

Curriculum Vitae Europass



Informazioni personali

Nome(i) / Cognome(i)	Maurizio Genuardi
Indirizzo(i)	Sezione di Medicina Genomica, Dipartimento di Scienze della Vita e di Sanità Pubblica, Università Cattolica del Sacro Cuore, Largo Francesco Vito 1, 00168 ROMA
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Cittadinanza	Italiana
Data di nascita	22-09-1957
Sesso	M
Codice fiscale	GNRMRZ57P22G273G

Esperienza professionale

Date	2014-presente
Posizione	Professore Ordinario di Genetica Medica
Principali attività e responsabilità	Responsabile Sezione di Medicina Genomica, Dipartimento di Scienze della Vita e di Sanità Pubblica, Direttore dell'Istituto di Medicina Genomica (già Istituto di Medina Genomica fino a novembre 2019) - Responsabile UOC Genetica Medica
Ente	Università Cattolica del Sacro Cuore – Fondazione Policlinico “Universitario A. Gemelli” IRCCS
Date	2001-2014
Posizione	Professore Ordinario di Genetica Medica
Principali attività e responsabilità	Responsabile SOD Complessa Genetica Medica, AOU Careggi; Responsabile UOC Genetica Medica AOU Meyer (2005-2012)
Ente	Università degli Studi di Firenze
Date	1988-2001
Posizione	Ricercatore Universitario
Principali attività e responsabilità	Assistente e dal 1994 Aiuto Servizio di Genetica Medica
Ente	Università Cattolica del Sacro Cuore – Policlinico Universitario A. Gemelli, Roma
Date	1986-1988
Posizione	Project Investigator

Ente Dept. Biochemistry and Molecular Biology, MD Anderson Cancer Center, Houston, Texas

Date 1985-1986

Posizione Borsista

Ente Istituto IRCCS OASI Maria SS, Troina (EN)

Date 1984

Posizione Medico Interno Universitario con Compiti Assistenziali

Ente Servizio di Citogenetica Clinica, Policlinico "A. Gemelli" – Università Cattolica del Sacro Cuore, Roma

Visite di Studio

Date 1994

Posizione Visiting Assistant Professor

Ente Genetics Laboratory, Vermont Cancer Center, University of Vermont, Burlington, USA

Date 2001

Posizione Visiting Assistant Professor and Clinical Scientist

Ente Victorian Clinical Genetics Services/Murdoch Children's Research Institute, Melbourne, Australia

2007

Visiting Professor and Clinical Scientist; Honorary Consultant

Victorian Clinical Genetics Services/Murdoch Children's Research Institute and Royal Melbourne Hospital, Melbourne, Australia

Istruzione

Date 1984

Titolo della qualifica rilasciata Specializzazione in Ematologia Generale (Clinica e Laboratorio)

Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione Università Cattolica del Sacro Cuore, Roma

Date 1981

Titolo della qualifica rilasciata Laurea in Medicina e Chirurgia

Nome e tipo d'organizzazione erogatrice dell'istruzione e formazione Università Cattolica del Sacro Cuore, Roma

Capacità e competenze personali

Madrelingua(e) Italiano

Altra(e) lingua(e) Inglese, francese

Incarichi accademici 2001-2007 e 2010-2014: Direttore della Scuola di Specializzazione in Genetica Medica, Università degli Studi di Firenze

2015-presente: Direttore della Scuola di Specializzazione in Genetica Medica, Università Cattolica del Sacro Cuore, Roma

2004-2007 e 2012-2015: Referente Nazionale presso il C.U.N. per le Scuole di Specializzazione italiane in Genetica Medica

Partecipazione a e incarichi in Società Scientifiche Membro delle seguenti società scientifiche: Società Italiana di Genetica Umana; European Society of Human Genetics; American Society of Human Genetics; International Society for the Study of Gastrointestinal Hereditary Tumors (InSiGHT); Associazione Italiana per lo Studio della Familiarità ed Ereditarietà dei Tumori Gastrointestinali (AIFEG-ONLUS); Human Genome Variation Society; European Society of Medical Oncology (ESMO).

