Maurizio Genuardi

List of Publications by Year
in descending order

204 papers	9,296 citations	61984 43 h-index	49909 87 g-index
211 all docs	211 docs citations	211 times ranked	12435 citing authors

#	Article	IF	Citations
1	Variants of uncertain significance (VUS) in cancer predisposing genes: What are we learning from multigene panels?. European Journal of Medical Genetics, 2022, 65, 104400.	1.3	4
2	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decisionâ€making. Clinical Genetics, 2022, 101, 454-458.	2.0	3
3	The challenge of the Molecular Tumor Board empowerment in clinical oncology practice: A Position Paper on behalf of the AIOM- SIAPEC/IAP-SIBioC-SIC-SIF-SIGU-SIRM Italian Scientific Societies. Critical Reviews in Oncology/Hematology, 2022, 169, 103567.	4.4	26
4	The use of polygenic risk scores in pre-implantation genetic testing: an unproven, unethical practice. European Journal of Human Genetics, 2022, 30, 493-495.	2.8	38
5	Gastrointestinal manifestations in PTEN hamartoma tumor syndrome. Bailliere's Best Practice and Research in Clinical Gastroenterology, 2022, 58-59, 101792.	2.4	6
6	Melanocytic nevi in RASopathies: insights on dermatological diagnostic handles. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e83-e85.	2.4	6
7	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <scp>LARS1</scp> gene: Clinical, genetic, and functional characterization. Clinical Genetics, 2021, 99, 601-603.	2.0	0
8	Intrafamilial communication of hereditary breast and ovarian cancer genetic information in Italian women: towards a personalised approach. European Journal of Human Genetics, 2021, 29, 250-261.	2.8	11
9	ESHG warns against misuses of genetic tests and biobanks for discrimination purposes. European Journal of Human Genetics, 2021, 29, 894-896.	2.8	14
10	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	10.7	58
11	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. European Journal of Medical Genetics, 2021, 64, 104350.	1.3	22
12	A new founder BRCA1 haplotype identified in the Puglia region is associated with a specific age-related cancer onset in three unrelated families. Clinical Chemistry and Laboratory Medicine, 2021, 59, e95-e98.	2.3	2
13	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
14	Role of extensive diagnostic workup in young athletes and nonathletes with complex ventricular arrhythmias. Heart Rhythm, 2020, 17, 230-237.	0.7	10
15	Methylated premutation of the FMR1 gene in three sisters: correlating CGG expansion and epigenetic inactivation. European Journal of Human Genetics, 2020, 28, 567-575.	2.8	6
16	Time for Change? The Why, What and How of Promoting Innovation to Tackle Rare Diseases â€“ Is It Time to Update the EUâ€™s Orphan Regulation? And if so, What Should be Changed?. Biomedicine Hub, 2020, 5, 1-11.	1.2	11
17	Disease expression in juvenile polyposis syndrome: a retrospective survey on a cohort of 221 European patients and comparison with a literature-derived cohort of 473 SMAD4/BMPR1A pathogenic variant carriers. Genetics in Medicine, 2020, 22, 1524-1532.	2.4	44
18	Deregulated expression of the imprinted DLK1-DIO3 region in glioblastoma stemlike cells: tumor suppressor role of lncRNA MEG3. Neuro-Oncology, 2020, 22, 1771-1784.	1.2	44

