Robert Hofstra

List of Publications by Year
in descending order

188 papers	13,401 citations	22153 59 h-index	25787 108 g-index
193 all docs	193 docs citations	193 times ranked	18974 citing authors

#	Article	IF	Citations
1	Universal Immunohistochemistry for Lynch Syndrome: A Systematic Review and Meta-analysis of 58,580 Colorectal Carcinomas. Clinical Gastroenterology and Hepatology, 2022, 20, e496-e507.	4.4	14
2	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	8.2	24
3	Loss of enteric neuronal Ndrg4 promotes colorectal cancer via increased release of Nid1 and Fbln2. EMBO Reports, 2021, 22, e51913.	4.5	14
4	Size matters: Large copy number losses in Hirschsprung disease patients reveal genes involved in enteric nervous system development. PLoS Genetics, 2021, 17, e1009698.	3.5	14
5	Intestinal multicellular organoids to study colorectal cancer. Biochimica Et Biophysica Acta: Reviews on Cancer, 2021, 1876, 188586.	7.4	13
6	Using Out-of-Batch Reference Populations to Improve Untargeted Metabolomics for Screening Inborn Errors of Metabolism. Metabolites, 2021, 11, 8.	2.9	14
7	The Somatic Mutation Paradigm in Congenital Malformations: Hirschsprung Disease as a Model. International Journal of Molecular Sciences, 2021, 22, 12354.	4.1	3
8	TALPID3/KIAA0586 Regulates Multiple Aspects of Neuromuscular Patterning During Gastrointestinal Development in Animal Models and Human. Frontiers in Molecular Neuroscience, 2021, 14, 757646.	2.9	3
9	Yield of Lynch Syndrome Surveillance for Patients With Pathogenic Variants in DNA Mismatch Repair Genes. Clinical Gastroenterology and Hepatology, 2020, 18, 1112-1120.e1.	4.4	14
10	Germline genome editing: public dialogue is urgent but not self-evident. European Journal of Human Genetics, 2020, 28, 4-5.	2.8	4
11	Three-step site-directed mutagenesis screen identifies pathogenic MLH1 variants associated with Lynch syndrome. Journal of Medical Genetics, 2020, 57, 308-315.	3.2	5
12	Goldbergâ€“Shprintzen syndrome is determined by the absence, or reduced expression levels, of KIFBP. Human Mutation, 2020, 41, 1906-1917.	2.5	6
13	Inhibition of ROCK signaling pathway accelerates enteric neural crest cellâ€based therapy after transplantation in a rat hypoganglionic model. Neurogastroenterology and Motility, 2020, 32, e13895.	3.0	6
14	Infantile hypertrophic pyloric stenosis in patients with esophageal atresia. Birth Defects Research, 2020, 112, 670-687.	1.5	1
15	Zebrafish: A Model Organism for Studying Enteric Nervous System Development and Disease. Frontiers in Cell and Developmental Biology, 2020, 8, 629073.	3.7	27
16	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. American Journal of Human Genetics, 2020, 106, 779-792.	6.2	25
17	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
18	Biallelic Variants in ASNA1 , Encoding a Cytosolic Targeting Factor of Tail-Anchored Proteins, Cause Rapidly Progressive Pediatric Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, 397-406.	3.6	16

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19	Exome chip association study excluded the involvement of rare coding variants with large effect sizes in the etiology of anorectal malformations. PLoS ONE, 2019, 14, e0217477.	2.5	3
20	Cardiac Phenotypes, Genetics, and RisksÂin Familial Noncompaction Cardiomyopathy. Journal of the American College of Cardiology, 2019, 73, 1601-1611.	2.8	65
21	Do RET somatic mutations play a role in Hirschsprung disease?. Genetics in Medicine, 2018, 20, 1477-1478.	2.4	7
22	Genetics, Clinical Features, and Long-TermÂOutcome of NoncompactionÂCardiomyopathy. Journal of the American College of Cardiology, 2018, 71, 711-722.	2.8	242