Presidente AIFEG: 2004-2007

Coordinatore dello Italian Country Node, Human Variome Project, 2013-presente

Membro del Variant Interpretation Committee (VIC), InSiGHT (2007-presente; coordinatore 2007-2018)

Membro del VIC Governance Committee, InSiGHT (2014-presente)

Coordinatore, gruppo di Lavoro di Genetica Oncologica, SIGU (2012-2015)

Membro, Scientific Program Planning Committee, European Society of Human Genetics (2012-2016 e 2018)

Membro eletto del Council InSiGHT per il quadriennio 2013-2017; Chairman per il biennio 2015-2017; Past Chairman, 2018-2019

Presidente, Società Italiana di Genetica Umana (SIGU), 2017-2019

Membro dell'Executive Board della European Society of Human Genetics per il triennio 2019-2022

Presidente Eletto, European Society of Human Genetics, 2019-20

Presidente, European Society of Human Genetics, in carica per l'anno 2020-2021

Attività editoriale e di revisore e altri riconoscimenti

Revisore per diverse riviste scientifiche, tra cui: American Journal of Medical Genetics, Journal of Medical Genetics, European Journal of Human Genetics, Human Genetics, Human Mutation, Lancet Oncology, International Journal of Cancer, British Journal of Cancer, Genes Chromosomes and Cancer, Melanoma Research, Neuroscience Letters, Human Reproduction, Oncogene, Clinical Genetics, Cancer Letters, Human Molecular Genetics, Gastroenterology, Digestive and Liver Disease, GeneReviews

Revisore di progetti scientifici per: MIUR, Dutch Cancer Society, Ateneo Italo-Tedesco, Scottish Executive, Institut National du Cancer (Francia), Swiss National Science Foundation

Membro del comitato di valutazione programma SIR (Scientific Independence of Young Investigators), MIUR 2014-2015

Guest Editor di un fascicolo monografico della rivista Seminars in Medical Genetics (American Journal of Medical Genetics Part C), Inherited Cancer Predisposition, Agosto 2004

Associate Editor, BMC Medical Genetics (2011-2015)

Communicating Editor, Human Mutation (2013-presente)

Section Editor (Cancer Genetics), European Journal of Human Genetics (2014-presente)

Membro dello IARC (International Agency for Research on Cancer) Working Group on Unclassified Sequence Variants in Cancer Predisposition Genes (Lione, 2008)

Coordinatore dello IARC Working Group on Unclassified Sequence Variants in Mismatch Repair Genes (Lione, 2009)

Membro del gruppo di lavoro "Medicina Personalizzata" – Fondazione Smith Kline (2008)

Membro del Comitato Etico Locale dell'Azienda Ospedaliero-Universitaria Careggi (2004-2012)

Membro del Comitato Etico del Policlinico "A. Gemelli", Roma, e successivamente della Fondazione Policlinico Universitario A. Gemelli e della Fondazione Policlinico Universitario A. Gemelli IRCCS (2013-)

Membro del Comitato Etico IRCCS Sicilia – Sezione Centro Neurolesi "Bonino-Pulejo", Messina (2016-presente)

Membro del Comitato Etico del Policlinico Universitario Campus Biomedico, Roma (2019-presente)

Membro del Comitato Nazionale per la Biosicurezza, le Biotecnologie e le Scienze della Vita presso la Presidenza del Consiglio dei Ministri per il quadriennio 2016-2020, confermato per il quadriennio 2020-2024

Relatore su invito a oltre 200 tra seminari, eventi scientifici e corsi nazionali e internazionali

Attività di ricerca

Titolare di progetti di ricerca MIUR, Ministero della Salute, AIRC, Ente Cassa di Risparmio di Firenze, Regione Toscana (Istituto Toscano Tumori), Fondazione FiorGen, LILT, altri privati
Autore di oltre 200 lavori scientifici pubblicati su riviste internazionali con Impact Factor
Autore di libro di testo per il Corso di Laurea in Medicina e Chirurgia: "Genetica Umana e Medica", Neri G., Genuardi M., Ed. Elsevier (1^a edizione 2007; 2^a edizione 2010; 3^a edizione 2014; 4^a edizione 2018)

Interessi di ricerca

Diversi ambiti della genetica clinica e molecolare, con particolare riguardo alla predisposizione ereditaria a tumori nell'uomo, in particolare tumori del colon, della mammella/ovaio, melanoma cutaneo. Classificazione clinica delle varianti di sequenza del DNA.

