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	A mutation in the RET proto-oncogene associated with multiple endocrine neoplasia type 2B and sporadic medullary thyroid carcinoma. Nature, 1994, 367, 375-376.	27.8	1,134
2	Evidence Based Selection of Housekeeping Genes. PLoS ONE, 2007, 2, e898.	2.5	617
3	Familial endometrial cancer in female carriers of MSH6 germline mutations. Nature Genetics, 1999, 23, 142-144.	21.4	378
4	C. elegans Model Identifies Genetic Modifiers of Î±-Synuclein Inclusion Formation During Aging. PLoS Genetics, 2008, 4, e1000027.	3.5	370
5	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
6	Plakophilin-2 Mutations Are the Major Determinant of Familial Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. Circulation, 2006, 113, 1650-1658.	1.6	326
7	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
8	RET as a Diagnostic and Therapeutic Target in Sporadic and Hereditary Endocrine Tumors. Endocrine Reviews, 2006, 27, 535-560.	20.1	311
9	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
10	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	21.4	269
11	Genetics, Clinical Features, and Long-TermÂOutcome of NoncompactionÂCardiomyopathy. Journal of the American College of Cardiology, 2018, 71, 711-722.	2.8	242
12	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
13	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
14	Mutation update on the CHD7 gene involved in CHARGE syndrome. Human Mutation, 2012, 33, 1149-1160.	2.5	224
15	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
16	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
17	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
18	TBX4mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension. Journal of Medical Genetics, 2013, 50, 500-506.	3.2	171

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19	Survival-Related Profile, Pathways, and Transcription Factors in Ovarian Cancer. PLoS Medicine, 2009, 6, e1000024.	8.4	156
20	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
21	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
22	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
23	Building a brain in the gut: development ofÂthe enteric nervous system. Clinical Genetics, 2013, 83, 307-316.	2.0	141
24	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
25	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
26	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
27	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
28	Identification of MOAG-4/SERF as a Regulator of Age-Related Proteotoxicity. Cell, 2010, 142, 601-612.	28.9	120
29	Contribution of rare and common variants determine complex diseasesâ€"Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119
30	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
31	White paper on guidelines concerning enteric nervous system stem cell therapy for enteric neuropathies. Developmental Biology, 2016, 417, 229-251.	2.0	112
32	The Role of Maternal-Fetal Cholesterol Transport in Early Fetal Life: Current Insights1. Biology of Reproduction, 2013, 88, 24.	2.7	108
33	A genetic variants database for arrhythmogenic right ventricular dysplasia/cardiomyopathy. Human Mutation, 2009, 30, 1278-1283.	2.5	105
34	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
35	Current concepts in RET-related genetics, signaling and therapeutics. Trends in Genetics, 2006, 22, 627-636.	6.7	101
36	MSH2 andMLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. , 1997, 18, 269-278.		99

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37	Candidate driver genes in microsatelliteâ€unstable colorectal cancer. International Journal of Cancer, 2012, 130, 1558-1566.	5.1	99
38	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
39	Studying the genetics of Hirschsprung's disease: unraveling an oligogenic disorder. Clinical Genetics, 2004, 67, 6-14.	2.0	93
40	Assessment of functional effects of unclassified genetic variants. Human Mutation, 2008, 29, 1314-1326.	2.5	93
41	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
42	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
43	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
44	MEN2A-RET-induced cellular transformation by activation of STAT3. Oncogene, 2001, 20, 5350-5358.	5.9	82
45	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
46	A Novel Point Mutation in the Intracellular Domain of theretProtooncogene in a Family with Medullary Thyroid Carcinoma1. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 4176-4178.	3.6	80
47	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
48	MEIS and PBX homeobox proteins in ovarian cancer. European Journal of Cancer, 2007, 43, 2495-2505.	2.8	79
49	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
50	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
51	Inclusion of malignant fibrous histiocytoma in the tumour spectrum associated with hereditary non-polyposis colorectal cancer. Genes Chromosomes and Cancer, 2000, 29, 353-355.	2.8	75
52	A DGGE system for comprehensive mutation screening ofBRCA1andBRCA2: application in a Dutch cancer clinic setting. Human Mutation, 2006, 27, 654-666.	2.5	75
53	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
54	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

