., Castells, A., The EPICOLON Consortium, Forsti, A., Hemminki, K., Vodicka, P., Naccarati, A., Lipton, L., Ho, J.W., Cheng, K.K., Sham, P.C., Luk, J., Agúndez, J.A., Ladero, J.M., de la Hoya, M., Caldes, T.,

Niittymäki, I., Tuupanen, S., Karhu, A., Aaltonen, L.A., Cazier, J.B., Tomlinson, I.P.M. & Houlston, R.S. Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer. **Human Molecular Genetics** 17, 3720-3727, 2008.

175. Alhopuro P, Karhu A, Winqvist R, Waltering K, Visakorpi T & Aaltonen LA. Somatic mutation analysis of *MYH11* in breast and prostate cancer. **BMC Cancer** 8, 263, 2008

176. Houlston RS, Webb E, Broderick P, Pittman AM, Di Bernardo MC, Lubbe S, Chandler I, Vijayakrishnan J, Sullivan K, Penegar S; Colorectal Cancer Association Study Consortium, Carvajal-Carmona L, Howarth K, Jaeger E, Spain SL, Walther A, Barclay E, Martin L, Gorman M, Domingo E, Teixeira AS; CoRGI Consortium, Kerr D, Cazier JB, Niittymäki I, Tuupanen S, Karhu A, Aaltonen LA, Tomlinson IP, Farrington SM, Tenesa A, Prendergast JG, Barnetson RA, Cetnarskyj R, Porteous ME, Pharoah PD, Koessler T, Hampe J, Buch S, Schafmayer C, Tepel J, Schreiber S, Völzke H, Chang-Claude J, Hoffmeister M, Brenner H, Zanke BW, Montpetit A, Hudson TJ, Gallinger S; International Colorectal Cancer Genetic Association Consortium, Campbell H, Dunlop MG. Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer. **Nature Genetics** 40, 1426-1435, 2008.

177. Jorissen RN, Lipton L, Gibbs P, Chapman M, Desai J, Jones IT, Yeatman TJ, East P, Tomlinson IP, Verspaget HW, Aaltonen LA, Kruhoffer M, Orntoft TF, Andersen CL, Sieber OM. DNA copy-number alterations underlie gene expression differences between microsatellite stable and unstable colorectal cancers. **Clinical Cancer Research** 14, 8061-8069, 2008.

178. Raitila A, Georgitsi M, Bonora E, Vargiolu M, Tuppurainen K, Mäkinen MJ, Vierimaa O, Salmela PI, Launonen V, Vahteristo P, Aaltonen LA, Romeo G & Karhu A. Aryl Hydrocarbon Receptor Interacting Protein (AIP) mutations seem not to associate with familial non-medullary thyroid cancer. **Journal of Endocrinological Investigation** 32, 426-429, 2009.

179. Raimundo N, Vanharanta S, Aaltonen LA, Hovatta I & Suomalainen A. Down-regulation of SRF-FOS-JUNB pathway in Fumarate Hydratase-deficiency and in uterine leiomyomas. **Oncogene** 28, 1261-1273, 2009.

180. Andersen CL, Christensen LL, Thorsen K, Schepeler T, Sørensen FB, Verspaget HW, Simon R, Kruhoffer M, Aaltonen LA, Laurberg S & Ørntoft TF. Dysregulation of the transcription factors SOX4, CBFB, and SMARCC1 correlates with outcome of colorectal cancer. **British Journal of Cancer** 100, 511-523, 2009.

181. Vierimaa O, Villablanca A, Alimov A, Georgitsi M, Raitila A, Vahteristo P, Larsson C, Ruokonen A, Eloranta E, Ebeling TML, Ingnatius J, Aaltonen LA, Leisti J & Salmela PI. Mutation analysis of MEN1, HRPT2, CASR, CDKN1B and AIP genes in primary hyperparathyroidism patients with features of genetic predisposition **Journal of Endocrinological Investigation** 32, 426-429, 2009.

182. Koski TA, Lehtonen HJ, Jee KJ, Ninomiya S, Joosse SA, Vahteristo P, Kiuru M, Karhu A, Sammalkorpi H, Vanharanta S, Lehtonen R, Edgren H, Nederlof PM, Hietala M, Aittomäki K, Herva R, Knuutila S, Aaltonen LA & Launonen V. Array Comparative Genomic Hybridization Identifies a Distinct DNA Copy Number Profile in Renal Cell Cancer Associated with Hereditary Leiomyomatosis and Renal Cell Cancer. **Genes Chromosomes & Cancer** 48, 544—551, 2009.

