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	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
2	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
3	Cancer risk in hereditary nonpolyposis colorectal cancer due to MSH6 mutations: impact on counseling and surveillance. Gastroenterology, 2004, 127, 17-25.	1.3	536
4	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
5	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
6	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
7	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
8	The DNA repair gene MBD4 (MED1) is mutated in human carcinomas with microsatellite instability. Nature Genetics, 1999, 23, 266-268.	21.4	211
9	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. EBioMedicine, 2017, 20, 39-49.	6.1	170
10	Evidence of a four-hit mechanism involvingSMARCB1andNF2in schwannomatosis-associated schwannomas. Human Mutation, 2008, 29, 227-231.	2.5	167
11	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
12	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
13	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
14	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
15	A new point mutation of the prion protein gene in Creutzfeldtâ€Jakob disease. Annals of Neurology, 1993, 34, 802-807.	5.3	104
16	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
17	Molecular Screening for Hereditary Nonpolyposis Colorectal Cancer: A Prospective, Population-Based Study. Journal of Clinical Oncology, 2001, 19, 3944-3950.	1.6	101
18	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

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19	Schwannomatosis associated with multiple meningiomas due to a familial SMARCB1 mutation. Neurogenetics, 2010, 11, 73-80.	1.4	90
20	Maternal-Fetal Flow, Negative Events, and Preeclampsia. Hypertension, 2003, 41, 932-937.	2.7	85
21	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
22	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
23	Polymorphisms of the prion protein gene in Italian patients with Creutzfeldt-Jakob disease. Human Genetics, 1994, 94, 375-9.	3.8	80
24	Hereditary nonpolyposis colorectal cancer: Review of clinical, molecular genetics, and counseling aspects. , 1996, 62, 353-364.		79
25	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
26	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
27	Hereditary nonpolyposis colorectal cancer and related conditions. American Journal of Medical Genetics Part A, 2003, 122A, 325-334.	2.4	70
28	Characterization ofMSH2 andMLH1 mutations in Italian families with hereditary nonpolyposis colorectal cancer. , 1997, 18, 8-18.		67
29	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
30	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
31	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
32	A split hand-split foot (SHFM3) gene is located at 10Q24â†'25. American Journal of Medical Genetics Part A, 1996, 62, 427-436.	2.4	57
33	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
34	Genetic testing among high-risk individuals in families with hereditary nonpolyposis colorectal cancer. British Journal of Cancer, 2004, 90, 882-887.	6.4	57
35	Characterization of MLH1 and MSH2 alternative splicing and its relevance to molecular testing of colorectal cancer susceptibility. Human Genetics, 1998, 102, 15-20.	3.8	56
36	Split hand/split foot anomaly in a family segregating a balanced translocation with breakpoint on 7q22.1. American Journal of Medical Genetics Part A, 1993, 47, 823-831.	2.4	51

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37	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
38	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
39	Survival analysis in families affected by hereditary non-polyposis colorectal cancer. , 1997, 71, 373-376.		50
40	MLH1 and MSH2 constitutinal mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer. , 1998, 75, 835-839.		50
41	A kindred withMYH-associated polyposis and pilomatricomas. American Journal of Medical Genetics, Part A, 2005, 134A, 212-214.	1.2	50
42	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
43	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
44	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
45	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
46	Analysis of 138 consecutive ovarian cancer patients: Incidence and characteristics of familial cases. Gynecologic Oncology, 1990, 39, 300-304.	1.4	47
47	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
48	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
49	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	1.8	43
50	Lone and secondary nonvalvular atrial fibrillation: Role of a genetic susceptibility. International Journal of Cardiology, 2007, 120, 59-65.	1.7	42
51	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
52	The CFC syndromeâ€"report of the first two cases outside the United States. American Journal of Medical Genetics Part A, 1987, 27, 767-771.	2.4	41
53	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
54	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