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19	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
20	Cost-effectiveness analysis of genetic diagnostic strategies for Lynch syndrome in Italy. PLoS ONE, 2020, 15, e0235038.	2.5	5
21	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
22	Altered mitochondrial function in cells carrying a premutation or unmethylated full mutation of the FMR1 gene. Human Genetics, 2020, 139, 227-245.	3.8	16
23	Insights into Genetic Susceptibility to Melanoma by Gene Panel Testing: Potential Pathogenic Variants in ACD, ATM, BAP1, and POT1. Cancers, 2020, 12, 1007.	3.7	19
24	Clinical utility of genetic testing in the early diagnosis of Danon disease mimicking hypertrophic cardiomyopathy: a case report. BMC Cardiovascular Disorders, 2020, 20, 156.	1.7	0
25	Complex Muco-cutaneous Manifestations of CARMIL2-associated Combined Immunodeficiency: A Novel Presentation of Dysfunctional Epithelial Barriers. Acta Dermato-Venereologica, 2020, 100, 1-2.	1.3	9
26	Lynch syndrome with exclusive skin involvement: time to consider a molecular definition?. Familial Cancer, 2019, 18, 421-427.	1.9	0
27	BRCA1/2 Molecular Assay for Ovarian Cancer Patients: A Survey through Italian Departments of Oncology and Molecular and Genomic Diagnostic Laboratories. Diagnostics, 2019, 9, 146.	2.6	3
28	A novel nonsense PTH1R variant shows incomplete penetrance of primary failure of eruption: a case report. BMC Oral Health, 2019, 19, 249.	2.3	5
29	Recommendations for the implementation of BRCA testing in ovarian cancer patients and their relatives. Critical Reviews in Oncology/Hematology, 2019, 140, 67-72.	4.4	51
30	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
31	Rare missense variants in the ALPK1 gene may predispose to periodic fever, aphthous stomatitis, pharyngitis and adenitis (PFAPA) syndrome. European Journal of Human Genetics, 2019, 27, 1361-1368.	2.8	21
32	Old treatments for new genetic conditions: Sirolimus therapy in a child affected by mosaic overgrowth with fibroadipose hyperplasia. Clinical Genetics, 2019, 96, 102-103.	2.0	6
33	Constitutional mismatch repair deficiencyâ€“associated brain tumors: report from the European C4CMMRD consortium. Neuro-Oncology Advances, 2019, 1, vdz033.	0.7	23
34	The Current Practice of Lynch Syndrome Diagnosis and Management in Italy: A Qualitative Assessment. Public Health Genomics, 2019, 22, 189-207.	1.0	7
35	Gastrointestinal juvenile-like (inflammatory/hyperplastic) mucosal polyps in neurofibromatosis type 1 with no concurrent genetic or clinical evidence of other syndromes. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2019, 474, 259-264.	2.8	4
36	Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility. Journal of Medical Genetics, 2018, 55, 669-674.	3.2	37

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37	Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut, 2018, 67, 1306-1316.	12.1	410
38	Workload measurement for molecular genetics laboratory: A survey study. PLoS ONE, 2018, 13, e0206855.	2.5	6
39	Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987.	6.4	22
40	Classification of Genetic Variants. , 2018, , 257-280.		0
41	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. EBioMedicine, 2017, 20, 39-49.	6.1	170
42	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. Gut, 2017, 66, 1657-1664.	12.1	127
43	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	2.8	34
44	Assessment of the InSiGHT Interpretation Criteria for the Clinical Classification of 24MLH1andMSH2Gene Variants. Human Mutation, 2017, 38, 64-77.	2.5	29
45	Intersociety policy statement on the use of whole-exome sequencing in the critically ill newborn infant. Italian Journal of Pediatrics, 2017, 43, 100.	2.6	51
46	Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. Hereditary Cancer in Clinical Practice, 2017, 15, 18.	1.5	49
47	The chromosome analysis of the miscarriage tissue. Miscarried embryo/fetal crown rump length (CRL) measurement: A practical use. PLoS ONE, 2017, 12, e0178113.	2.5	4
48	The Role of Genetic Testing in the Identification of Young Athletes with Inherited Primitive Cardiac Disorders at Risk of Exercise Sudden Death. Frontiers in Cardiovascular Medicine, 2016, 3, 28.	2.4	7
49	Recommendations for the implementation of BRCA testing in the care and treatment pathways of ovarian cancer patients. Future Oncology, 2016, 12, 2071-2075.	2.4	21
50	Recurrent candidiasis and early-onset gastric cancer in a patient with a genetically defined partial MYD88 defect. Familial Cancer, 2016, 15, 289-296.	1.9	13
51	The Intestinal Polyposes: Clinical and Molecular Overview. , 2016, , 1-24.		0
52	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. Human Mutation, 2015, 36, 712-719.	2.5	39
53	Colchicine trial in PFAPA Syndrome and MEFV-negative patients. Pediatric Rheumatology, 2015, 13, .	2.1	2
54	Correlation between mutations and mRNA expression of APC and MUTYH genes: new insight into hereditary colorectal polyposis predisposition. Journal of Experimental and Clinical Cancer Research, 2015, 34, 131.	8.6	19