183. Järvinen HJ, Renkonen-Sinisalo L, Aktán-Collán K, Peltomäki P, Aaltonen LA & Mecklin J-P. Ten Years after Mutation Testing for Lynch Syndrome: Cancer Incidence and Outcome in Mutation-positive and Mutation-negative Family Members. *Journal of Clinical Oncology* 27, 4793-4797, 2009.
184. Tuupanen S, Turunen M, Lehtonen R, Hallikas O, Vanharanta S, Kivioja T, Björklund M, Wei G, Yan J, Niittymäki I, Mecklin J-P, Järvinen H, Ristimäki A, Di-Bernardo M, East P, Carvajal-Carmona L, Houlston RS, Tomlinson I, Palin K, Ukkonen E, Karhu A, Taipale J & Aaltonen LA. The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. *Nature Genetics* 41, 885-890, 2009.
185. Heliövaara E, Raitila A, Launonen V, Paetau A, Arola J, Lehtonen H, Sane T, Weil RJ, Vierimaa O, Salmela P, Tuppurainen K, Mäkinen M, Aaltonen LA, & Karhu A. The expression of AIP-related molecules in elucidation of cellular pathways in pituitary adenomas. *American Journal of Pathology* 175, 2501-2507, 2009.
186. Yuan Z, Shin J, Wilson A, Goel S, Ling Y-H, Ahmed N, Dopeso H, Jhaver M, Nasser S, Montagna C, Fordyce K, Augenlicht LH, Aaltonen LA, Arango D, Weber TK & Mariadason JM. An A13 repeat within the 3'UTR of EGFR is frequently mutated in MSI colon cancers and is associated with increased EGFR expression. *Cancer Research* 69, 7811-7818, 2009.
187. Jennings J, Georgitsi M, Holdaway I, Daly A, Tichomirowa M, Beckers A, Aaltonen LA, Karhu A, & Cameron F. Aggressive pituitary adenomas occurring in young patients in a large Polynesian kindred with a germline R271W mutation in the AIP gene. *European Journal of Endocrinology* 161, 799-804, 2009.
188. Jorissen RN, Gibbs P, Christie M, Prakash S, Lipton L, Desai J, Kerr D, Aaltonen LA, Arango D, Kruhøffer M, Ørntoft TF, Andersen CL, Gruidl M, Kamath VP, Eschrich S, Yeatman TJ & Sieber OM. Metastasis-associated gene expression changes predict poor outcomes in patients with Dukes' stage B and C colorectal cancer. *Clinical Cancer Research* 15, 7642-7651, 2009.
189. Vahteristo P, Koski TA, Näätsaari L, Kiuru M, Karju A, Herva R, Sallinen S-L, Vierimaa O, Björck E, Richard S, Gardie B, Bessis D, Van Glabeke E, Blanco I, Houlston R, Senter L, Hietala M, Aittomäki K, Aaltonen LA, Launonen V & Lehtonen R. No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. *Familial Cancer* 9, 245-251, 2010.
190. Velho S, Oliveira C, Paredes J, Sousa S, Leite M, Matos P, Milanezi F, Ribeiro AS, Mendes N, Licastro D, Karhu A, Oliveira MJ, Ligtenberg M, Hamelin R, Carneiro F, Lindblom A, Peltomäki P, Castedo S, Schwartz Jr S, Jordan P, Aaltonen LA, Hofstra RMW, Suriano G, Stupka E, Fialho AM & Seruca R. *Mixed lineage kinase 3* gene mutations in mismatch repair deficient gastrointestinal tumours. *Human Molecular Genetics* 19, 697-796, 2010.
191. Niittymäki I, Kaasinen E, Tuupanen S, Karhu A, Järvinen H, Mecklin JP, Tomlinson IP, Di Bernardo MC, Houlston RS & Aaltonen LA. Low-penetrance susceptibility variants in familial colorectal cancer. *Cancer Epidemiology, Biomarkers and Prevention* 19, 1478-1483, 2010.

192. Kaasinen E, Aittomäki K, Eronen M, Vahteristo P, Karhu A, Mecklin J-P, Kajantie E, Aaltonen LA & Lehtonen R. Recessively Inherited Right Atrial Isomerism Caused by Mutations in *Growth/Differentiation Factor 1 (GDF1)*. **Human Molecular Genetics** 19, 2747-2753, 2010.
193. Alhopuro P, Bjorklund M, Sammalkorpi H, Turunen M, Tuupanen S, Biström M, Niittymäki I, Lehtonen HJ, Kivioja T, Launonen V, Saharinen J, Nousiainen K, Hautaniemi S, Nuorva K, Mecklin J-P, Järvinen H, Orntoft T, Arango D, Lehtonen R, Karhu A, Taipale J & Aaltonen LA. Mutations in the Circadian Gene *CLOCK* in Colorectal Cancer. **Molecular Cancer Research** 8, 952-960, 2010.
194. Gylfe AE, Sirkiä J, Ahlsten M, Järvinen H, Mecklin J-P, Karhu A & Aaltonen LA. Somatic mutations and germline sequence variants in patients with familial colorectal cancer. **International Journal of Cancer** 127, 2974-2980, 2010.
195. Daly AF, Tichomirowa MA, Petrossians P, Heliövaara E, Jaffrain-Rea ML, Barlier A, Naves LA, Ebeling T, Karhu A, Raappana A, Cazabat L, De Menis E, Montañana CF, Raverot G, Weil RJ, Sane T, Maiter D, Neggers S, Yaneva M, Tabarin A, Verrua E, Eloranta E, Murat A, Vierimaa O, Salmela PI, Emy P, Toledo RA, Sabaté MI, Villa C, Popelier M, Salvatori R, Jennings J, Ferrandez Longás A, Labarta Aizpún JI, Georgitsi M, Paschke R, Ronchi C, Valimaki M, Saloranta C, De Herder W, Cozzi R, Guitelman M, Magri F, Lagonigro MS, Halaby G, Corman V, Hagelstein MT, Vanbellinghen JF, Barra GB, Gimenez-Roqueplo AP, Cameron FJ, Borson-Chazot F, Holdaway I, Toledo SP, Stalla GK, Spada A, Zacharieva S, Bertherat J, Brue T, Bours V, Chanson P, Aaltonen LA & Beckers A. Clinical characteristics and therapeutic responses in patients with germ-line AIP mutations and pituitary adenomas: An international collaborative study. **Journal of Clinical Endocrinology and Metabolism** 95, E373-E383, 2010.
196. Allelic variation at the 8q23.3 colorectal cancer risk locus functions as a cis-acting regulator of *EIF3H*. Pittman AM, Naranjo S, Jalava SE, Twiss P, Ma Y, Olver B, Lloyd A, Vijayakrishnan J, Qureshi M, Broderick P, van Wezel T, Morreau H, Tuupanen S, Aaltonen LA, Alonso ME, Manzanares M, Gavilán A, Visakorpi T, Gómez-Skarmeta JL & Houlston RS. **PLoS Genetics** 6, e1001126, 2010.
197. Raitila A, Lehtonen HJ, Arola J, Heliövaara E, Ahlsten M, Georgitsi M, Jalanko A, Paetau A, Aaltonen LA & Karhu A. Mice with Inactivation of *Aryl Hydrocarbon Receptor Interacting Protein (Aip)* Display Complete Penetrance of Pituitary Adenomas with aberrant ARNT Expression. **American Journal of Pathology** 177, 1969-1976, 2010.
198. Houlston RS, Cheadle J, Dobbins SE, Tenesa A, Jones AM, Howarth K, Spain SL, Broderick P, Domingo E, Farrington S, Prendergast JGD, Pittman AM, Theodoratou E, Smith CG, Olver B, Walther A, Barnetson RA, Churchman M, Jaeger EEM, Penegar S, Barclay E, Martin L, Gorman M, Mager R, Johnstone E, Midgley R, Niittymäki I, Tuupanen S, Colley J, Idziaszczyk S, The COGENT Consortium, Thomas HJM, Lucassen AM, Evans DGR, Maher ER, The CORGI Consortium, The COIN Collaborative Group, The COINB Collaborative Group, Maughan T, Dimas A, Dermizakis E, Cazier J-B, Aaltonen LA, Pharoah P, Kerr DJ, Carvajal-Carmona LG, Campbell H, Dunlop MG & Tomlinson IPM. Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33. **Nature Genetics** 42, 973-977, 2010.
199. Theodoratou E, Campbell H, Tenesa A, Houlston R, Webb E, Lubbe S, Broderick P, Gallinger S, Croitoru EM, Jenkins MA, Win,⁷ Sean P. Cleary,⁶ Thibaud Koessler,⁸ Paul D. Pharoah,⁸ Sébastien Küry