**Pubblicazioni su riviste
indexate**

1. Neri G., Sabatino G., Bertini E., Genuardi M. "The CFC syndrome. Report of the first two cases outside the United States". *Am. J. Med. Genet.* 27:767-771 (1987).
2. Neri G., Genuardi M., Natoli G., Costa P., Maggioni G. "A girl with G syndrome and agenesis of corpus callosum". *Am. J. Med. Genet.* 28:287-291 (1987).
3. Calvieri F., Tozzi C., Benincori A., De Merulis M.L., Bellussi A., Genuardi M., Neri G. "Partial tetrasomy 9 in an infant with clinical and radiological evidence of multiple joint dislocations". *Eur. J. Pediatr.* 147:645-648 (1988).
4. Zollino M., Genuardi M., De Santis R., Leone G., Marra R., Mancini R., Mango G., Neri G. "Partial duplication of chromosome 1q preceding the development of an L3 lymphoblastic leukemia with t(8;14) secondary to treatment for Hodgkin disease". *Eur. J. Hematol.* 40:193-197 (1988).
5. Genuardi M., Zollino M., Serra A., Leone G., Mancini R., Mango G., Neri G. "Long-term cytogenetic effects of antineoplastic treatment in relation to secondary leukemia". *Cancer Genet. Cytogenet.* 33:201-211 (1988).
6. Genuardi M., Saunders G.F. "Localization of the HLA class II-associated invariant chain gene to human chromosome band 5q32". *Immunogenetics* 28:53-56 (1988).
7. Genuardi M., Tshira H., Anderson D.E., Saunders G.F. "Distal deletion of chromosome 1p in ductal carcinoma of the breast". *Am. J. Hum. Genet.* 45:73-82 (1989).
8. Zollino M., Genuardi M., Neri G. "Differential expression of FRA16B in peripheral lymphocytes and bone marrow cells". *Cancer Genet. Cytogenet.* 49:229-233 (1990).
9. Genuardi M., Zollino M., Bellussi A., Fuhrmann W., Neri G. "Brachy/ectrodactyly and absence or hypoplasia of the fibula: an autosomal dominant condition with low penetrance and variable expressivity". *Clin. Genet.* 38:321-326 (1990).
10. Greggi S., Genuardi M., Benedetti-Panici P., Cento R., Scambia G., Neri G., Mancuso S. "Analysis of 138 consecutive ovarian cancer patients: incidence and characteristics of familial cases". *Gynecol. Oncol.* 39:300-304 (1990).
11. Zollino M., Genuardi M., Tanci P., Mango G., Rumi C., Mancini R., Neri G. "Chronic myelogenous leukemia in the course of chronic lymphocytic leukemia: evidence for an independent clonal origin". *Leukemia Res.* 15:269-273 (1991).
12. Mars W.M., Genuardi M., Tshira H., Anderson D.E., Saunders G.F. "Genomic changes on the short arm of chromosome 1 in breast cancer". *Cancer Detect. Prev.* 15:145-149 (1991).
13. Ferrandina G., Scambia G., Benedetti-Panici P., Bonanno G., De Vincenzo R., Rumi C., Bussa S., Genuardi M., Romano-Spica V., Mancuso S. "Effects of dexamethasone on the growth and EGFR expression of the OVCA433 ovarian cancer cells". *Mol. Cell. Endocrinol.* 83:183-193 (1992).
14. Genuardi M., Flamia R., Palka G., Parruti G., Neri G. "Dosage analysis at the CSF1 and CSF1R loci in a new case of partial trisomy 5q". *Clin. Genet.* 41:259-262 (1992).
15. Tanci P., Genuardi M., Santini S.A., Neri G. "PCR detection of an insertion/deletion polymorphism in intron 1 of the HRAS locus". *Nucleic Acids Res.* 20:1157 (1992).
16. Pocchiari M., Masullo C., Salvatore M., Genuardi M., Galgani S. "Creutzfeldt-Jakob disease after non-commercial dura mater graft". *Lancet* 340:614-615 (1992).
17. Genuardi M., Dionisi-Vici C., Sabetta G., Mignozzi M., Rizzoni G., Cotugno G., Martini-Neri M.E. "Brief clinical report: Cerebro-reno-digital (Meckel-like) syndrome with Dandy-Walker malformation, cystic kidneys, hepatic fibrosis, and polydactyly". *Am. J. Med. Genet.* 47:50-53 (1993).
18. Kozak L., Chiurazzi P., Genuardi M., Pomponi M.G., Zollino M., Neri G. "Mapping of a gene for non-specific X-linked mental retardation: evidence for linkage to chromosomal region Xp21.1-Xp22.3". *J. Med. Genet.* 30:866-869 (1993).
19. Pocchiari M., Salvatore M., Cutruzzola F., Genuardi M., Travaglini Allocatelli C., Masullo C., Macchi G., Alema G., Galgani S., Xi Y.G., Petraroli R., Silvestrini M.C., Brunori M.: "A new point mutation of the prion protein gene in Creutzfeldt-Jakob disease". *Ann. Neurol.* 34:802-807 (1993).
20. Genuardi M., Chiurazzi P., Capelli A., Neri G. "X-linked VACTERL with hydrocephalus: the VACTERL-H syndrome". *Birth Defects: Original Article Series* 29:235-241 (1993).
21. Genuardi M., Pomponi M.G., Sammito V., Bellussi A., Zollino M., Neri G. "Split hand/split foot anomaly in a family segregating a balanced translocation with breakpoint on 7q22.1" *Am. J. Med. Genet.* 47:823-831 (1993).
22. Masullo C., Salvatore M., Macchi G., Genuardi M., Pocchiari M. "Progressive dementia in a young patient with a homozygous deletion of the PrP gene". *Ann. N.Y. Acad. Sci.* 724:358-360 (1994).
23. Genuardi M., Gurrieri F., Neri G. "Genes for split hand/split foot and laterality defects on 7q22.1 and Xq24-q27.1". *Am. J. Med. Genet.* 50:101 (1994).
24. Orth U., Gurrieri F., Behmel A., Genuardi M., Cremer M., Gal A., Neri G. "Gene for Simpson-Golabi-Behmel syndrome is linked to HPRT in Xq26 in two European families". *Am. J. Med. Genet.* 50:388-390 (1994).
25. Genuardi M., Silvestri E., Tozzi C. "Split hand/split foot, syndactyly, urinary tract obstruction, radial, diaphragmatic, and neural tube defects: Czeizel-Losonci syndrome?". *Am. J. Med. Genet.*, 51:247-250 (1994).