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55	PFAPA syndrome as an hereditary autoinflamatory disorder. Pediatric Rheumatology, 2015, 13, .	2.1	0
56	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
57	Variable expressivity of a familial 1.9ÂMb microdeletion in 3q28 leading to haploinsufficiency of TP63: Refinement of the critical region for a new microdeletion phenotype. European Journal of Medical Genetics, 2015, 58, 400-405.	1.3	4
58	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	2.9	91
59	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
60	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	1.8	43
61	Anti-miR21 oligonucleotide enhances chemosensitivity of T98G cell line to doxorubicin by inducing apoptosis. American Journal of Cancer Research, 2015, 5, 231-42.	1.4	25
62	Characterization of the rs2802292 SNP identifies FOXO3Aas a modifier locus predicting cancer risk in patients with PJS and PHTS hamartomatous polyposis syndromes. BMC Cancer, 2014, 14, 661.	2.6	11
63	MLH1constitutional and somatic methylation in patients with MLH1 negative tumors fulfilling the revised Bethesda criteria. Epigenetics, 2014, 9, 1431-1438.	2.7	22
64	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
65	MUTYH-associated polyposis (MAP): evidence for the origin of the common European mutations p.Tyr179Cys and p.Gly396Asp by founder events. European Journal of Human Genetics, 2014, 22, 923-929.	2.8	39
66	Gene variants of unknown clinical significance in Lynch syndrome. An introduction for clinicians. Familial Cancer, 2013, 12, 181-187.	1.9	27
67	The policy of public health genomics in Italy. Health Policy, 2013, 110, 214-219.	3.0	33
68	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. Gut, 2013, 62, 812-823.	12.1	630
69	Cancer risk associated with STK11/LKB1 germline mutations in Peutzâ€“Jeghers syndrome patients: Results of an Italian multicenter study. Digestive and Liver Disease, 2013, 45, 606-611.	0.9	113
70	Clinical and genetic study of a family with a paternally inherited 15q11â€“q13 duplication. American Journal of Medical Genetics, Part A, 2013, 161, 1459-1464.	1.2	17
71	Duodenal carcinoma in a 37-year-old man with Cowden/Bannayan syndrome. Digestive and Liver Disease, 2013, 45, 75-78.	0.9	9
72	The growing complexity of the intestinal polyposis syndromes. American Journal of Medical Genetics, Part A, 2013, 161, 2777-2787.	1.2	37

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73	MUTYH c.933+3A>C, associated with a severely impaired gene expression, is the first Italian founder mutation in MUTYHâ€Associated Polyposis. International Journal of Cancer, 2013, 132, 1060-1069.	5.1	16
74	Clinical utility gene card for: MUTYH-associated polyposis (MAP), Autosomal recessive colorectal adenomatous polyposis, Multiple colorectal adenomas, Multiple adenomatous polyps (MAP) - update 2012. European Journal of Human Genetics, 2013, 21, 118-118.	2.8	33
75	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
76	Encomium: Giovanni Neriâ€"Polyhedral and downâ€toâ€earth mentor. American Journal of Medical Genetics, Part A, 2013, 161, 2687-2690.	1.2	0
77	Morquio A syndrome due to Maternal Uniparental Isodisomy of the telomeric end of chromosome 16. Molecular Genetics and Metabolism, 2012, 105, 438-442.	1.1	17
78	Fabry disease: polymorphic haplotypes and a novel missense mutation in the GLA gene. Clinical Genetics, 2012, 81, 224-233.	2.0	28
79	Thymidylate synthase expression and genotype have no major impact on the clinical outcome of colorectal cancer patients treated with 5-fluorouracil. Pharmacological Research, 2011, 64, 242-248.	7.1	21
80	Constitutional FLCN mutations in patients with suspected Birt-Hogg-DubÃ© syndrome ascertained for non-cutaneous manifestations. Clinical Genetics, 2011, 79, 345-354.	2.0	36
81	High resolution melting analysis for a rapid identification of heterozygous and homozygous sequence changes in the MUTYH gene. BMC Cancer, 2011, 11, 305.	2.6	11
82	Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). Clinical Endocrinology, 2010, 72, 839-844.	2.4	29
83	Schwannomatosis associated with multiple meningiomas due to a familial SMARCB1 mutation. Neurogenetics, 2010, 11, 73-80.	1.4	90
84	A PALB2 germline mutation associated with hereditary breast cancer in Italy. Familial Cancer, 2010, 9, 181-185.	1.9	39
85	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
86	Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). European Journal of Endocrinology, 2010, 163, 329-337.	3.7	25
87	Prognostic Relevance of MLH1 and MSH2 Mutations in Hereditary Non-Polyposis Colorectal Cancer Patients. Tumori, 2009, 95, 731-738.	1.1	8
88	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H+-ATPase genes. Nephrology Dialysis Transplantation, 2009, 24, 2734-2738.	0.7	29
89	Germline mutations in MEN1 and BRCA1 genes in a woman with familial multiple endocrine neoplasia type 1 and inherited breastâ€"ovarian cancer syndromes: a case report. Cancer Genetics and Cytogenetics, 2009, 195, 75-79.	1.0	14
90	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. Pediatric Nephrology, 2009, 24, 2147-2153.	1.7	32