23	Identification of Variants in RET and IHH Pathway Members inÂaÂLarge Family With History of Hirschsprung Disease. Gastroenterology, 2018, 155, 118-129.e6.	1.3	25
24	Routine Molecular Analysis for Lynch Syndrome Among Adenomas or Colorectal Cancer Within a National Screening Program. Gastroenterology, 2018, 155, 1410-1415.	1.3	9
25	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1603-1610.	2.8	4
26	A "late-but-fitter revertant cell" explains the high frequency of revertant mosaicism in epidermolysis bullosa. PLoS ONE, 2018, 13, e0192994.	2.5	18
27	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
28	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129.	6.2	67
29	Loss of LMOD1 impairs smooth muscle cytocontractility and causes megacystis microcolon intestinal hypoperistalsis syndrome in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E2739-E2747.	7.1	97
30	Suspected Lynch syndrome associated MSH6 variants: A functional assay to determine their pathogenicity. PLoS Genetics, 2017, 13, e1006765.	3.5	18
31	Correspondence: SEMA4A variation and risk of colorectal cancer. Nature Communications, 2016, 7, 10611.	12.8	7
32	ACTG2variants impair actin polymerization in sporadic Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. Human Molecular Genetics, 2016, 25, 571-583.	2.9	56
33	White paper on guidelines concerning enteric nervous system stem cell therapy for enteric neuropathies. Developmental Biology, 2016, 417, 229-251.	2.0	112
34	Two new mutations of the CLMP gene identified in a newborn presenting congenital short-bowel syndrome. Clinics and Research in Hepatology and Gastroenterology, 2016, 40, e65-e67.	1.5	11
35	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	2.9	38
36	Genetics of enteric neuropathies. Developmental Biology, 2016, 417, 198-208.	2.0	44

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37	Regulators of gene expression in Enteric Neural Crest Cells are putative Hirschsprung disease genes. Developmental Biology, 2016, 416, 255-265.	2.0	31
38	Paroxysmal exercise-induced dystonia within the phenotypic spectrum ofECHS1deficiency. Movement Disorders, 2016, 31, 1041-1048.	3.9	58
39	Cardiovascular malformations caused by NOTCH1 mutations do not keep left: data on 428 probands with left-sided CHD and their families. Genetics in Medicine, 2016, 18, 914-923.	2.4	104
40	Common arterial trunk and in Lrp2 knock out mice indicate a crucial role of LRP2 in cardiac development. DMM Disease Models and Mechanisms, 2016, 9, 413-25.	2.4	33
41	Review: Clinical aspects of hereditary DNA Mismatch repair gene mutations. DNA Repair, 2016, 38, 155-162.	2.8	49
42	Biallelic Truncating Mutations in ALPK3 Cause Severe Pediatric Cardiomyopathy. Journal of the American College of Cardiology, 2016, 67, 515-525.	2.8	70
43	Oligonucleotide-directed mutagenesis screen to identify pathogenic Lynch syndrome-associated MSH2 DNA mismatch repair gene variants. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 4128-4133.	7.1	28
44	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Scientific Reports, 2015, 5, 16286.	3.3	24
45	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. American Journal of Human Genetics, 2015, 96, 581-596.	6.2	118
46	Hirschsprung Disease and Activation of Hedgehog Signaling via GLI1-3 Mutations. Gastroenterology, 2015, 149, 1672-1675.	1.3	6
47	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	2.8	238
48	ENDOCRINE TUMOURS: Progressive metastatic medullary thyroid carcinoma: first- and second-line strategies. European Journal of Endocrinology, 2015, 172, R241-R251.	3.7	22
49	Congenital Short Bowel Syndrome: from clinical and genetic diagnosis to the molecular mechanisms involved in intestinal elongation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2352-2361.	3.8	27