26. Scherer S.W., Poorkaj P., Allen T., Kim J., Geshuri D., Nunes M., Soder S., Stephens K., Pagon R.A., Patton M.A., Berg M.A., Donlon T., Rivera H., Pfeiffer R.A., Naritomi K., Hughes H., Genuardi M., Gurrieri F., Neri G., Lovrein E., Magenis E., Tsui L.-C., Evans J.P.: "Fine mapping of the autosomal dominant split hand/split foot locus on chromosome 7, band q21.3-q22.1". *Am. J. Hum. Genet.* 55:12-20 (1994).
27. Gurrieri F., Genuardi M., Chiurazzi P., Gillissen-Kaesbach G., Neri G.: "Exclusion of linkage between autosomal dominant split hand/split foot and markers from chromosome 7q: further evidence for genetic heterogeneity". *Am. J. Hum. Genet.* 55:853-855 (1994).
28. Salvatore M., Genuardi M., Petraroli R., Masullo C., D'Alessandro M., Pocchiari M. "Polymorphisms of the prion protein gene in Italian patients with Creutzfeldt-Jakob disease". *Hum. Genet.* 94:375-379 (1994).
29. Genuardi M., Calvieri F., Tozzi C., Coslovi R., Neri G. "A new case of interstitial deletion of chromosome 3q, del(3q)(q13.12q21.3), with agenesis of the corpus callosum". *Clinical Dysmorphol.* 3:292-296 (1994).
30. Genuardi M., Neri G. "Reply to dr. Rivera: Split hand/split foot anomaly and 7q22.1". *Am. J. Med. Genet.* 53:90 (1994).
31. Genuardi M., Bardoni B., Florida G., Chiurazzi P., Scarano G., Zollino M., Garcea N., Martini-Neri M.E., Neri G. "Dicentric chromosome Y associated with Leydig cell agenesis and sex reversal". *Clin. Genet.* 47:38-41 (1995).
32. Gurrieri F., Cammarata M., Avarello R.M., Genuardi M., Pomponi M.G., Neri G., Giuffrè L. "Ulnar ray defect in an infant with a 6q21;7q31.2 translocation: further evidence for the existence of a limb defect gene in 6q21". *Am. J. Med. Genet.* 55:315-318 (1995).
33. DeBrasi D., Genuardi M., D'Agostino A., Calvieri F., Tozzi C., Varrone S., Neri G. "Double autosomal/gonosomal mosaic aneuploidy: study of nondisjunction in two cases with trisomy of chromosome 8". *Hum. Genet.* 95:519-525 (1995).
34. Cox-Froncillo M.C., Genuardi M., Bajer J., Livdi E., Adorno G., Venditti A., Masi M., Giudiceandrea P., Neri G., Papa G. "First report of t(8;21)(q22;q22) in a case of de novo acute monoblastic leukemia". *Cancer Genet. Cytogenet.* 79:82-85 (1995).
35. Zollino M., Genuardi M., Bajer J., Tornesello A., Mastrangelo S., Zampino G., Mastrangelo R., Neri G. "Constitutional trisomy 8 and myelodysplasia: report of a case and review of the literature". *Leukemia Res.* 15:269-273 (1995).
36. Neri G., Gurrieri F., Genuardi M. "Oral-facial-digital syndromes". *Am. J. Med. Genet.* 59:365-368 (1995).