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91	Founder mutations account for the majority of BRCA1-attributable hereditary breast/ovarian cancer cases in a population from Tuscany, Central Italy. Breast Cancer Research and Treatment, 2009, 117, 497-504.	2.5	31
92	Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome. European Journal of Human Genetics, 2009, 17, 919-927.	2.8	42
93	Genetic profiling of Bolivian population using 15 STR markers of forensic importance. Legal Medicine, 2009, 11, 149-151.	1.3	2
94	Malignant extra-adrenal pheochromocytoma caused by an SDHB intronic variation leading to a 54-bp deletion in exon 4. Journal of Endocrinological Investigation, 2009, 32, 111-114.	3.3	1
95	Endometrial cancer and somatic G>T KRAS transversion in patients with constitutional MUTYH biallelic mutations. Cancer Letters, 2009, 274, 266-270.	7.2	24
96	Tacrolimus causes reduced GLI1 expression and phenotypic changes in the TE 354.T basal cell carcinoma cell line. Journal of Dermatological Science, 2009, 54, 52-54.	1.9	3
97	Evidence of a four-hit mechanism involvingSMARCB1andNF2in schwannomatosis-associated schwannomas. Human Mutation, 2008, 29, 227-231.	2.5	167
98	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	2.5	782
99	Locus-specific databases and recommendations to strengthen their contribution to the classification of variants in cancer susceptibility genes. Human Mutation, 2008, 29, 1273-1281.	2.5	41
100	A proteomics approach to identify changes in protein profiles in serum of Familial Adenomatous Polyposis patients. Cancer Letters, 2008, 272, 40-52.	7.2	22
101	Somatic hypermutability of microsatellite sequences in Turcot syndrome: Implications for forensic genetics. Forensic Science International: Genetics Supplement Series, 2008, 1, 557-558.	0.3	0
102	Interleukin-10 promoter polymorphisms influence susceptibility to ulcerative colitis in a gender-specific manner. Scandinavian Journal of Gastroenterology, 2008, 43, 712-718.	1.5	50
103	NF2 Mutation Screening by Denaturing High-Performance Liquid Chromatography and High-Resolution Melting Analysis. Genetic Testing and Molecular Biomarkers, 2008, 12, 311-318.	1.7	17
104	Genomic rearrangements of the CDKN2A locus are infrequent in Italian malignant melanoma families without evidence of CDKN2A/CDK4 point mutations. Melanoma Research, 2008, 18, 431-437.	1.2	9
105	Genotype-phenotype correlations in individuals with a founder mutation in the MLH1 gene and hereditary non-polyposis colorectal cancer. Scandinavian Journal of Gastroenterology, 2007, 42, 746-753.	1.5	10
106	The p.G23S CDKN2A founder mutation in high-risk melanoma families from Central Italy. Melanoma Research, 2007, 17, 387-392.	1.2	20
107	Lone and secondary nonvalvular atrial fibrillation: Role of a genetic susceptibility. International Journal of Cardiology, 2007, 120, 59-65.	1.7	42
108	Genetic STRs variation in a large population from Tuscany (Italy). Forensic Science International: Genetics, 2007, 1, e10-e11.	3.1	7