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55	BRCA1-Related Malignancies in a Family Presenting with von Recklinghausen's Disease. Gynecologic Oncology, 2002, 86, 375-378.	1.4	39
56	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
57	A PALB2 germline mutation associated with hereditary breast cancer in Italy. Familial Cancer, 2010, 9, 181-185.	1.9	39
58	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
59	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
60	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
61	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
62	Oral-facial-skeletal syndromes. American Journal of Medical Genetics Part A, 1995, 59, 365-368.	2.4	37
63	Two novel mutations and a new STK11/LKB1 gene isoform in Peutz-Jeghers patients. Human Mutation, 2002, 20, 78-79.	2.5	37
64	The growing complexity of the intestinal polyposis syndromes. American Journal of Medical Genetics, Part A, 2013, 161, 2777-2787.	1.2	37
65	Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility. Journal of Medical Genetics, 2018, 55, 669-674.	3.2	37
66	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
67	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
68	Different molecular mechanisms underlie genomic deletions in theMLH1 Gene. Human Mutation, 2002, 20, 368-374.	2.5	34
69	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
70	Aetiology of colorectal cancer and relevance of monogenic inheritance. Gut, 2004, 53, 115-122.	12.1	33
71	The policy of public health genomics in Italy. Health Policy, 2013, 110, 214-219.	3.0	33
72	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

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73	Creutzfeldt-Jakob disease after non-commercial dura mater graft. Lancet, The, 1992, 340, 614-615.	13.7	32
74	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. Pediatric Nephrology, 2009, 24, 2147-2153.	1.7	32
75	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
76	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
77	Proximal femoral focal deficiency (PFFD) and fibular a/hypoplasia (FA/H): A model of a developmental field defect. American Journal of Medical Genetics Part A, 1995, 55, 427-432.	2.4	30
78	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
79	Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). Clinical Endocrinology, 2010, 72, 839-844.	2.4	29
80	Assessment of the InSiGHT Interpretation Criteria for the Clinical Classification of 24MLH1andMSH2Gene Variants. Human Mutation, 2017, 38, 64-77.	2.5	29
81	Nonhomologous Robertsonian translocations(NHRTs) and uniparental disomy(UPD) risk: an Italian multicentric prenatal survey. Prenatal Diagnosis, 2004, 24, 647-652.	2.3	28
82	Fabry disease: polymorphic haplotypes and a novel missense mutation in the GLA gene. Clinical Genetics, 2012, 81, 224-233.	2.0	28
83	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
84	Gene variants of unknown clinical significance in Lynch syndrome. An introduction for clinicians. Familial Cancer, 2013, 12, 181-187.	1.9	27
85	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
86	Constitutional trisomy 8 and myelodysplasia: Report of a case and review of the literature. Leukemia Research, 1995, 19, 733-736.	0.8	25
87	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
88	Simple and complex genetics of colorectal cancer susceptibility. American Journal of Medical Genetics Part A, 2004, 129C, 35-43.	2.4	25
89	Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). European Journal of Endocrinology, 2010, 163, 329-337.	3.7	25
90	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

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91	Partial tetrasomy 9 in an infant with clinical and radiological evidence of multiple joint dislocations. European Journal of Pediatrics, 1988, 147, 645-648.	2.7	24
92	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
93	A Mononucleotide Markers Panel to Identify hMLH1/hMSH2 Germline Mutations. Disease Markers, 2007, 23, 179-187.	1.3	24
94	Endometrial cancer and somatic G>T KRAS transversion in patients with constitutional MUTYH biallelic mutations. Cancer Letters, 2009, 274, 266-270.	7.2	24
95	Double autosomal/gonosomal mosaic aneuploidy: study of nondisjunction in two cases with trisomy of chromosome 8. Human Genetics, 1995, 95, 519-25.	3.8	23
96	Constitutional mismatch repair deficiencyâ€“associated brain tumors: report from the European C4CMMRD consortium. Neuro-Oncology Advances, 2019, 1, vdz033.	0.7	23
97	Ulnar ray defect in an infant with a 6q21;7q31.2 translocation: Further evidence for the existence of a limb defect gene in 6q21. American Journal of Medical Genetics Part A, 1995, 55, 315-318.	2.4	22
98	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
99	MLH1constitutional and somatic methylation in patients with MLH1 negative tumors fulfilling the revised Bethesda criteria. Epigenetics, 2014, 9, 1431-1438.	2.7	22
100	Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987.	6.4	22
101	Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. European Journal of Medical Genetics, 2021, 64, 104350.	1.3	22
102	A girl with G syndrome and agenesis of the corpus callosum. American Journal of Medical Genetics Part A, 1987, 28, 287-291.	2.4	21
103	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
104	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
105	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
106	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
107	Localization of the HLA class 11-associated invariant chain gene to human chromosome band 5832. Immunogenetics, 1988, 28, 53-56.	2.4	20
108	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