50	New Target Genes in Endometrial Tumors Show a Role for the Estrogen-Receptor Pathway in Microsatellite-Unstable Cancers. Human Mutation, 2014, 35, 1514-1523.	2.5	10
51	High Frequency ofRPL22Mutations in Microsatellite-Unstable Colorectal and Endometrial Tumors. Human Mutation, 2014, 35, 1442-1445.	2.5	38
52	Re: Role of the Oxidative DNA Damage Repair Gene OGG1 in Colorectal Tumorigenesis. Journal of the National Cancer Institute, 2014, 106, .	6.3	9
53	The MLH1 c.1852_1853delinsGC (p.K618A) Variant in Colorectal Cancer: Genetic Association Study in 18,723 Individuals. PLoS ONE, 2014, 9, e95022.	2.5	7
54	Contribution of rare and common variants determine complex diseasesâ€"Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119

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55	Response to: Design of a Core Classification Process for DNA Mismatch Repair Variations of A Priori Unknown Functional Significance. Human Mutation, 2013, 34, 923-924.	2.5	1
56	The entire miRâ€200 seed family is strongly deregulated in clear cell renal cell cancer compared to the proximal tubular epithelial cells of the kidney. Genes Chromosomes and Cancer, 2013, 52, 165-173.	2.8	26
57	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
58	Building a brain in the gut: development ofÂthe enteric nervous system. Clinical Genetics, 2013, 83, 307-316.	2.0	141
59	Brush border myosin Ia inactivation in gastric but not endometrial tumors. International Journal of Cancer, 2013, 132, 1790-1799.	5.1	21
60	TBX4mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension. Journal of Medical Genetics, 2013, 50, 500-506.	3.2	171
61	Congenital short bowel syndrome as the presenting symptom in male patients with FLNA mutations. Genetics in Medicine, 2013, 15, 310-313.	2.4	32
62	The Cardiac Phenotype in Patients With a CHD7 Mutation. Circulation: Cardiovascular Genetics, 2013, 6, 248-254.	5.1	53
63	Novel noâ€stop FLNA mutation causes multiâ€organ involvement in males. American Journal of Medical Genetics, Part A, 2013, 161, 2376-2384.	1.2	33
64	The Role of Maternal-Fetal Cholesterol Transport in Early Fetal Life: Current Insights1. Biology of Reproduction, 2013, 88, 24.	2.7	108
65	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
66	CLMP Is Essential for Intestinal Development, but Does Not Play a Key Role in Cellular Processes Involved in Intestinal Epithelial Development. PLoS ONE, 2013, 8, e54649.	2.5	17
67	Combined adverse effects of maternal smoking and high body mass index on heart development in offspring: evidence for interaction?. Heart, 2012, 98, 474-479.	2.9	42
68	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. European Journal of Human Genetics, 2012, 20, 917-920.	2.8	8
69	Clinical Relevance of ¹⁸F-FDG PET and ¹⁸F-DOPA PET in Recurrent Medullary Thyroid Carcinoma. Journal of Nuclear Medicine, 2012, 53, 1863-1871.	5.0	86
70	Phospholamban R14del mutation in patients diagnosed with dilated cardiomyopathy or arrhythmogenic right ventricular cardiomyopathy: evidence supporting the concept of arrhythmogenic cardiomyopathy. European Journal of Heart Failure, 2012, 14, 1199-1207.	7.1	369
71	Natural Gene Therapy in Dystrophic Epidermolysis Bullosa. Archives of Dermatology, 2012, 148, 213.	1.4	27
72	Pathological assessment of mismatch repair gene variants in Lynch syndrome: Past, present, and future. Human Mutation, 2012, 33, 1617-1625.	2.5	60

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73	CLMP Is Required for Intestinal Development, and Loss-of-Function Mutations Cause Congenital Short-Bowel Syndrome. Gastroenterology, 2012, 142, 453-462.e3.	1.3	49