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109	A Mononucleotide Markers Panel to Identify hMLH1/hMSH2 Germline Mutations. Disease Markers, 2007, 23, 179-187.	1.3	24
110	Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. Human Mutation, 2007, 28, 459-468.	2.5	41
111	Reply to Jaskowski et al. European Journal of Human Genetics, 2007, 15, 141-142.	2.8	2
112	Frequency of constitutional MSH6 mutations in a consecutive series of families with clinical suspicion of HNPCC. Clinical Genetics, 2007, 72, 230-237.	2.0	16
113	Fatal Malonyl CoA Decarboxylase Deficiency Due to Maternal Uniparental Isodisomy of the Telomeric End of Chromosome 16. Annals of Human Genetics, 2007, 71, 705-712.	0.8	21
114	A Single Mutation in the FGA Locus Responsible for False Homozygosities and Discrepancies Between Commercial Kits in an Unusual Paternity Test Case. Journal of Forensic Sciences, 2007, 52, 393-396.	1.6	17
115	Identification and Classification of Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome): Adapting Old Concepts to Recent Advancements. Report from the Italian Association for the Study of Hereditary Colorectal Tumors Consensus Group. Diseases of the Colon and Rectum, 2007, 50, 2126-2134.	1.3	12
116	Introduction of the DNase in forensic analysis. International Congress Series, 2006, 1288, 607-609.	0.2	0
117	Endothelial nitric oxide synthase gene influences the risk of pre-eclampsia, the recurrence of negative pregnancy events, and the maternalâ€“fetal flow. Journal of Hypertension, 2006, 24, 1823-1829.	0.5	38
118	The use of microsatellite instability, immunohistochemistry and other variables in determining the clinical significance of MLH1 and MSH2 unclassified variants in Lynch syndrome. Cancer Biomarkers, 2006, 2, 11-27.	1.7	14
119	A genetic model for determining MSH2 and MLH1 carrier probabilities based on family history and tumor microsatellite instability. Clinical Genetics, 2006, 69, 254-262.	2.0	20
120	Stability of BAT26 in tumours of hereditary nonpolyposis colorectal cancer patients with MSH2 intragenic deletion. European Journal of Human Genetics, 2006, 14, 63-68.	2.8	39
121	SDH Mutations in Patients Affected by Paraganglioma Syndromes: A Personal Experience. Annals of the New York Academy of Sciences, 2006, 1073, 183-189.	3.8	10
122	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
123	Analysis of minK and eNOS genes as candidate loci for predisposition to non-valvular atrial fibrillation. European Heart Journal, 2006, 27, 1712-1718.	2.2	84
124	Human Bone Marrow MSC Transformation in Different Culture Conditions.. Blood, 2006, 108, 4253-4253.	1.4	0
125	A novel microdeletion syndrome with loss of the MSH2 locus and hereditary nonâ€polyposis colorectal cancer. Clinical Genetics, 2005, 67, 178-182.	2.0	11
126	Microsatellite instability is not related to response to cisplatin-based chemotherapy in cervical cancer. International Journal of Gynecological Cancer, 2005, 15, 308-311.	2.5	7

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127	A kindred withMYH-associated polyposis and pilomatricomas. American Journal of Medical Genetics, Part A, 2005, 134A, 212-214.	1.2	50
128	Molecular Genetic Alterations and Clinical Features in Early-Onset Colorectal Carcinomas and Their Role for the Recognition of Hereditary Cancer Syndromes. American Journal of Gastroenterology, 2005, 100, 2280-2287.	0.4	66
129	Relationships between promoter polymorphisms in the thymidylate synthase gene and mRNA levels in colorectal cancers. European Journal of Cancer, 2005, 41, 2176-2183.	2.8	79
130	Aetiology of colorectal cancer and relevance of monogenic inheritance. Gut, 2004, 53, 115-122.	12.1	33
131	A rare combination consisting of aldosterone-producing adenoma and adrenal myelolipoma in a patient with heterozygosity for retinoblastoma (RB) gene. JRAAS - Journal of the Renin-Angiotensin-Aldosterone System, 2004, 5, 45-48.	1.7	8
132	Genetic testing among high-risk individuals in families with hereditary nonpolyposis colorectal cancer. British Journal of Cancer, 2004, 90, 882-887.	6.4	57
133	Susceptibility to Refractory Ulcerative Colitis Is Associated with Polymorphism in the hMLH1 Mismatch Repair Gene. Inflammatory Bowel Diseases, 2004, 10, 705-708.	1.9	25
134	Nonhomologous Robertsonian translocations(NHRTs) and uniparental disomy(UPD) risk: an Italian multicentric prenatal survey. Prenatal Diagnosis, 2004, 24, 647-652.	2.3	28
135	Inherited cancer predisposition. American Journal of Medical Genetics Part A, 2004, 129C, 1-4.	2.4	0
136	Simple and complex genetics of colorectal cancer susceptibility. American Journal of Medical Genetics Part A, 2004, 129C, 35-43.	2.4	25
137	Cancer risk in hereditary nonpolyposis colorectal cancer due to MSH6 mutations: impact on counseling and surveillance. Gastroenterology, 2004, 127, 17-25.	1.3	536
138	Hereditary nonpolyposis colorectal cancer and related conditions. American Journal of Medical Genetics Part A, 2003, 122A, 325-334.	2.4	70
139	Maternal-Fetal Flow, Negative Events, and Preeclampsia. Hypertension, 2003, 41, 932-937.	2.7	85
140	Different molecular mechanisms underlie genomic deletions in theMLH1 Gene. Human Mutation, 2002, 20, 368-374.	2.5	34
141	Two novel mutations and a new STK11/LKB1 gene isoform in Peutz-Jeghers patients. Human Mutation, 2002, 20, 78-79.	2.5	37
142	BRCA1-Related Malignancies in a Family Presenting with von Recklinghausen's Disease. Gynecologic Oncology, 2002, 86, 375-378.	1.4	39
143	Multiple lipomas linked to an RB1 gene mutation in a large pedigree with low penetrance retinoblastoma. European Journal of Human Genetics, 2001, 9, 690-694.	2.8	49
144	Investigation of G2-phase chromosomal radiosensitivity in hereditary non-polyposis colorectal cancer cells. International Journal of Radiation Biology, 2001, 77, 773-780.	1.8	5