- AK, Bézieau S, Buecher B, Ellis NA, Peterlongo P, Offit K, Aaltonen LA, Enholm S, Lindblom A, Zhou X-L, Tomlinson IP, Moreno V, Blanco I, Capellà G, Barnetson R, Porteous ME, Dunlop MG & Farrington SM. A large-scale meta-analysis to refine colorectal cancer risk estimates associated with *MUTYH* variants. *British Journal of Cancer* 103, 1875-1884, 2010.
200. Ashrafian H, O'Flaherty L, Adam J, Steeples V, Chung Y-L, East P, Vanharanta S, Lehtonen H, Nye E, Hatipoglu E, Miranda M, Howarth K, Shukla D, Troy H, Griffiths JR, Spencer-Dene B, Yusuf M, Volpi E, Maxwell P, Stamp G, Poulson R, Pugh C, Costa B, Bardella C, Di Renzo MF, Kotlikoff M, Launonen V, Aaltonen LA, El-Bahrawy M, Tomlinson I & Pollard P. Fumarate Hydratase Deficiency Promotes An Oncogenic Metabolic Milieu Reminiscent of the Warburg Effect. *Cancer Research* 70, 9153-9165, 2010.
201. Heliövaara E, Tuupanen S, Ahlsten M, Hodgson S, de Menis E, Kuusmin O, Izatt L, McKinlay Gardner RJ, Gundogdu S, Lucassen A, Arola J, Tuomisto A, Mäkinen M, Karhu A & Aaltonen LA. No evidence of *RET* germline mutations in familial pituitary adenoma. *Journal of Molecular Endocrinology* 46, 1-8, 2011.
202. Knappskog S, Bjørnslett M, Myklebust L, Huijts P, Vreeswijk MP, Edvardsen H, Guo Y, Zhang X, Yang M, Ylisaukko-oja SK, Alhopuro P, Arola J, Tollenaar RAEM, van Asperen CJ, Seynaeve C, Staalesen V, Chiranthar R, Løkkevik E, Salvesen H, Evans G, Newman W, Lin D, Aaltonen LA, Børresen-Dale A-L, Tell G, Stoltenberg C, Romundstad P, Hveem K, Lillehaug JR, Vatten L, Devilee P, Dørum A & Lønning PE. The *MDM2* promoter SNP285C/309G haplotype diminish Sp1 transcription factor binding and reduces risk for breast and ovarian cancer in Caucasians. *Cancer Cell* 19, 273-282, 2011.
- 203 Stefanius K, Kantola T, Tuomisto A, Vahteristo P, Karttunen T, Aaltonen LA, Mäkinen M & Karhu A. Downregulation of the hedgehog receptor *PTCH1* in colorectal serrated adenocarcinomas is not caused by *PTCH1* mutations *Virchows Archiv* 458, 213-219, 2011.
204. Niittymäki I, Gylfe A, Laine L, Laakso M, Lehtonen H, Sirkiä J, Tolvanen J, Nousiainen K, Pouwels J, Järvinen H, Nuorva K, Mecklin JP, Mäkinen M, Ristimäki A, Ørntoft TF, Hautaniemi S, Karhu A, Kallio MJ & Aaltonen LA. High frequency of *TTK* mutations in microsatellite-unstable colorectal cancer and evaluation of their effect on spindle-assembly checkpoint. *Carcinogenesis* 32, 305-311, 2011.
205. Niittymäki I, Tuupanen S, Li Y, Järvinen H, Mecklin JP, Tomlinson IPM, Houlston RS, Karhu A & Aaltonen LA. Systematic search for enhancer elements and somatic allelic imbalance at seven low-penetrance colorectal cancer predisposition loci. *BMC Medical Genetics* 2011 Feb; 12:23.
206. Alhopuro P, Sammalkorpi H, Niittymäki I, Biström M, Raitila A, Saharinen J, Nousiainen K, Lehtonen HJ, Heliövaara E, Puhakka J, Tuupanen S, Sousa S, Seruca R, Ferreira AM, Hofstra RMW, Mecklin JP, Järvinen H, Ristimäki A, Ørntoft TF, Hautaniemi S, Arango D, Karhu A & Aaltonen LA. Candidate driver genes in microsatellite-unstable colorectal cancer. *International Journal of Cancer* 130, 1558-1566, 2012.
207. Saarinen S, Vahteristo P, Launonen V, Franssila K, Kivirikko S, Lehtonen R, Bain BJ, Bauduer F, Unal A, Aaltonen LA & Aittomäki K. Analysis of *KLHDC8B* in familial nodular lymphocyte predominant Hodgkin lymphoma. *British Journal of Haematology* 154, 413-415, 2011.