37. Sorge G., Ardito S., Genuardi M., Pavone V., Rizzo R., Conti G., Neri G., Katz B.E., Opitz J.M. "Proximal femoral focal deficiency (PFFD) and fibular a/hypoplasia (FA/H): a model of a developmental field defect". *Am. J. Med. Genet.* 57:427-432 (1995).
38. Genuardi M., Scarano G., Tozzi C., Chinca M., Della Monica M., Martini-Neri M.E., Picardi P., Neri G. "Malformation syndromes with kidney dysplasia". *Birth Defects: Original Article Series* 32:377-393 (1996).
39. Gurrieri F., Prinso P., Tackels D., Kilpatrick M.W., Allanson J., Genuardi M., Vuckov A., Nanni L., Sangiorgi E., Garofalo G., Nunes M.E., Neri G., Schwartz C., Tsiouras P. "A split hand-split foot (SHFM3) gene is located at 10q24-25". *Am. J. Med. Genet.* 62:427-436 (1996).
40. Heouaine A., Mareni C., Varesco L., Genuardi M., Neri G. "Genetic counseling in hereditary non-polyposis colorectal cancer". *Tumori* 82:136-142 (1996).
41. Cama A., Genuardi M., Guanti G., Radice P., Varesco L. "Molecular genetics of hereditary non-polyposis colorectal cancer (HNPCC)". *Tumori* 82:1-13 (1996).
42. Bellacosa A., Genuardi M., Anti M., Viel A., Ponz de Leon M. "Hereditary nonpolyposis colorectal cancer: clinical, molecular genetics and counseling aspects. A review". *Am. J. Med. Genet.* 62:353-364 (1996).
43. Chiurazzi P., Destro-Bisol G., Genuardi M., Oostra B.A., Spedini G., Neri G. "Extended gene diversity at the FMR1 locus and neighbouring CA repeats in a subsaharian population". *Am. J. Med. Genet.* 64:216-219 (1996).
44. Chiurazzi P., Genuardi M., Kozak L., Giovannucci-Uzielli M.L., Bussani C., Dagna-Bricarelli F., Grasso M., Perroni L., Sebastio G., Sperandeo M.P., Oostra B.A., Neri G. "Fragile X founder chromosomes in Italy. Few initial events as possible explanation for their heterogeneity". *Am. J. Med. Genet.* 64:209-215 (1996).
45. Hohauser S., Cavallo S., Bellacosa A., Genuardi M., Galli J., Cadoni G., Almadori G., Lauriola L., Litwin S., Maurizi M., Neri G. "Telomerase activity in human laryngeal squamous cell carcinomas". *Clin. Cancer Res.* 2:1895-1900 (1996).
46. Greggi S., Genuardi M. "Familial ovarian and breast cancers". *Oncol. in Practice* 3: 17-20 (1996).
47. Viel A., Genuardi M., Capozzi E., Leonardi F., Bellacosa A., Paravatou-Petsotas M., Pomponi M.G., Fornasari M., Percesepe A., Roncucci L., Tamassia M.G., Benatti P., Ponz de Leon M., Valenti A., Covino M., Anti M., Boiocchi M., Neri G. "Characterization of *MSH2* and *MLH1* mutations in Italian families with hereditary non polyposis colorectal cancer". *Genes Chromosom. Cancer* 18:8-18 (1997).
48. Genuardi M., Gasparini P., Zelante L., Neri G. "Limb-pelvis hypoplasia/aplasia: a discrete entity in the fibuloulnar developmental field complex". *Am. J. Med. Genet.* 68:1190-194 (1997).

