Riferimenti Bibliografici

1. Institute, N.C. Cancer Stat Fact Sheets October 22, 2014]; Available from: http://seer.cancer.gov/.

2. Institute, N.C. October 22, 2014]; Available from: http://www.cancer.gov/.

3. Castera, L., et al., Next-generation sequencing for the diagnosis of hereditary breast and ovarian cancer using genomic capture targeting multiple candidate genes. Eur J Hum Genet, 2014. 22(11): p. 1305-13.

4. Walsh, T., et al., Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. Proc Natl Acad Sci U S A, 2010. 107(28): p. 12629-33.

5. van der Groep, P., E. van der Wall, and P.J. van Diest, Pathology of hereditary breast cancer. Cell Oncol (Dordr), 2011. 34(2): p. 71-88.

6. Walsh, T. and M.C. King, Ten genes for inherited breast cancer. Cancer Cell, 2007. 11(2): p. 103-5.

7. Meindl, A., et al., Hereditary breast and ovarian cancer: new genes, new treatments, new concepts. Dtsch Arztebl Int, 2011. 108(19): p. 323-30.

8. Antoniou, A., et al., Average risks of breast and ovarian cancer associated with BRCA1 or BRCA2 mutations detected in case Series unselected for family history: a combined analysis of 22 studies. Am J Hum Genet, 2003. 72(5): p. 1117-30.

9. Chen, S. and G. Parmigiani, Meta-analysis of BRCA1 and BRCA2 penetrance. J Clin Oncol, 2007. 25(11): p. 1329-33.

10. Ford, D., et al., Genetic heterogeneity and penetrance analysis of the BRCA1 and BRCA2 genes in breast cancer families. The Breast Cancer Linkage Consortium. Am J Hum Genet, 1998. 62(3): p. 676-89.

11. Loveday, C., et al., Germline RAD51C mutations confer susceptibility to ovarian cancer. Nat Genet, 2012. 44(5): p. 475-6; author reply 476.

12. 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.

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. Pennington, K.P., et al., Germline and somatic mutations in homologous recombination genes predict platinum response and survival in ovarian, fallopian tube, and peritoneal carcinomas. Clin Cancer Res, 2014. 20(3): p. 764-75.

15. Pennington, K.P. and E.M. Swisher, Hereditary ovarian cancer: beyond the usual suspects. Gynecol Oncol, 2012. 124(2): p. 347-53.

16. Loveday, C., et al., Germline mutations in RAD51D confer susceptibility to ovarian cancer. Nat Genet, 2011. 43(9): p. 879-82.

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

17. Hampel, H., Genetic testing for hereditary colorectal cancer. Surg Oncol Clin N Am, 2009. 18(4): p. 687-703.

18. Shi, C., R.H. Hruban, and A.P. Klein, Familial pancreatic cancer. Arch Pathol Lab Med, 2009. 133(3): p. 365-74.

19. Rosner, I., et al., The clinical implications of the genetics of renal cell carcinoma. Urol Oncol, 2009. 27(2): p. 131-6.

20. Chan-Smutko, G., Genetic testing by cancer site: urinary tract. Cancer J, 2012. 18(4): p. 343-9.

21. Coleman, J.A. and P. Russo, Hereditary and familial kidney cancer. Curr Opin Urol, 2009. 19(5): p. 478-85.

22. Rini, B.I., S.C. Campbell, and W.K. Rathmell, Renal cell carcinoma. Curr Opin Oncol, 2006. 18(3): p. 289-96.

23. Fishbein, L. and K.L. Nathanson, Pheochromocytoma and paraganglioma: understanding the complexities of the genetic background. Cancer Genet, 2012. 205(1-2): p. 1-11.

24. Welander, J., P. Soderkvist, and O. Gimm, Genetics and clinical characteristics of hereditary pheochromocytomas and paragangliomas. Endocr Relat Cancer, 2011. 18(6): p. R253-76.

25. DeLellis, R.A., Pathology and genetics of tumours of endocrine organs. World Health Organization classification of tumours. 2004, Lyon, France: IARC Press.

26. Fishbein, L., et al., Inherited mutations in pheochromocytoma and paraganglioma: why all patients should be offered genetic testing. Ann Surg Oncol, 2013. 20(5): p. 1444-50.

27. Mannelli, M., et al., Clinically guided genetic screening in a large cohort of italian patients with pheochromocytomas and/or functional or nonfunctional paragangliomas. J Clin Endocrinol Metab, 2009. 94(5): p. 1541-7.