208. Saarinen S, Aavikko M, Aittomäki K, Launonen V, Lehtonen R, Franssila K, Lehtonen HJ, Kaasinen E, Broderick P, Tarkkanen J, Bain BJ, Bauduer F, Ünal A, Swerdlow AJ, Cooke R, Mäkinen MJ, Houlston R, Vahteristo P & Aaltonen LA. Exome sequencing reveals germline *NPAT* mutation as a candidate risk factor for Hodgkin lymphoma. *Blood* 118, 493-498, 2011.
209. Bardella C, El-Bahrawy M, Frizzell N, Adam J, Ternette N, Hatipoglu E, Howarth K, O'Flaherty L, Roberts I, Turner G, Taylor J, Giaslakiotis K, Macaulay VM, Harris AL, Chandra A, Lehtonen HJ, Launonen V, Aaltonen LA, Pugh CW, Mihai R, Trudgian D, Kessler B, Baynes JW, Ratcliffe PJ, Tomlinson IP & Pollard PJ. Aberrant succination of proteins in fumarate hydratase-deficient mice and HLRCC patients is a robust biomarker of mutation status. *Journal of Pathology* 225, 4-11, 2011.
210. Tomlinson IP, Carvajal-Carmona LG, Dobbins SE, Tenesa A, Jones AM, Howarth K, Palles C, Broderick P, Jaeger EE, Farrington S, Lewis A, Prendergast JG, Pittman AM, Theodoratou E, Olver B, Walker M, Penegar S, Barclay E, Whiffin N, Martin L, Ballereau S, Lloyd A, Gorman M, Lubbe S; COGENT Consortium; CORGI Collaborators; EPICOLON Consortium, Howie B, Marchini J, Ruiz-Ponte C, Fernandez-Rozadilla C, Castells A, Carracedo A, Castellvi-Bel S, Duggan D, Conti D, Cazier JB, Campbell H, Sieber O, Lipton L, Gibbs P, Martin NG, Montgomery GW, Young J, Baird PN, Gallinger S, Newcomb P, Hopper J, Jenkins MA, Aaltonen LA, Kerr DJ, Cheadle J, Pharoah P, Casey G, Houlston RS & Dunlop MG. Multiple common susceptibility variants near BMP pathway loci *GREM1*, *BMP4*, and *BMP2* explain part of the missing heritability of colorectal cancer. *PLoS Genetics* Jun;7(6):e1002105, 2011.
211. Coissieux M-M, Tomsic J, Castets M, Hampel H, Tuupanen S, Andrieu N, Comeras I, Drouet Y, Lasset C, Liyanarachchi S, Mazelin L, Puisieux A, Saurin J-C, Scoazec J-Y, Wang Q, Aaltonen L, Tanner SM, Chapelle A, Bernet A & Mehlen P. Variants in the Netrin-1 Receptor *UNC5C* Prevent Apoptosis and Increase Risk for Familial Colorectal Cancer *Gastroenterology* in 141, 2039-2046, 2011.
212. Mäkinen N, Mehine M, Tolvanen J, Kaasinen E, Li Y, Lehtonen HJ, Gentile M, Yan J, Enge M, Taipale M, Aavikko M, Katainen R, Virolainen E, Böhling T, Koski TA, Launonen V, Sjöberg J, Taipale J, Vahteristo P & Aaltonen LA. *MED12*, the *Mediator Complex Subunit 12* Gene, Is Mutated at High Frequency in Uterine Leiomyomas. *Science* 334, 252-255, 2011.
213. Rafnar T, Gudbjartsson DF, Sulem P, Jonasdottir A, Sigurdsson A, Jonasdottir A, Besenbacher S, Lundin P, Stacey SN, Gudmundsson J, Magnusson OT, le Roux L, Orlygsdottir G, Helgadottir H, Johannsdottir H, Gylfason A, Tryggvadottir L, Jonasson JG, De Juan A, Ortega E, Ramon-Cajal JM, García-Prats MD, Mayordomo C, Panadero A, Rivera F, Aben KKH, van Altena AM, Massuger LFAG, Aavikko M, Kujala PM, Staff S, Aaltonen LA, Olafsdottir K, Bjornsson J, Kong A, Salvarsdottir A, Saemundsson H, Olafsson K, Benediksdottir KR, Gulcher J, Masson G, Kiemeny LA, Mayordomo JI, Thorsteinsdottir U & Stefansson K. Mutations in *BRIP1* confer high risk of ovarian cancer. *Nature Genetics* 43, 1104-1107, 2011.
214. Spain SL, Carvajal-Carmona LG, Howarth KM, Jones AM, Su Z, Cazier JB, Williams J, Aaltonen LA, Pharoah P, Kerr DJ, Cheadle J, Li L, Casey G, Vodicka P, Sieber O, Lipton L, Gibbs P, Martin NG, Montgomery GW, Young J, Baird PN, Morreau H, van Wezel T, Ruiz-Ponte C, Fernandez-Rozadilla C, Carracedo A, Castells A, Castellvi-Bel S, Dunlop M, Houlston RS & Tomlinson IP. Refinement of the associations between risk of colorectal cancer and polymorphisms on chromosomes 1q41 and 12q13.13.

Human Molecular Genetics 21, 934-946, 2012.

215. Mäkinen N, Heinonen HR, Moore S, Tomlinson IPM, van der Spuy ZM & Aaltonen LA. *MED12* exon 2 mutations are common in uterine leiomyomas from South African patients **Oncotarget** 2, 966-969, 2011.

216. Mazzolini R, Dopeso H, Mateo-Lozano S, Chang W, Rodrigues P, Bazzocco S, Alazzouzi H, Landolfi S, Hernandez-Losa J, Alhopuro P, Espín E, Armengo M, Tabernero J, Ramón y Cajal S, Kloor M, Gebert J, Mariadason JM, Schwartz Jr S, Aaltonen LA, Mooseker MS & Arango D. Brush border Myosin Ia has tumor suppressor activity in the intestine **Proceedings of the National Academy of Sciences** 109, 1530-1535, 2012.

217. Arango D, Al-Obaidi S, Williams D, Dopeso J, Mazzolini R, Corner G, Byun D-S, Carr AA, Murone C, Tögel L, Zeps N, Aaltonen LA, Iacopetta B & Mariadason JM. *Villin* expression is frequently lost in poorly differentiated colon cancer. **American Journal of Pathology** 180, 1509-1521, 2012

218. Tuupanen S, Yan J, Turunen M, Gylfe AE, Kaasinen E, Li L, Eng C, Culver DA, Kalady MF, Pennison MJ, Pasche B, Manne U, de la Chapelle A, Hampel H, Henderson BE, Le Marchand L, Hautaniemi S, Askhtorab H, Smoot D, Sandler RS, Keku T, Kupfer SS, Ellis NA, Haiman CA, Taipale J & Aaltonen LA. Characterization of the colorectal cancer associated enhancer MYC-335 at 8q24: the role of rs67491583. **Cancer Genetics** 205, 25-33, 2012.