- 49.** Percesepe A., Benatti P., Roncucci L., Sassatelli R., Fante R., Ganazzi D., Bellacosa A., Genuardi M., Neri G., Viel A., Ponz de Leon M. "Survival analysis in families affected by hereditary non-polyposis colorectal cancer". *Int. J. Cancer* 71:373-376 (1997).
- 50.** Hohauser S., Voso M.T., Ortu-La Barbera E., Cavallo S., Bellacosa A., Rutella S., Rumi C., Genuardi M., Neri G., Leone G. "Telomerase activity in human hematopoietic progenitor cells. *Haematologica* 82:262-268 (1997).
- 51.** Greggi S., Benedetti-Panici P., Paratore M.P., Genuardi M. "Genetic counseling for families with inherited susceptibility to breast and ovarian cancer". *CME J. Gynecol. Oncol.* 2:85-89 (1997).
- 52.** EUCROMIC Quality Assessment Group "Quality guidelines and standards for genetic laboratories/clinics in prenatal diagnosis on fetal samples obtained by invasive procedures". *Eur. J. Hum. Genet.* 5:342-350 (1997).
- 53.** Guanti G., Varesco L., Crotti N., Hehouaine A., De Stefano F., Genuardi M. "Recommendations for genetic counseling of familial adenomatous polyposis". *Tumori* 83:791-794 (1997).
- 54.** Genuardi M., Viel A., Bonora D., Capozzi E., Bellacosa A., Leonardi F., Valle R., Ventura A., Pedroni M., Boiocchi M., Neri G. "Characterization of *MLH1* and *MSH2* alternative splicing and its relevance to molecular testing of colorectal cancer susceptibility". *Hum. Genet.* 102:15-20 (1998).
- 55.** Genuardi M., Anti M., Capozzi E., Leonardi F., Fornasarig M., Novella E., Bellacosa A., Valenti A., Gasbarrini G.B., Roncucci L., Benatti P., Percesepe A., Ponz de Leon M., Coco C., De Paoli A., Valentini M., Boiocchi M., Neri G., Viel A. "*MLH1* and *MSH2* constitutional mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer". *Int. J. Cancer* 75:835-839 (1998).
- 56.** Viel A., Novella E., Genuardi M., Capozzi E., Fornasarig M., Pedroni M., Santarosa M., Ponz de Leon M., Della Puppa L., Anti M., Boiocchi M. "Lack of *PMS2* gene-truncating mutations in patients with hereditary colorectal cancer". *Int. J. Oncol.* 13:565-569 (1998).
- 57.** Cattani P., Hohauser S., Bellacosa A., Genuardi M., Cavallo S., Rovella V., Almadori G., Cadoni G., Galli J., Maurizi M., Fadda G., Neri G. "Association between cyclin D1 (*CCND1*) gene amplification and human papillomavirus infection in human laryngeal squamous cell carcinoma". *Clin. Cancer Res.* 4:2585-2589 (1998).
- 58.** Viel A., Genuardi M., Lucci-Cordisco E., Capozzi E., Rovella V., Fornasarig M., Ponz de Leon M., Anti M., Pedroni M., Bellacosa A., Percesepe A., Covino M., Benatti P., Del Tin L., Roncucci L., Valentini M., Boiocchi M., Neri G. "Hereditary nonpolyposis colorectal cancer: an approach to the selection of candidates to genetic testing based on clinical and molecular characteristics". *Community Genet.* 1:229-236 (1998).
- 59.** Genuardi M., Pomponi M.G., Torrisi L., Neri G., Stagni M.L., Tozzi C. "45,X/47,XX,+18 constitutional mosaicism: clinical presentation and evidence for a somatic origin of the aneuploid cell lines". *J. Med. Genet.* 36:496-498 (1999).