74	The origin of fetal sterols in second-trimester amniotic fluid: endogenous synthesis or maternal-fetal transport?. American Journal of Obstetrics and Gynecology, 2012, 207, 202.e19-202.e25.	1.3	42
75	Mutation update on the CHD7 gene involved in CHARGE syndrome. Human Mutation, 2012, 33, 1149-1160.	2.5	224
76	Targeted exome sequencing in clear cell renal cell carcinoma tumors suggests aberrant chromatin regulation as a crucial step in ccRCC development. Human Mutation, 2012, 33, 1059-1062.	2.5	74
77	A novel classification system to predict the pathogenic effects of CHD7 missense variants in CHARGE syndrome. Human Mutation, 2012, 33, 1251-1260.	2.5	65
78	What monozygotic twins discordant for phenotype illustrate about mechanisms influencing genetic forms of neurodegeneration. Clinical Genetics, 2012, 81, 325-333.	2.0	36
79	RET/PTC rearrangement is prevalent in follicular HÃ¼rthle cell carcinomas. Histopathology, 2012, 61, 833-843.	2.9	42
80	A rapid and cell-free assay to test the activity of lynch syndrome-associated MSH2 and MSH6 missense variants. Human Mutation, 2012, 33, 488-494.	2.5	46
81	Candidate driver genes in microsatelliteâ€unstable colorectal cancer. International Journal of Cancer, 2012, 130, 1558-1566.	5.1	99
82	Functional analyses of RET mutations in Chinese hirschsprung disease patients. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 47-51.	1.6	5
83	Variants in RET Associated With Hirschsprung's Disease Affect Binding of Transcription Factors and Gene Expression. Gastroenterology, 2011, 140, 572-582.e2.	1.3	38
84	Paediatric intestinal cancer and polyposis due to bi-allelic PMS2 mutations: Case series, review and follow-up guidelines. European Journal of Cancer, 2011, 47, 965-982.	2.8	64
85	Haplotype sharing test maps genes for familial cardiomyopathiesâ€. Clinical Genetics, 2011, 79, 459-467.	2.0	6
86	Perspectives for tailored chemoprevention and treatment of colorectal cancer in Lynch syndrome. Critical Reviews in Oncology/Hematology, 2011, 80, 264-277.	4.4	11
87	The international dystrophic epidermolysis bullosa patient registry: An online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. Human Mutation, 2011, 32, 1100-1107.	2.5	74
88	Focal Adhesion Kinase (FAK) Binds RET Kinase via Its FERM Domain, Priming a Direct and Reciprocal RET-FAK Transactivation Mechanism. Journal of Biological Chemistry, 2011, 286, 17292-17302.	3.4	50
89	The Effects of Four Different Tyrosine Kinase Inhibitors on Medullary and Papillary Thyroid Cancer Cells. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E991-E995.	3.6	77
90	The inversa type of recessive dystrophic epidermolysis bullosa is caused by specific arginine and glycine substitutions in type VII collagen. Journal of Medical Genetics, 2011, 48, 160-167.	3.2	35

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91	Trans-eQTLs Reveal That Independent Genetic Variants Associated with a Complex Phenotype Converge on Intermediate Genes, with a Major Role for the HLA. PLoS Genetics, 2011, 7, e1002197.	3.5	324
92	Recurrent and founder mutations in the Netherlands. Netherlands Heart Journal, 2010, 18, 583-591.	0.8	33
93	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. American Journal of Human Genetics, 2010, 87, 60-74.	6.2	230
94	Screening for germline DND1 mutations in testicular cancer patients. Familial Cancer, 2010, 9, 439-442.	1.9	5
95	An updated and upgradedL1CAMmutation database. Human Mutation, 2010, 31, E1102-E1109.	2.5	54
96	Cell-free assay breakthrough forMLH1variants. Human Mutation, 2010, 31, v-v.	2.5	0
97	A genome-wide association study of Hodgkin's lymphoma identifies new susceptibility loci at 2p16.1 (REL), 8q24.21 and 10p14 (GATA3). Nature Genetics, 2010, 42, 1126-1130.	21.4	177