28. Mannelli, M., et al., Subclinical phaeochromocytoma. Best Pract Res Clin Endocrinol Metab, 2012. 26(4): p. 507-15.

29. Lipton, L. and I. Tomlinson, The genetics of FAP and FAP-like syndromes. Fam Cancer, 2006. 5(3): p. 221-6.

30. 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.

31. 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.

32. Renwick, A., et al., ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. Nat Genet, 2006. 38(8): p. 873-5.

33. Roberts NJ, J.Y., Yu J, Kopelovich L, Petersen GM, Bondy ML, Steven Gallinger, Schwartz AG, Syngal S, Cote ML, Axilbund J, Schulick R, Ali SZ, Eshleman JR, Velculescu VE, Goggins M, Bert Vogelstein, Papadopoulos M, Hruban RH, Kinzler KW, Klein AP, ATM Mutations in Patients with hereditary Pancreatic cancer. Cancer Discovery, 2011. 2(1): p. OF1-OF6.

34. Janavicius, R., Founder BRCA1/2 mutations in the Europe: implications for hereditary breast-ovarian cancer prevention and control. EPMA J, 2010. 1(3): p. 397-412.

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CHIAMATA GRATUITA
NUMEROVERDE
800-501651

35. Ferla, R., et al., Founder mutations in BRCA1 and BRCA2 genes. Ann Oncol, 2007. 18 Suppl 6: p. vi93-8.

36. Tulinius, H., et al., The effect of a single BRCA2 mutation on cancer in Iceland. J Med Genet, 2002. 39(7): p. 457-62.

37. Tai, Y.C., et al., Breast cancer risk among male BRCA1 and BRCA2 mutation carriers. J Natl Cancer Inst, 2007. 99(23): p. 1811-4.

38. Thompson, D., D.F. Easton, and C. Breast Cancer Linkage, Cancer Incidence in BRCA1 mutation carriers. J Natl Cancer Inst, 2002. 94(18): p. 1358-65.

39. Folkins, A.K. and T.A. Longacre, Hereditary gynaecological malignancies: advances in screening and treatment. Histopathology, 2013. 62(1): p. 2-30.

40. Shannon, K.M. and A. Chittenden, Genetic testing by cancer site: breast. Cancer J, 2012. 18(4): p. 310-9.

41. Kote-Jarai, Z., et al., BRCA2 is a moderate penetrance gene contributing to young-onset prostate cancer: implications for genetic testing in prostate cancer patients. Br J Cancer, 2011. 105(8): p. 1230-4.

42. van Asperen, C.J., et al., Cancer risks in BRCA2 families: estimates for sites other than breast and ovary. J Med Genet, 2005. 42(9): p. 711-9.

43. Damiola, F., et al., Rare key functional domain missense substitutions in MRE11A, RAD50, and NBN contribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. Breast Cancer Res, 2014. 16(3): p. R58.

44. Seal, S., et al., Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. Nat Genet, 2006. 38(11): p. 1239-41.

45. Meindl, A., et al., Germline mutations in breast and ovarian cancer pedigrees establish RAD51C as a human cancer susceptibility gene. Nat Genet, 2010. 42(5): p. 410-4.

46. van Hattem, W.A., et al., Large genomic deletions of SMAD4, BMPR1A and PTEN in juvenile polyposis. Gut, 2008. 57(5): p. 623-7.

47. Chow, E. and F. Macrae, A review of juvenile polyposis syndrome. J Gastroenterol Hepatol, 2005. 20(11): p. 1634-40.

48. 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.

49. 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.

50. Cybulski, C., et al., CHEK2 is a multiorgan cancer susceptibility gene. Am J Hum Genet, 2004. 75(6): p. 1131-5.

51. 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.

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CHIAMATA GRATUITA
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800-501651

52. 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.

53. 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.

54. Goldstein, A.M., et al., High-risk melanoma susceptibility genes and pancreatic cancer, neural system tumors, and uveal melanoma across GenoMEL. Cancer Res, 2006. 66(20): p. 9818-28.

55. Puntervoll, H.E., et al., Melanoma prone families with CDK4 germline mutation: phenotypic profile and associations with MC1R variants. J Med Genet, 2013. 50(4): p. 264-70.

56. Begg, C.B., et al., Lifetime risk of melanoma in CDKN2A mutation carriers in a population-based sample. J Natl Cancer Inst, 2005. 97(20): p. 1507-15.

57. Bishop, D.T., et al., Geographical variation in the penetrance of CDKN2A mutations for melanoma. J Natl Cancer Inst, 2002. 94(12): p. 894-903.