219. Karinen S, Saarinen S, Lehtonen R, Rastas P, Vahteristo P, Aaltonen LA & Hautaniemi S. Rule-based induction method for haplotype comparison and identification of candidate disease loci **Genome Medicine** 4, 21, 2012.

220. Dunlop MG, Tenesa A, Farrington SM, Ballereau S, Brewster DH, Kossler T, Pharoah P, Schafmayer C, Hampe J, Völzke H, Chang-Claude J, Hoffmeister M, Brenner H, von Holst S, Picelli S, Lindblom A, Jenkins MA, Hopper JL, Casey G, Duggan D, Newcomb PA, Abulí A, Bessa X, Ruiz-Ponte C, Castellvi-Bel S, Niittymäki I, Tuupanen S, Karhu A, Aaltonen L, Zanke B, Hudson T, Gallinger S, Barclay E, Martin L, Gorman M, Carvajal-Carmona L, Walther A, Kerr D, Lubbe S, Broderick P, Chandler I, Pittman A, Penegar S, Campbell H, Tomlinson I & Houlston RS. Cumulative impact of common genetic variants and other risk factors on colorectal cancer risk in 42 103 individuals **Gut** 62, 871-881, 2013.

221. Dunlop MG, Dobbins SE, Farrington SM, Jones AM, Palles C, Whiffin N, Tenesa A, Spain S, Broderick P, Ooi L-Y, Domingo E, Smillie C, Henrion M, Frampton M, Martin L, Grimes G, Gorman M, Semple C, Ma Y, Barclay E, Prendergast J, Cazier J-B, Olver B, Carvajal-Carmona LG, Ballereau S, Lloyd A, Vijayakrishnan J, Zgaga L, Rudan I, Theodoratou E, The CORGI Consortium, Starr JM, Deary I, Kirac I, Kovačević D, Aaltonen LA, Renkonen-Sinisalo L, Mecklin J-P, Matsuda K, Nakamura Y, Okada Y, Gallinger S, Duggan DJ, Conti D, Newcomb P, Hopper J, Jenkins MA, Schumacher F, Casey G, Easton D, Shah M, Pharoah P, Lindblom A, Liu T, The Swedish Low-Risk Colorectal Cancer Study Group, Smith CG, West H, Cheadle JP, The COIN Collaborative Group, Midgley R, Kerr DJ, Campbell H, Tomlinson IP & Houlston RS. Common variation near CDKN1A, POLD3 and SHROOM2 influences colorectal cancer risk. **Nature Genetics** 44, 770-776, 2012.

222. Ogundiran T, Tuupanen S, Aaltonen LA, Akarolo-Anthony S & Adebamowo C. delGA (rs67491583) Variant and Colorectal Cancer Risk in an Indigenous African Population. *African Journal of Medicine and Medical Sciences* 41, 271-275, 2012.
223. Aavikko M, Li S-P, Saarinen S, Alhopuro P, Kaasinen E, Morgunova E, Li Y, Vesanen K, Smith MJ, DGR Evans, Pöyhönen M, Kiuru A, Auvinen A, Aaltonen LA, Taipale J & Vahteristo P. Loss of SUFU function in familial multiple meningioma. *American Journal of Human Genetics* 91, 520-526, 2012.
224. Lehtonen HJ, Sipponen T, Tojkander S, Karikoski R, Järvinen H, Laing NG, Lappalainen P, Aaltonen LA & Tuupanen S. Segregation of a Missense Variant in Enteric Smooth Muscle Actin γ -2 with Autosomal Dominant Familial Visceral Myopathy. *Gastroenterology* 143, 1482-1491, 2012.
225. Kämpjärvi K, Mäkinen N, Kilpivaara O, Arola J, Heinonen H-R, Böhm J, Abdel-Wahab O, Lehtonen HJ, Pelttari LM, Mehine M, Schrewe H, Nevanlinna H, Levine RL, Hokland P, Böhling T, Mecklin J-P, Bützow R, Aaltonen LA & Vahteristo P. Somatic *MED12* mutations in uterine leiomyosarcoma and colorectal cancer. *British Journal of Cancer* 107, 1761-1765, 2012.
226. Moreira L, Balaguer F, Lindor N, Chapelle A, Hampel H, Aaltonen LA, Hopper JL, Le Marchand L, Gallinger S, Newcomb PA, Haile R, Thibodeau SN, Gunawardena S, Jenkins MA, Buchanan DD, Potter JD, Baron JA, Ahnen DJ, Moreno V, Andreu M, Ponz de Leon M, Rustgi AK, the EPICOLON Consortium & Castells A. Identification of Lynch syndrome among patients with colorectal cancer. *Journal of the American Medical Association* 308, 1555-1565, 2012.
227. Saarinen S, Pukkala E, Vahteristo P, Mäkinen MJ, Franssila K & Aaltonen LA. High Familial Risk in Nodular Lymphocyte Predominant Hodgkin Lymphoma. *Journal of Clinical Oncology* 31, 938-943, 2013.
228. Sur IK, Hallikas O, Vähärautio A, Yan J, Turunen M, Enge M, Taipale M, Karhu A, Aaltonen LA & Taipale J, Mice Lacking a Myc Enhancer Element that Includes Human SNP rs6983267 Are Resistant to Intestinal Tumors. *Science* 338, 1360-1363, 2012.
229. Pelttari LM, Nurminen R, Gylfe A, Aaltonen LA, Schleutker J & Nevanlinna H. Screening of Finnish RAD51C Founder Mutations in Prostate and Colorectal Cancer Patients. *BMC Cancer* 12:552, 2012.
230. Laitinen VH, Wahlfors T, Saaristo L, Rantapero T, Pelttari LM, Kilpivaara O, Laasanen S-L, Kallioniemi A, Nevanlinna H, Aaltonen LA, Vessella RL, Auvinen A, Visakorpi T, Tammela TLJ & Schleutker J. HOXB13 G84E mutation in Finland; population-based analysis of prostate, breast and colorectal cancer risk. *Cancer Epidemiology, Biomarkers & Prevention* 22, 452-460, 2013.
231. Kaasinen E, Aavikko M, Vahteristo P, Patama T, Li Y, Saarinen S, Kilpivaara O, Pitkänen E, Knekt P, Laaksonen M, Artama M, Lehtonen R, Aaltonen LA & Pukkala E. Nationwide registry-based analysis of cancer clustering detects strong familial occurrence of Kaposi sarcoma. *PLoS One* 8:e55209, 2013.
232. Ngeow J, Heald B, Rybicki LA, Orloff MS, Chen JL, Liu X, Yerian L, Willis J, Lehtonen, Rainer Lehtonen, Mester JL, Moline J, Burke CA, Church J, Aaltonen LA & Eng C. Prevalence of Germline

PTEN, BMPR1A, SMAD4, STK11, and ENG Mutations in Patients with Moderate-Load Colorectal Polyps. *Gastroenterology* 144, 1402-1409, 2013.

