

## ColonScreen<sup>®</sup>: Riferimenti Bibliografici

1. National Cancer Institute. Cancer Stat Fact Sheets. Accessed October 22, 2014; Available from: <http://seer.cancer.gov/>.
2. National Cancer Institute. Accessed October 22, 2014; Available from: <http://www.cancer.gov/>.
3. Hampel, H., Genetic testing for hereditary colorectal cancer. *Surg Oncol Clin N Am*, 2009. 18(4): p. 687-703.
4. Lipton, L. and I. Tomlinson, The genetics of FAP and FAP-like syndromes. *Fam Cancer*, 2006. 5(3): p. 221-6.
5. Petersen, G.M., J. Slack, and Y. Nakamura, Screening guidelines and premorbid diagnosis of familial adenomatous polyposis using linkage. *Gastroenterology*, 1991. 100(6): p. 1658-64.
6. Pedace, L., et al., Identification of a novel duplication in the APC gene using multiple ligation probe amplification in a patient with familial adenomatous polyposis. *Cancer Genet Cytogenet*, 2008. 182(2): p. 130-5.
7. van Hattem, W.A., et al., Large genomic deletions of SMAD4, BMPR1A and PTEN in juvenile polyposis. *Gut*, 2008. 57(5): p. 623-7.
8. Chow, E. and F. Macrae, A review of juvenile polyposis syndrome. *J Gastroenterol Hepatol*, 2005. 20(11): p. 1634-40.
9. Gallione, C.J., et al., A combined syndrome of juvenile polyposis and hereditary haemorrhagic telangiectasia associated with mutations in MADH4 (SMAD4). *Lancet*, 2004. 363(9412): p. 852-9.
10. Bahassi, E.M., et al., The checkpoint kinases Chk1 and Chk2 regulate the functional associations between hBRCA2 and Rad51 in response to DNA damage. *Oncogene*, 2008. 27(28): p. 3977-85.
11. Cybulski, C., et al., CHEK2 is a multiorgan cancer susceptibility gene. *Am J Hum Genet*, 2004. 75(6): p. 1131-5.
12. Walsh, T., et al., Spectrum of mutations in BRCA1, BRCA2, CHEK2, and TP53 in families at high risk of breast cancer. *Jama*, 2006. 295(12): p. 1379-88.
13. Walsh, T., et al., Mutations in 12 genes for inherited ovarian, fallopian tube, and peritoneal carcinoma identified by massively parallel sequencing. *Proc Natl Acad Sci U S A*, 2011. 108(44): p. 18032-7.
14. Pharoah, P.D., et al., Incidence of gastric cancer and breast cancer in CDH1 (E-cadherin) mutation carriers from hereditary diffuse gastric cancer families. *Gastroenterology*, 2001. 121(6): p. 1348-53.
15. Guilford, P., B. Humar, and V. Blair, Hereditary diffuse gastric cancer: translation of CDH1 germline mutations into clinical practice. *Gastric Cancer*, 2010. 13(1): p. 1-10.
16. Hegde, M.R. and B.B. Roa, Genetic Testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC) *Current Protocols in Human Genetics*, 2009. 61(Unit 10.12): p. 10.12.1-10.12.28.
17. Capelle, L.G., et al., Risk and epidemiological time trends of gastric cancer in Lynch syndrome carriers in the Netherlands. *Gastroenterology*, 2010. 138(2): p. 487-92.
18. Bonadona, V., et al., Cancer risks associated with germline mutations in MLH1, MSH2, and MSH6 genes in Lynch syndrome. *JAMA*, 2011. 305(22): p. 2304-10.

19. Engel, C., et al., Risks of less common cancers in proven mutation carriers with lynch syndrome. *J Clin Oncol*, 2012. 30(35): p. 4409-15.
20. Win, A.K., et al., Colorectal and other cancer risks for carriers and noncarriers from families with a DNA mismatch repair gene mutation: a prospective cohort study. *J Clin Oncol*, 2012. 30(9): p. 958-64.
21. Jenkins, M.A., et al., Risk of colorectal cancer in monoallelic and biallelic carriers of MYH mutations: a population-based case-family study. *Cancer Epidemiol Biomarkers Prev*, 2006. 15(2): p. 312-4.
22. Win, A.K., et al., Cancer risks for monoallelic MUTYH mutation carriers with a family history of colorectal cancer. *Int J Cancer*, 2011. 129(9): p. 2256-62.
23. Vogt, S., et al., Expanded extracolonic tumor spectrum in MUTYH-associated polyposis. *Gastroenterology*, 2009. 137(6): p. 1976-85 e1-10.
24. Rennert, G., et al., MutYH mutation carriers have increased breast cancer risk. *Cancer*, 2012. 118(8): p. 1989-93.
25. Eng, C., Will the real Cowden syndrome please stand up: revised diagnostic criteria. *J Med Genet*, 2000. 37(11): p. 828-30.
26. Starink, T.M., et al., The Cowden syndrome: a clinical and genetic study in 21 patients. *Clin Genet*, 1986. 29(3): p. 222-33.
27. Heald, B., et al., Frequent gastrointestinal polyps and colorectal adenocarcinomas in a prospective series of PTEN mutation carriers. *Gastroenterology*, 2010. 139(6): p. 1927-33.
28. Tan, M.H., et al., Lifetime cancer risks in individuals with germline PTEN mutations. *Clin Cancer Res*, 2012. 18(2): p. 400-7.
29. Mester, J.L., et al., Papillary renal cell carcinoma is associated with PTEN hamartoma tumor syndrome. *Urology*, 2012. 79(5): p. 1187 e1-7.
30. Hearle, N., et al., Frequency and spectrum of cancers in the Peutz-Jeghers syndrome. *Clin Cancer Res*, 2006. 12(10): p. 3209-15.
31. Lim, W., et al., Relative frequency and morphology of cancers in STK11 mutation carriers. *Gastroenterology*, 2004. 126(7): p. 1788-1794.
32. Hwang, S.J., et al., Germline p53 mutations in a cohort with childhood sarcoma: sex differences in cancer risk. *Am J Hum Genet*, 2003. 72(4): p. 975-83.
33. Birch, J.M., et al., Prevalence and diversity of constitutional mutations in the p53 gene among 21 Li-Fraumeni families. *Cancer Res*, 1994. 54(5): p. 1298-304.
34. Olivier, M., et al., Li-Fraumeni and related syndromes: correlation between tumor type, family structure, and TP53 genotype. *Cancer Res*, 2003. 63(20): p. 6643-50.
35. Gonzalez, K.D., et al., Beyond Li Fraumeni Syndrome: clinical characteristics of families with p53 germline mutations. *J Clin Oncol*, 2009. 27(8): p. 1250-6.
36. McCuaig, J.M., et al., Routine TP53 testing for breast cancer under age 30: ready for prime time? *Fam Cancer*, 2012. 11(4): p. 607-13.