58. Cust, A.E., et al., Melanoma risk for CDKN2A mutation carriers who are relatives of population-based case carriers in Australia and the UK. J Med Genet, 2011. 48(4): p. 266-72.

59. Vasen, H.F., et al., Risk of developing pancreatic cancer in families with familial atypical multiple mole melanoma associated with a specific 19 deletion of p16 (p16-Leiden). Int J Cancer, 2000. 87(6): p. 809-11.

60. McWilliams, R.R., et al., Prevalence of CDKN2A mutations in pancreatic cancer patients: implications for genetic counseling. Eur J Hum Genet, 2011. 19(4): p. 472-8.

61. de Snoo, F.A., et al., Increased risk of cancer other than melanoma in CDKN2A founder mutation (p16-Leiden)-positive melanoma families. Clin Cancer Res, 2008. 14(21): p. 7151-7.

62. Laud, K., et al., Comprehensive analysis of CDKN2A (p16INK4A/p14ARF) and CDKN2B genes in 53 melanoma index cases considered to be at heightened risk of melanoma. J Med Genet, 2006. 43(1): p. 39-47.

63. Binni, F., et al., Novel and recurrent p14 mutations in Italian familial melanoma. Clin Genet, 2010. 77(6): p. 581-6.

64. 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.

65. 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.

66. 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.

67. 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.

68. 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.

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E-mail: info@genomamilano.it

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800-501651

69. 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.

70. 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.

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

72. Rennert, G., et al., MutYH mutation carriers have increased breast cancer risk. Cancer, 2012. 118(8): p. 1989-93.

73. Slater, E.P., et al., PALB2 mutations in European familial pancreatic cancer families. Clin Genet, 2010. 78(5): p. 490-4.

74. Casadei, S., et al., Contribution of inherited mutations in the BRCA2-interacting protein PALB2 to familial breast cancer. Cancer Res, 2011. 71(6): p. 2222-9.

75. Antoniou, A.C., et al., Breast-cancer risk in families with mutations in PALB2. N Engl J Med, 2014. 371(6): p. 497-506.

76. Tischkowitz, M.D., et al., Analysis of the gene coding for the BRCA2-interacting protein PALB2 in familial and sporadic pancreatic cancer. Gastroenterology, 2009. 137(3): p. 1183-6.

77. Jones, S., et al., Exomic sequencing identifies PALB2 as a pancreatic cancer susceptibility gene. Science, 2009. 324(5924): p. 217.

78. Eng, C., Will the real Cowden syndrome please stand up: revised diagnostic criteria. J Med Genet, 2000. 37(11): p. 828-30.

79. Starink, T.M., et al., The Cowden syndrome: a clinical and genetic study in 21 patients. Clin Genet, 1986. 29(3): p. 222-33.

80. 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.

81. Tan, M.H., et al., Lifetime cancer risks in individuals with germline PTEN mutations. Clin Cancer Res, 2012. 18(2): p. 400-7.

82. Mester, J.L., et al., Papillary renal cell carcinoma is associated with PTEN hamartoma tumor syndrome. Urology, 2012. 79(5): p. 1187 e1-7.

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

84. Lim, W., et al., Relative frequency and morphology of cancers in STK11 mutation carriers. Gastroenterology, 2004. 126(7): p. 1788-1794.

85. 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.

86. 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.

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87. 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.

88. 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.

89. Gardie, B., et al., Novel FH mutations in families with hereditary leiomyomatosis and renal cell cancer (HLRCC) and patients with isolated type 2 papillary renal cell carcinoma. J Med Genet, 2011. 48(4): p. 226-34.

90. Barrisford, G.W., et al., Familial renal cancer: molecular genetics and surgical management. Int J Surg Oncol, 2011. 2011: p. 658767.

91. Baba, M., et al., Folliculin encoded by the BHD gene interacts with a binding protein, FNIP1, and AMPK, and is involved in AMPK and mTOR signaling. Proc Natl Acad Sci U S A, 2006. 103(42): p. 15552-7.

92. Pavlovich, C.P., et al., Evaluation and management of renal tumors in the Birt-Hogg-Dube syndrome. J Urol, 2005. 173(5): p. 1482-6.

93. Schmidt, L.S., et al., Germline BHD-mutation spectrum and phenotype analysis of a large cohort of families with Birt-Hogg-Dube syndrome. Am J Hum Genet, 2005. 76(6): p. 1023-33.