233. Tanskanen T, Gylfe AE, Katainen R, Taipale M, Renkonen-Sinisalo L, Mecklin J-P, Järvinen H, Tuupanen S, Kilpivaara O, Vahteristo P & Aaltonen LA. Exome Sequencing in Diagnostic Evaluation of Colorectal Cancer Predisposition in Young Patients. *Scandinavian Journal of Gastroenterology* 48, 672-678, 2013.

234. Saarinen S, Kaasinen E, Karjalainen-Lindsberg M-L, Vesanen K, Aavikko M, Katainen R, Taskinen M, Kytölä S, Leppä S, Hietala M, Vahteristo P & Aaltonen LA. Primary mediastinal large B-cell lymphoma segregating in a family: exome sequencing identifies MLL as a candidate predisposition gene. *Blood* 121, 3428-3430, 2013.

235. Raizis AM, Van Mater D, Aaltonen LA, Lohmann D, Cheale MS, Bickley VM, George PM, Zhou Y & Rosoff PM. Trilateral retinoblastoma in a patient with Peutz-Jeghers syndrome. *American Journal of Medical Genetics* 161, 1096-1100, 2013.

236. Mehine M, Kaasinen E, Mäkinen N, Katainen R, Kämpjärvi K, Pitkänen E, Heinonen H-R, Bützow R, Kilpivaara O, Kuosmanen A, Ristolainen H, Gentile M, Sjöberg J, Vahteristo P & Aaltonen LA. Characterization of Uterine Leiomyomas by Whole Genome Sequencing. *New England Journal of Medicine* 369, 453-463, 2013.

237. Gylfe AE, Kondelin J, Turunen M, Ristolainen H, Katainen R, Pitkänen E, Kaasinen E, Rantanen V, Tanskanen T, Varjosalo M, Lehtonen H, Palin K, Taipale M, Taipale J, Renkonen-Sinisalo L, Järvinen H, Böhm J, Mecklin J-P, Ristimäki A, Kilpivaara O, Tuupanen S, Karhu A, Vahteristo P & Aaltonen LA. Identification of candidate oncogenes discovered in human colorectal cancers with microsatellite instability. *Gastroenterology* 145, 540-543, 2013.

238. Mäkinen N, Vahteristo P, Bützow R, Sjöberg J & Aaltonen LA. Exomic landscape of MED12 mutation negative and positive uterine leiomyomas. *International Journal of Cancer* 134, 1008-1012, 2014.

239. Gylfe AE, Katainen R, Kondelin J, Tanskanen T, Cajuso T, Hänninen U, Taipale J, Taipale M, Renkonen-Sinisalo L, Järvinen H, Mecklin J-P, Kilpivaara O, Pitkänen E, Vahteristo P, Tuupanen S, Karhu A & Aaltonen LA. Eleven Candidate Susceptibility Genes for Common Familial Colorectal Cancer. *PLOS Genetics* 9:e1003876, 2013.

240. Mäkinen N, Heinonen H-R, Sjöberg J, Taipale J, Vahteristo P, & Aaltonen LA. Mutation analysis of components of the Mediator kinase module in MED12 mutation-negative uterine leiomyomas. *British Journal of Cancer* 110, 2246-2249, 2014.

241. Pitkänen E, Cajuso T, Katainen R, Kaasinen E, Välimäki N, Palin K, Taipale J, Aaltonen LA & Kilpivaara O. Frequent L1 retrotranspositions originating from TTC28 in colorectal cancer. *Oncotarget* 5, 853-859, 2014.

242. Turunen M, Spaeth JM, Keskitalo S, Park MJ, Kivioja T, Clark AD, Mäkinen N, Gao F, Palin K, Vähärautio A, Aavikko M, Kämpjärvi K, Nurkkala H, Vahteristo P, Kim CA, Aaltonen LA, Varjosalo

M, Taipale J & Boyer TG. Uterine leiomyoma-linked MED12 mutations disrupt Mediator-associated CDK activity. *Cell Reports*, 7, 654-660, 2014.

243. Heinonen H-R, Sarvilinna NS, Sjöberg J, Kämpjärvi K, Pitkänen E, Vahteristo P, Mäkinen N & Aaltonen LA. MED12 mutation frequency in unselected sporadic uterine leiomyomas. *Fertility and Sterility* 102, 1137-1142, 2014.

244. Tuupanen S, Hänninen UA, Kondelin J, von Nandelstadh P, Cajuso T, Gylfe AE, Katainen R, Tanskanen T, Ristolainen H, Böhm J, Mecklin JP, Järvinen H, Renkonen-Sinisalo L, Andersen CL, Taipale M, Taipale J, Vahteristo P, Lehti K, Pitkänen E & Aaltonen LA. Identification of 33 candidate oncogenes by screening for base-specific mutations. *British Journal of Cancer* 111, 1657-1662, 2014.

245. Kaasinen E, Rahikkala E, Koivunen P, Miettinen S, Wamelink MM, Aavikko M, Palin K, Myllyharju J, Moilanen JS, Pajunen L, Karhu A & Aaltonen LA. Clinical characterization, genetic mapping and whole-genome sequence analysis of a novel autosomal recessive intellectual disability syndrome. *European Journal of Medical Genetics* 57, 543-551, 2014.

246. Ferreira AM, Tuominen I, van Dijk-Bos K, Sanjabi B, van der Sluis T, van der Zee AG, Hollema H, Zazula M, Sijmons RH, Aaltonen LA, Westers H & Hofstra RM. High frequency of RPL22 mutations in microsatellite-unstable colorectal and endometrial tumors. *Human Mutation* 35, 1442-1445, 2014.