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145	Molecular Screening for Hereditary Nonpolyposis Colorectal Cancer: A Prospective, Population-Based Study. Journal of Clinical Oncology, 2001, 19, 3944-3950.	1.6	101
146	Microsatellite Instability Is an Independent Indicator of Recurrence in Sporadic Stage I-II Endometrial Adenocarcinoma. Journal of Clinical Oncology, 2001, 19, 1008-1014.	1.6	57
147	Familial microsatellite-stable non-polyposis colorectal cancer: Incidence and characteristics in a clinic-based population. Annals of Oncology, 2001, 12, 813-818.	1.2	7
148	Four novelMSH2 andMLH1 frameshift mutations and occurrence of a breast cancer phenocopy in hereditary nonpolyposis colorectal cancer. Human Mutation, 2001, 17, 521-521.	2.5	17
149	CDKN2A germline splicing mutation affecting both p16ink4 and p14arf RNA processing in a melanoma/neurofibroma kindred. Genes Chromosomes and Cancer, 2001, 31, 398-401.	2.8	65
150	Mutations of the 'minor' mismatch repair gene MSH6 in typical and atypical hereditary nonpolyposis colorectal cancer. Familial Cancer, 2001, 1, 95-101.	1.9	24
151	Clinical and biologic heterogeneity of hereditary nonpolyposis colorectal cancer. International Journal of Cancer, 2001, 95, 323-328.	5.1	19
152	Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts. , 2000, 27, 424-429.		19
153	Investigation of the substrate spectrum of the human mismatch-specific DNAN-glycosylase MED1 (MBD4): Fundamental role of the catalytic domain. Journal of Cellular Physiology, 2000, 185, 473-480.	4.1	101
154	Biphasic Kinetics of the Human DNA Repair Protein MED1 (MBD4), a Mismatch-specific DNA N-Glycosylase. Journal of Biological Chemistry, 2000, 275, 32422-32429.	3.4	157
155	Problems in the identification of hereditary nonpolyposis colorectal cancer in two families with late development of full-blown clinical spectrum. American Journal of Gastroenterology, 2000, 95, 2110-2115.	0.4	3
156	Microsatellite instability in gastric carcinogenesis. Gastroenterology, 2000, 118, A57.	1.3	0
157	Problems in the identification of hereditary nonpolyposis colorectal cancer in two families with late development of full-blown clinical spectrum. American Journal of Gastroenterology, 2000, 95, 2110-2115.	0.4	2
158	Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts. Genes Chromosomes and Cancer, 2000, 27, 424-429.	2.8	1
159	MED1, a novel human methyl-CpG-binding endonuclease, interacts with DNA mismatch repair protein MLH1. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 3969-3974.	7.1	239
160	The DNA repair gene MBD4 (MED1) is mutated in human carcinomas with microsatellite instability. Nature Genetics, 1999, 23, 266-268.	21.4	211
161	Mosaic trisomy 17 in amniocytes: phenotypic outcome, tissue distribution, and uniparental disomy studies. European Journal of Human Genetics, 1999, 7, 421-426.	2.8	36
162	Assessment of pathogenicity criteria for constitutional missense mutations of the hereditary nonpolyposis colorectal cancer genes MLH1 and MSH2. European Journal of Human Genetics, 1999, 7, 778-782.	2.8	31

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163	MLH1 and MSH2 constitutinal mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer. , 1998, 75, 835-839.		50
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