94. Lim, D.H., et al., A new locus-specific database (LSDB) for mutations in the folliculin (FLCN) gene. Hum Mutat, 2010. 31(1): p. E1043-51.

95. Zbar, B., et al., Risk of renal and colonic neoplasms and spontaneous pneumothorax in the Birt-Hogg-Dube syndrome. Cancer Epidemiol Biomarkers Prev, 2002. 11(4): p. 393-400.

96. Vocke, C.D., et al., High frequency of somatic frameshift BHD gene mutations in Birt-Hogg-Dube-associated renal tumors. J Natl Cancer Inst, 2005. 97(12): p. 931-5.

97. Comino-Mendez, I., et al., Exome sequencing identifies MAX mutations as a cause of hereditary pheochromocytoma. Nat Genet, 2011. 43(7): p. 663-7.

98. Schmidt, L., et al., Two North American families with hereditary papillary renal carcinoma and identical novel mutations in the MET proto-oncogene. Cancer Res, 1998. 58(8): p. 1719-22.

99. Bertolotto, C., et al., A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. Nature, 2011. 480(7375): p. 94-8.

100. Yokoyama, S., et al., A novel recurrent mutation in MITF predisposes to familial and sporadic melanoma. Nature, 2011. 480(7375): p. 99-103.

101. Eng, C., et al., The relationship between specific RET proto-oncogene mutations and disease phenotype in multiple endocrine neoplasia type 2. International RET mutation consortium analysis. JAMA, 1996. 276(19): p. 1575-9.

102. Carney, J.A. and C.A. Stratakis, Familial paraganglioma and gastric stromal sarcoma: a new syndrome distinct from the Carney triad. Am J Med Genet, 2002. 108(2): p. 132-9.

103. Ricketts, C., et al., Germline SDHB mutations and familial renal cell carcinoma. J Natl Cancer Inst, 2008. 100(17): p. 1260-2.

104. Vanharanta, S., et al., Early-onset renal cell carcinoma as a novel extraparaganglial component of SDHB-associated heritable paraganglioma. Am J Hum Genet, 2004. 74(1): p. 153-9.

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800-501651

105. Ricketts, C.J., et al., Tumor risks and genotype-phenotype-proteotype analysis in 358 patients with germline mutations in SDHB and SDHD. Hum Mutat, 2010. 31(1): p. 41-51.

106. Baysal, B.E., Mitochondrial complex II and genomic imprinting in inheritance of paraganglioma tumors. Biochim Biophys Acta, 2013. 1827(5): p. 573-7.

107. Hao, H.X., et al., SDH5, a gene required for flavination of succinate dehydrogenase, is mutated in paraganglioma. Science, 2009. 325(5944): p. 1139-42.

108. Kunst, H.P., et al., SDHAF2 (PGL2-SDH5) and hereditary head and neck paraganglioma. Clin Cancer Res, 2011. 17(2): p. 247-54.

109. Ni, Y., et al., Germline mutations and variants in the succinate dehydrogenase genes in Cowden and Cowden-like syndromes. Am J Hum Genet, 2008. 83(2): p. 261-8.

110. Neumann, H.P., et al., Germline mutations of the TMEM127 gene in patients with paraganglioma of head and neck and extraadrenal abdominal sites. J Clin Endocrinol Metab, 2011. 96(8): p. E1279-82.

111. Sancak, O., et al., Mutational analysis of the TSC1 and TSC2 genes in a diagnostic setting: genotype--phenotype correlations and comparison of diagnostic DNA techniques in Tuberous Sclerosis Complex. Eur J Hum Genet, 2005. 13(6): p. 731-41.

112. Borkowska, J., et al., Tuberous sclerosis complex: tumors and tumorigenesis. Int J Dermatol, 2011. 50(1): p. 13-20.

113. Hoogeveen-Westerveld, M., et al., Functional assessment of TSC1 missense variants identified in individuals with tuberous sclerosis complex. Hum Mutat, 2012. 33(3): p. 476-9.

114. Rodrigues, D.A., C.M. Gomes, and I.M. Costa, Tuberous sclerosis complex. An Bras Dermatol, 2012. 87(2): p. 184-96.

115. Sasongko, T.H., et al., Novel mutations in 21 patients with tuberous sclerosis complex and variation of tandem splice-acceptor sites in TSC1 exon 14. Kobe J Med Sci, 2008. 54(1): p. E73-81.

116. Lonser, R.R., et al., von Hippel-Lindau disease. Lancet, 2003. 361(9374): p. 2059-67.

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