247. Nummela P, Saarinen L, Thiel A, Järvinen P, Lehtonen R, Lepistö A, Järvinen H, Aaltonen LA, Hautaniemi S & Ristimäki A. Genomic profile of pseudomyxoma peritonei analyzed using next-generation sequencing and immunohistochemistry. *International Journal of Cancer* 136, E282-289, 2015.

248. Knappskog S, Gansmo LB, Dibirova K, Metspalu A, Cybulski C, Peterlongo P, Aaltonen L, Vatten L, Romundstad P, Hveem K, Devilee P, Evans GD, Lin D, Van Camp G, Manolopoulos VG, Osorio A, Milani L, Ozcelik T, Zalloua P, Mouzaya F, Bliznetz E, Balanovska E, Pocheshkova E, Kučinskas V, Atramentova L, Nymadawa P, Titov K, Lavryashina M, Yusupov Y, Bogdanova N, Koshel S, Zamora J, Wedge DC, Charlesworth D, Dörk T, Balanovsky O & Lønning PE. Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget* 5, 8223-8234, 2014.

249. Ferreira AM, Tuominen I, Sousa S, Gerbens F, van Dijk-Bos K, Osinga J, Kooi KA, Sanjabi B, Esendam C, Oliveira C, Terpstra P, Hardonk M, van der Sluis T, Zazula M, Stachura J, van der Zee AG, Hollema H, Sijmons RH, Aaltonen LA, Seruca R, Hofstra RM & Westers H. New Target Genes in Endometrial Tumors Show a Role for the Estrogen-Receptor Pathway in Microsatellite Unstable Cancers. *Human Mutation* 35, 1514-1523, 2015.

250. Aavikko M, Kaasinen E, Nieminen JK, Byun M, Donner I, Mancuso R, Ferrante P, Clerici M, Brambilla L, Tournalaki A, Sarid R, Guttman-Yassky E, Taipale M, Morgunova E, Pekkonen P, Ojala P, Pukkala E, Casanova JL, Vaarala O, Vahteristo P & Aaltonen LA. Whole genome sequencing identifies STAT4 as a putative susceptibility gene in classic Kaposi sarcoma. *Journal of Infectious Diseases* 211, 1842-1851, 2015.

251. Ristolainen H, Kilpivaara O, Kamper P, Taskinen M, Saarinen S, Leppä S, d'Amore F, Aaltonen LA. Identification of homozygous deletion in ACAN and other candidate variants in familial classical

Hodgkin lymphoma by exome sequencing. *British Journal of Haematology* 170, 428-431, 2015.

252. Donner I, Kiviluoto T, Ristimäki A, Aaltonen LA, & Vahteristo P. Exome sequencing reveals three novel candidate predisposition genes for diffuse gastric cancer. *Familial Cancer* 14, 241-246, 2015,

253. Tanskanen T, Gylfe AE, Katainen R, Taipale M, Renkonen-Sinisalo L, Järvinen H, Mecklin JP, Böhm J, Kilpivaara O, Pitkänen E, Palin K, Vahteristo P, Tuupanen S, Aaltonen LA. Systematic search for rare variants in Finnish early-onset colorectal cancer patients. *Cancer Genetics* 208, 35-40, 2015.

254. Kondelin J, Tuupanen S, GylfeAE, Aavikko M, Renkonen-Sinisalo L, Järvinen H, Böhm J, Mecklin J-P, Andersen CL, Vahteristo P, Pitkänen E & Aaltonen LA. 3'-UTR Poly(T/U) Repeat of *EWSR1* is Altered in Microsatellite Unstable Colorectal Cancer with Nearly Perfect Sensitivity. *Familial Cancer* 14, 449-453, 2015.

255. Välimäki N, Demir H, Pitkänen E, Kaasinen E, Karppinen A, Kivipelto L, Schalin-Jäntti C, Aaltonen LA & Karhu A. Whole-Genome Sequencing of Growth Hormone (GH)-Secreting Pituitary Adenomas. *Journal of Clinical Endocrinology and Metabolism* 100, 3918-3927, 2016.

256. Katainen R, Dave K, Pitkänen E, Palin K, Kivioja T, Välimäki N, Gylfe A, Ristolainen H, Hänninen UA, Cajuso T, Kondelin J, Tanskanen T, Mecklin J-P, Järvinen H, Renkonen-Sinisalo L, Lepistö A, Kaasinen E, Kilpivaara O, Tuupanen S, Enge M, Taipale J & Aaltonen LA. CTCF/cohesin binding sites are frequently mutated in cancer. *Nature Genetics* 47, 818-821, 2015.

257. Mehine M, Heinonen H-R, Sarvilinna N, Pitkänen E, Mäkinen N, Katainen R, Tuupanen S, BützowR, Sjöberg J & Aaltonen LA. Clonally related uterine leiomyomas are common and display branched tumor evolution. *Human Molecular Genetics* 24, 4407-4416, 2015.

258. Mehine M, Kaasinen E, Heinonen H-R, Mäkinen N, Kämpjärvi K, Sarvilinna N, Aavikko M, Vähärautio A, Pasanen A, Bützow R, Heikinheimo O, Sjöberg J, Pitkänen E, Vahteristo P & Aaltonen LA. Integrated data analysis reveals uterine leiomyoma subtypes with distinct driver pathways and biomarkers. *Proceedings of the National Academy of Sciences* 113, 1315-1320, 2016.

259. Orlando G, Law PJ, Palin K, Tuupanen S, Gylfe A, Hänninen U, Cajuso T, Tanskanen T, Kondelin J, Kaasinen E, Sarin AP, Kaprio J, Eriksson JG, Rissanen H, Knekt P, Pukkala E, Jousilahti P, Salomaa V, Ripatti S, Palotie A, Järvinen H, Renkonen-Sinisalo L, Lepistö A, Böhm J, Meklin JP, Al-Tassan NA, Palle C, Martin L, Barclay E, Tenesa A, Farrington S, Timofeeva MN, Meyer BF, Wakil SM, Campbell H, Smith CG, Idziaszczyk S, Maughan TS, Kaplan R, Kerr R, Kerr D, Buchanan DD, Win AK, Hopper J, Jenkins M, Lindor NM, Newcomb PA, Gallinger S, Conti D, Schumacher F, Casey G, Taipale J, Cheadle JP, Dunlop MG, Tomlinson IP, Aaltonen LA & Houlston RS. Variation at 2q35 (*PNKD* and *TMBIM1*) influences colorectal cancer risk and identifies a pleiotropic effect with inflammatory bowel disease. *Human Molecular Genetics* 25, 2349-2359, 2016.