- 60.** Genuardi M., Tozzi C., Pomponi M.G., Stagni M.L., Della Monica M., Scarano G., Calvieri F., Torrisi L., Neri G. "Mosaic trisomy 17 in amniocytes: phenotypic outcome, tissue distribution and uniparental disomy studies". *Eur. J. Hum. Genet.* 7:421-426 (1999).
- 61.** Ponz de Leon M., Pedroni M., Benatti P., Percesepe A., Rossi G., Genuardi M., Roncucci L. "Epidemiologic and genetic factors in colorectal cancer: development of cancer in dizygotic twins in a family with Lynch syndrome". *Ital. J. Gastroenterol. Hepatol.* 31:218-222 (1999).
- 62.** Bellacosa A., Cicchilitti L., Schepis F., Riccio A., Yeung A.T., Matsumoto Y., Golemis E.A., Genuardi M., Neri G. "MED1, a novel human methyl-CpG-binding endonuclease, interacts with DNA mismatch repair protein MLH1". *Proc. Natl. Acad. Sci. USA* 96:3969-3974 (1999).
- 63.** Ponz de Leon M., Pedroni M., Benatti P., Percesepe A., Di Gregorio C., Foroni M., Rossi G., Genuardi M., Neri G., Leonardi F., Viel A., Capozzi E., Boiocchi M., Roncucci L. "Hereditary colorectal cancer in the general population: from cancer registration to molecular diagnosis". *Gut* 45:32-38 (1999).
- 64.** Genuardi M., Carrara S., Anti M., Ponz de Leon M., Viel A. "Assessment of pathogenicity criteria for constitutional missense mutations of the hereditary nonpolyposis colorectal cancer genes *MLH1* and *MSH2*". *Eur. J. Hum. Genet.*, 7:778-782 (1999).
- 65.** Riccio A., Aaltonen L.A., Godwin A.K., Loukola A., Percesepe A., Salovaara R., Masciullo V., Genuardi M., Paravatou-Petsotas M., Bassi D.E., Ruggeri B.A., Klein-Szanto A.J.P., Testa J.R., Neri G., Bellacosa A. "The novel DNA repair gene *MED1* is mutated in human carcinomas exhibiting microsatellite instability". *Nature Genet.* 23:266-268 (1999).
- 66.** Neri G., Moreira F., Gurrieri F., Sangiorgi E., Genuardi M., Moretti-Ferreira D. "Split hand/split foot: a clinical and molecular overview". *Braz. J. Dysmorphol. Speech-Hear. Disord.* 3:5-14 (1999).
- 67.** Percesepe A., Pedroni M., Sala E., Menigatti M., Borghi F., Losi L., Viel A., Genuardi M., Benatti P., Roncucci L., Peltomäki P., Ponz de Leon M. "Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts". *Genes Chromosom. Cancer* 27:424-429 (2000).
- 68.** Ponz de Leon M., Benatti P., Pedroni M., Viel A., Genuardi M., Percesepe A., Roncucci L. "Problems in the identification of hereditary nonpolyposis colorectal cancer in two families with late development of full-blown clinical spectrum". *Am. J. Gastroenterol.* 95:2110-5 (2000).

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Autorizzo il trattamento dei miei dati personali ai sensi del Decreto Legislativo 30 giugno 2003, n. 196 "Codice in materia di protezione dei dati personali" e successive modifiche ed integrazioni.

Dichiaro che le informazioni contenute nel curriculum corrispondono a verità, e che sono consapevole delle sanzioni penali in cui posso incorrere in caso di dichiarazioni mendaci ai sensi dell'art.46 e 76 del D.P.R. 445/2000 e successive modifiche e integrazioni.

Firma



Data: 22/01/2021