98	Mutations in SCG10 Are Not Involved in Hirschsprung Disease. PLoS ONE, 2010, 5, e15144.	2.5	6
99	KBP interacts with SCG10, linking Goldbergâ€“Shprintzen syndrome to microtubule dynamics and neuronal differentiation. Human Molecular Genetics, 2010, 19, 3642-3651.	2.9	37
100	Genotype-phenotype correlations in L1 syndrome: a guide for genetic counselling and mutation analysis. Journal of Medical Genetics, 2010, 47, 169-175.	3.2	82
101	Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours. Human Molecular Genetics, 2010, 19, 697-706.	2.9	26
102	Histone Methyltransferase Gene SETD2 Is a Novel Tumor Suppressor Gene in Clear Cell Renal Cell Carcinoma. Cancer Research, 2010, 70, 4287-4291.	0.9	216
103	Identification of MOAG-4/SERF as a Regulator of Age-Related Proteotoxicity. Cell, 2010, 142, 601-612.	28.9	120
104	Estrogens, MSI and Lynch syndrome-associated tumors. Biochimica Et Biophysica Acta: Reviews on Cancer, 2009, 1796, 194-200.	7.4	14
105	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	2.5	57
106	A genetic variants database for arrhythmogenic right ventricular dysplasia/cardiomyopathy. Human Mutation, 2009, 30, 1278-1283.	2.5	105
107	PMS2 involvement in patients suspected of Lynch syndrome. Genes Chromosomes and Cancer, 2009, 48, 322-329.	2.8	34
108	Biochemical characterization of MLH3 missense mutations does not reveal an apparent role of MLH3 in Lynch syndrome. Genes Chromosomes and Cancer, 2009, 48, 340-350.	2.8	18

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109	Do microsatellite instability profiles really differ between colorectal and endometrial tumors?. Genes Chromosomes and Cancer, 2009, 48, 552-557.	2.8	10
110	Germline hypermethylation of MLH1 and EPCAM deletions are a frequent cause of Lynch syndrome. Genes Chromosomes and Cancer, 2009, 48, 737-744.	2.8	186
111	TP53 germline mutations in Portugal and genetic modifiers of age at cancer onset. Familial Cancer, 2009, 8, 383-390.	1.9	14
112	Mononucleotide precedes dinucleotide repeat instability during colorectal tumour development in Lynch syndrome patients. Journal of Pathology, 2009, 219, 96-102.	4.5	22
113	Complex pathogenesis of Hirschsprung's disease in a patient with hydrocephalus, vesico-ureteral reflux and a balanced translocation t(3;17)(p12;q11). European Journal of Human Genetics, 2009, 17, 483-490.	2.8	26
114	Long-term follow-up of patients with recessive dystrophic epidermolysis bullosa in the Netherlands: Expansion of the mutation database and unusual phenotypeâ€“genotype correlations. Journal of Dermatological Science, 2009, 56, 9-18.	1.9	27
115	Severe cardiac phenotype with right ventricular predominance in a large cohort of patients with a single missense mutation in the DES gene. Heart Rhythm, 2009, 6, 1574-1583.	0.7	143
116	Survival-Related Profile, Pathways, and Transcription Factors in Ovarian Cancer. PLoS Medicine, 2009, 6, e1000024.	8.4	156
117	A novel MSH2 germline mutation in a Druze HNPCC family. Familial Cancer, 2008, 7, 135-139.	1.9	4
118	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
119	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. Human Mutation, 2008, 29, 1292-1303.	2.5	54
120	Assessment of functional effects of unclassified genetic variants. Human Mutation, 2008, 29, 1314-1326.	2.5	93
121	A database to support the interpretation of human mismatch repair gene variants. Human Mutation, 2008, 29, 1337-1341.	2.5	51
122	Somatic mutations in mismatch repair genes in sporadic gastric carcinomas are not a cause but a consequence of the mutator phenotype. Cancer Genetics and Cytogenetics, 2008, 180, 110-114.	1.0	26