260. Kämpjärvi K, Mäkinen N, Mehine M, Välipakka S, Uimari O, Pitkänen E, Heinonen H-R, Heikkinen T, Tolvanen J, Ahtikoski A, Frizzell N, Sarvilinna N, Sjöberg J, Bützow R, Aaltonen LA & Vahteristo P. *MED12* mutations and *FH* inactivation are mutually exclusive in uterine leiomyomas. *British Journal of Cancer* 114, 1405-1411, 2016.

261. Jarvis D, Mitchell JS, Law PJ, Palin K, Tuupanen S, Gylfe A, Hänninen UA, Cajuso T, Tanskanen T, Kondelin J, Kaasinen E, Sarin AP, Kaprio J, Eriksson JG, Rissanen H, Knekt P, Pukkala E, Jousilahti P, Salomaa V, Ripatti S, Palotie A, Järvinen H, Renkonen-Sinisalo L, Lepistö A, Böhm J, Meklin JP, Al-Tassan NA, Palles C, Martin L, Barclay E, Farrington SM, Timofeeva MN, Meyer BF, Wakil SM, Campbell H, Smith CG, Idziaszczyk S, Maughan TS, Kaplan R, Kerr R, Kerr D, Buchanan DD, Win AK, Hopper JL, Jenkins MA, Lindor NM, Newcomb PA, Gallinger S, Conti D, Schumacher F, Casey G, Taipale J, Aaltonen LA, Cheadle JP, Dunlop MG, Tomlinson IP & Houlston RS. Mendelian randomization analysis strongly implicates adiposity with risk of developing colorectal cancer. *British Journal of Cancer* 115, 266-272, 2016.

262. Donner I, Katainen R, Tanskanen T, Kaasinen E, Aavikko M, Ovaska K, Artama M, Pukkala E, Aaltonen LA. Candidate susceptibility variants for esophageal squamous cell carcinoma. *Genes, Chromosomes and Cancer* in press.

REVIEWS

1. Aaltonen LA & Peltomäki P. Genes involved in hereditary nonpolyposis colorectal cancer. *Anticancer Research* 14, 1657-1660, 1994.

2. Mecklin J-P, Peltomäki P, Aaltonen LA & Kääriäinen H. Periytyvä syöpä. *Suomen Lääkärilehti* 18-19, 2057-2064, 1995.

3. Wolf M, Aaltonen LA, Szymanska J, Tarkkanen M, Elomaa I & Knuutila S. Microsatellite markers as tools for characterization of DNA amplifications evaluated by comparative genomic hybridization. *Cancer Genetics and Cytogenetics* 93, 33-38, 1997.

4. Aaltonen LA. Molecular epidemiology of hereditary nonpolyposis colorectal cancer in Finland. *Recent Results in Cancer Research* 1998, 154:306-311, 1998.

5. Aaltonen LA. Hereditary intestinal cancer. *Seminars in Cancer Biology* 10, 289-298, 2000.

6. Shibata D & Aaltonen LA. Genetic predisposition and somatic diversification in tumor development and progression, *Advances in Cancer Research* 80:83-114, 2001.

7. Launonen V & Aaltonen LA. Periytyvän kolorektaalisyövän geenimuutokset. *Suomen Lääkärilehti* 33, 3125 – 3130, 2002.

8. Eng C, Kiuru M, Fernandez MJ & Aaltonen LA. Role for mitochondrial enzymes in inherited neoplasia and beyond. *Nature Reviews Cancer* 3, 193-202, 2003.

9. Aaltonen LA. Periytyvän kasvainttiuden molekyyliasta (Äyräpään luento 2006). *Duodecim* 122, 2221-30, 2006.

10. Karhu A & Aaltonen LA. Susceptibility to pituitary neoplasia related to MEN-1, CDKN1B and AIP mutations; an update. *Human Molecular Genetics* 16 (R1), R73-R79, 2007.

11. Tomlinson IP, Dunlop M, Campbell H, Zanke B, Gallinger S, Hudson T, Koessler T, Pharoah PD, Niittymäki I, Tuupanen S, Aaltonen LA, Hemminki K, Lindblom A, Försti A, Sieber O, Lipton L, van Wezel T, Morreau H, Wijnen JT, Devilee P, Matsuda K, Nakamura Y, Castellví-Bel S, Ruiz-Ponte C, Castells A, Carracedo A, Ho JW, Sham P, Hofstra RM, Vodicka P, Brenner H, Hampe J, Schafmayer C, Tepel J, Schreiber S, Völzke H, Lerch MM, Schmidt CA, Buch S, Moreno V, Villanueva CM, Peterlongo P, Radice P, Echeverry MM, Velez A, Carvajal-Carmona L, Scott R, Penegar S, Broderick P, Tenesa A, Houlston RS. COGENT (COlorectal cancer GENeTics): an international consortium to study the role of polymorphic variation on the risk of colorectal cancer. *British Journal of Cancer* 102, 447-454, 2010.
12. Beckers A, Aaltonen LA, Daly A & Karhu A. Familial Isolated Pituitary Adenomas (FIPA) and the Pituitary Adenoma Predisposition due to Mutations in the Aryl Hydrocarbon Receptor Interacting Protein (AIP) gene (Review). *Endocrine Reviews* 34, 239-277, 2013.
13. Kilpivaara O & Aaltonen LA. Diagnostic Cancer Genome Sequencing and the Contribution of Germline Variants. *Science* 339, 1559-1562, 2013.
14. Sur I, Tuupanen S, Whittington T, Aaltonen LA & Taipale J. Lessons from functional analysis of genome-wide association studies. *Cancer Research* 73, 4180-4184, 2013.
15. Mehine M, Mäkinen N, Heinonen H-R, Aaltonen LA & Vahteristo P. Genomics of uterine leiomyomas: insights from high-throughput sequencing. *Fertility and Sterility* 102, 621-629, 2014.