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109	The p.G23S CDKN2A founder mutation in high-risk melanoma families from Central Italy. Melanoma Research, 2007, 17, 387-392.	1.2	20
110	Effects of dexamethasone on the growth and epidermal growth factor receptor expression of the OVCA 433 ovarian cancer cells. Molecular and Cellular Endocrinology, 1992, 83, 183-193.	3.2	19
111	Cerebro-reno-digital (Meckel-like) syndrome with Dandy-Walker malformation, cystic kidneys, hepatic fibrosis, and polydactyly. American Journal of Medical Genetics Part A, 1993, 47, 50-53.	2.4	19
112	Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts. , 2000, 27, 424-429.		19
113	Clinical and biologic heterogeneity of hereditary nonpolyposis colorectal cancer. International Journal of Cancer, 2001, 95, 323-328.	5.1	19
114	Dicentric chromosome Y associated with Leydig cell agenesis and sex reversal. Clinical Genetics, 1995, 47, 38-41.	2.0	19
115	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
116	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
117	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
118	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
119	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
120	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
121	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
122	Gene for Simpson-Golabi-Behmel syndrome is linked to HPRT in Xq26 in two European families. American Journal of Medical Genetics Part A, 1994, 50, 388-390.	2.4	16
123	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
124	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
125	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
126	PCR detection of an insertion/deletion polymorphism in intron 1 of the HRAS1 locus. Nucleic Acids Research, 1992, 20, 1157-1157.	14.5	15

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127	Long-term cytogenetic effects of antineoplastic treatment in relation to secondary leukemia. Cancer Genetics and Cytogenetics, 1988, 33, 201-211.	1.0	14
128	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
129	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
130	ESHG warns against misuses of genetic tests and biobanks for discrimination purposes. European Journal of Human Genetics, 2021, 29, 894-896.	2.8	14
131	Brachy/ectrodactyly and absence or hypoplasia of the fibula: an autosomal dominant condition with low penetrance and variable expressivity. Clinical Genetics, 1990, 38, 321-326.	2.0	13
132	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
133	Genes for split hand/split foot and laterality defects on 7q22.1 and xq24-q27.1. American Journal of Medical Genetics Part A, 1994, 50, 101-101.	2.4	12
134	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
135	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
136	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
137	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
138	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
139	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
140	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
141	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
142	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
143	Role of extensive diagnostic workup in young athletes and nonathletes with complex ventricular arrhythmias. Heart Rhythm, 2020, 17, 230-237.	0.7	10
144	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

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145	Duodenal carcinoma in a 37-year-old man with Cowden/Bannayan syndrome. Digestive and Liver Disease, 2013, 45, 75-78.	0.9	9
146	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
147	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
148	Prognostic Relevance of MLH1 and MSH2 Mutations in Hereditary Non-Polyposis Colorectal Cancer Patients. Tumori, 2009, 95, 731-738.	1.1	8
149	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
150	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
151	Genetic STRs variation in a large population from Tuscany (Italy). Forensic Science International: Genetics, 2007, 1, e10-e11.	3.1	7
152	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
153	The Current Practice of Lynch Syndrome Diagnosis and Management in Italy: A Qualitative Assessment. Public Health Genomics, 2019, 22, 189-207.	1.0	7
154	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
155	Limb-pelvis hypoplasia/aplasia: A discrete entity in the fibuloulnar developmental field complex. , 1997, 68, 190-194.		6
156	Workload measurement for molecular genetics laboratory: A survey study. PLoS ONE, 2018, 13, e0206855.	2.5	6
157	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
158	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
159	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
160	Gastrointestinal manifestations in PTEN hamartoma tumor syndrome. Bailliere's Best Practice and Research in Clinical Gastroenterology, 2022, 58-59, 101792.	2.4	6
161	Chronic myelogenous leukemia in the course of chronic lymphocytic leukemia: Evidence for an independent clonal origin. Leukemia Research, 1991, 15, 269-273.	0.8	5
162	Progressive Dementia in a Young Patient with a Homozygous Deletion of the PrP Gene.. Annals of the New York Academy of Sciences, 1994, 724, 358-360.	3.8	5

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164	Molecular Genetics of Hereditary Non-Polyposis Colorectal Cancer (HNPCC). Tumori, 1996, 82, 122-135.	1.1	5
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