123	C. elegans Model Identifies Genetic Modifiers of Î±-Synuclein Inclusion Formation During Aging. PLoS Genetics, 2008, 4, e1000027.	3.5	370
124	A New Perspective on Transcriptional System Regulation (TSR): Towards TSR Profiling. PLoS ONE, 2008, 3, e1656.	2.5	11
125	Ras/ERK1/2-mediated STAT3 Ser727 Phosphorylation by Familial Medullary Thyroid Carcinoma-associated RET Mutants Induces Full Activation of STAT3 and Is Required for c-fos Promoter Activation, Cell Mitogenicity, and Transformation. Journal of Biological Chemistry, 2007, 282, 6415-6424.	3.4	52
126	Sorafenib Functions to Potently Suppress RET Tyrosine Kinase Activity by Direct Enzymatic Inhibition and Promoting RET Lysosomal Degradation Independent of Proteasomal Targeting. Journal of Biological Chemistry, 2007, 282, 29230-29240.	3.4	90

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127	Nuclear localization of human DNA mismatch repair protein exonuclease 1 (hEXO1). Nucleic Acids Research, 2007, 35, 2609-2619.	14.5	30
128	High yield of LMNA mutations in patients with dilated cardiomyopathy and/or conduction disease referred to cardiogenetics outpatient clinics. American Heart Journal, 2007, 154, 1130-1139.	2.7	150
129	MEIS and PBX homeobox proteins in ovarian cancer. European Journal of Cancer, 2007, 43, 2495-2505.	2.8	79
130	Severe Myocardial Fibrosis Caused by a Deletion of the 5â€™ End of the Lamin A/C Gene. Journal of the American College of Cardiology, 2007, 49, 2430-2439.	2.8	79
131	Evidence Based Selection of Housekeeping Genes. PLoS ONE, 2007, 2, e898.	2.5	617
132	Getting rid of thePMS2 pseudogenes: mission impossible?. Human Mutation, 2007, 28, 414-414.	2.5	12
133	Functional analysis helps to clarify the clinical importance of unclassified variants in DNA mismatch repair genes. Human Mutation, 2007, 28, 1047-1054.	2.5	51
134	MUTYH and the mismatch repair system: partners in crime?. Human Genetics, 2006, 119, 206-211.	3.8	32
135	Current concepts in RET-related genetics, signaling and therapeutics. Trends in Genetics, 2006, 22, 627-636.	6.7	101
136	A biological question and a balanced (orthogonal) design: the ingredients to efficiently analyze two-color microarrays with Confirmatory Factor Analysis. BMC Genomics, 2006, 7, 232.	2.8	17
137	A DGGE system for comprehensive mutation screening ofBRCA1andBRCA2: application in a Dutch cancer clinic setting. Human Mutation, 2006, 27, 654-666.	2.5	75
138	RET as a Diagnostic and Therapeutic Target in Sporadic and Hereditary Endocrine Tumors. Endocrine Reviews, 2006, 27, 535-560.	20.1	311
139	Plakophilin-2 Mutations Are the Major Determinant of Familial Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. Circulation, 2006, 113, 1650-1658.	1.6	326
140	BRAF-V600E is not involved in the colorectal tumorigenesis of HNPCC in patients with functional MLH1 and MSH2 genes. Oncogene, 2005, 24, 3995-3998.	5.9	155
141	Concomitant RASSF1A hypermethylation and KRAS/BRAF mutations occur preferentially in MSI sporadic colorectal cancer. Oncogene, 2005, 24, 7630-7634.	5.9	45
142	A substantial proportion of microsatellite-unstable colon tumors carryTP53 mutations while not showing chromosomal instability. Genes Chromosomes and Cancer, 2005, 43, 194-201.	2.8	5
143	Colorectal cancer and theCHEK2 1100delC mutation. Genes Chromosomes and Cancer, 2005, 43, 377-382.	2.8	27
144	The Human Leukocyte Antigen Region and Colorectal Cancer Risk. Diseases of the Colon and Rectum, 2005, 48, 303-306.	1.3	7

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145	RET-Familial Medullary Thyroid Carcinoma Mutants Y791F and S891A Activate a Src/JAK/STAT3 Pathway, Independent of Glial Cell Lineâ€“Derived Neurotrophic Factor. Cancer Research, 2005, 65, 1729-1737.	0.9	84
146	Determination of TP53 Mutation Is More Relevant Than Microsatellite Instability Status for the Prediction of Disease-Free Survival in Adjuvant-Treated Stage III Colon Cancer Patients. Journal of Clinical Oncology, 2005, 23, 5635-5643.	1.6	127
147	Predictive value of thymidylate synthase and dihydropyrimidine dehydrogenase protein expression on survival in adjuvantly treated stage III colon cancer patients. Annals of Oncology, 2005, 16, 1646-1653.	1.2	43
148	Medullary Thyroid Cancer in a Patient with Hirschsprung Disease with a C609Y Germline RET-mutation. Journal of Pediatric Gastroenterology and Nutrition, 2005, 40, 226-229.	1.8	3
149	Identifying Candidate Hirschsprung Diseaseâ€“Associated RET Variants. American Journal of Human Genetics, 2005, 76, 850-858.	6.2	49
150	Homozygous Nonsense Mutations in KIAA1279 Are Associated with Malformations of the Central and Enteric Nervous Systems. American Journal of Human Genetics, 2005, 77, 120-126.	6.2	138
151	Distinct patterns of KRAS mutations in colorectal carcinomas according to germline mismatch repair defects and hMLH1 methylation status. Human Molecular Genetics, 2004, 13, 2303-2311.	2.9	127
152	Studying the genetics of Hirschsprung's disease: unraveling an oligogenic disorder. Clinical Genetics, 2004, 67, 6-14.	2.0	93
153	Localizing a putative mutation as the major contributor to the development of sporadic Hirschsprung disease to the RET genomic sequence between the promoter region and exon 2. European Journal of Human Genetics, 2004, 12, 604-612.	2.8	47
154	Distinct Transcriptional Changes in Donor Kidneys upon Brain Death Induction in Rats: Insights in the Processes of Brain Death. American Journal of Transplantation, 2004, 4, 1972-1981.	4.7	72
155	No association between two MLH3 variants (S845G and P844L)and colorectal cancer risk. Cancer Genetics and Cytogenetics, 2004, 152, 70-71.	1.0	15
156	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. Human Mutation, 2004, 23, 57-66.	2.5	50
157	No association between the Arg201Gly polymorphism of the DCC gene and colorectal cancer. Digestive and Liver Disease, 2004, 36, 821-823.	0.9	0
158	Two mismatch repair gene mutations found in a colon cancer patient â€“ which one is pathogenic?. Human Genetics, 2003, 112, 105-109.	3.8	30
159	MBD4 mutations are rare in gastric carcinomas with microsatellite instability. Cancer Genetics and Cytogenetics, 2003, 145, 103-107.	1.0	16
160	Detection of point mutation in dystrophin gene reveals somatic and germline mosaicism in the mother of a patient with Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 2003, 118A, 296-298.	2.4	29
161	Prognostic factors in ovarian cancer: current evidence and future prospects. European Journal of Cancer, Supplement, 2003, 1, 127-145.	2.2	18
162	Acceptable age for prophylactic surgery in children with multiple endocrine neoplasia type 2a. European Journal of Surgical Oncology, 2003, 29, 331-335.	1.0	33

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163	Toward New Strategies to Select Young Endometrial Cancer Patients for Mismatch Repair Gene Mutation Analysis. Journal of Clinical Oncology, 2003, 21, 4364-4370.	1.6	120
164	Molecular and Clinical Characteristics of MSH6 Variants: An Analysis of 25 Index Carriers of a Germline Variant. American Journal of Human Genetics, 2002, 70, 26-37.	6.2	271
165	A Rare Haplotype of the RET Proto-Oncogene Is a Risk-Modifying Allele in Hirschsprung Disease. American Journal of Human Genetics, 2002, 71, 969-974.	6.2	41
166	ABCD syndrome is caused by a homozygous mutation in theEDNRB gene. American Journal of Medical Genetics Part A, 2002, 108, 223-225.	2.4	44
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