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55	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
56	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
57	Biallelic Truncating Mutations in ALPK3 Cause Severe Pediatric Cardiomyopathy. Journal of the American College of Cardiology, 2016, 67, 515-525.	2.8	70
58	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
59	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
60	Cardiac Phenotypes, Genetics, and RisksÂin Familial Noncompaction Cardiomyopathy. Journal of the American College of Cardiology, 2019, 73, 1601-1611.	2.8	65
61	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
62	Pathological assessment of mismatch repair gene variants in Lynch syndrome: Past, present, and future. Human Mutation, 2012, 33, 1617-1625.	2.5	60
63	Paroxysmal exercise-induced dystonia within the phenotypic spectrum ofECHS1deficiency. Movement Disorders, 2016, 31, 1041-1048.	3.9	58
64	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	2.5	57
65	ACTG2variants impair actin polymerization in sporadic Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. Human Molecular Genetics, 2016, 25, 571-583.	2.9	56
66	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. Human Mutation, 2008, 29, 1292-1303.	2.5	54
67	An updated and upgradedL1CAMmutation database. Human Mutation, 2010, 31, E1102-E1109.	2.5	54
68	The Cardiac Phenotype in Patients With a CHD7 Mutation. Circulation: Cardiovascular Genetics, 2013, 6, 248-254.	5.1	53
69	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
70	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
71	A database to support the interpretation of human mismatch repair gene variants. Human Mutation, 2008, 29, 1337-1341.	2.5	51
72	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

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73	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
74	Identifying Candidate Hirschsprung Diseaseâ€"Associated RET Variants. American Journal of Human Genetics, 2005, 76, 850-858.	6.2	49
75	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
76	Review: Clinical aspects of hereditary DNA Mismatch repair gene mutations. DNA Repair, 2016, 38, 155-162.	2.8	49
77	Hydrocephalus and intestinal aganglionosis: IsL1CAMa modifier gene in Hirschsprung disease?. American Journal of Medical Genetics Part A, 2002, 108, 51-56.	2.4	47
78	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
79	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
80	A single-nucleotide polymorphic variant of the RET proto-oncogene is underrepresented in sporadic Hirschsprung disease. European Journal of Human Genetics, 2000, 8, 721-724.	2.8	45
81	Concomitant RASSF1A hypermethylation and KRAS/BRAF mutations occur preferentially in MSI sporadic colorectal cancer. Oncogene, 2005, 24, 7630-7634.	5.9	45
82	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
83	Genetics of enteric neuropathies. Developmental Biology, 2016, 417, 198-208.	2.0	44
84	MSI-L Gastric Carcinomas Share the hMLH1 Methylation Status of MSI-H Carcinomas but Not Their Clinicopathological Profile. Laboratory Investigation, 2000, 80, 1915-1923.	3.7	43
85	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
86	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
87	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
88	RET/PTC rearrangement is prevalent in follicular HÃ¼rthle cell carcinomas. Histopathology, 2012, 61, 833-843.	2.9	42
89	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
90	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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91	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
92	High Frequency ofRPL22Mutations in Microsatellite-Unstable Colorectal and Endometrial Tumors. Human Mutation, 2014, 35, 1442-1445.	2.5	38
93	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	2.9	38
94	KBP interacts with SCG10, linking Goldbergâ€“Shprintzen syndrome to microtubule dynamics and neuronal differentiation. Human Molecular Genetics, 2010, 19, 3642-3651.	2.9	37
95	Mutations of UFD1L Are Not Responsible for the Majority of Cases of DiGeorge Syndrome/Velocardiofacial Syndrome without Deletions within Chromosome 22q11. American Journal of Human Genetics, 1999, 65, 247-249.	6.2	36
96	What monozygotic twins discordant for phenotype illustrate about mechanisms influencing genetic forms of neurodegeneration. Clinical Genetics, 2012, 81, 325-333.	2.0	36
97	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
98	PMS2 involvement in patients suspected of Lynch syndrome. Genes Chromosomes and Cancer, 2009, 48, 322-329.	2.8	34
99	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
100	Recurrent and founder mutations in the Netherlands. Netherlands Heart Journal, 2010, 18, 583-591.	0.8	33
101	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
102	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
103	MUTYH and the mismatch repair system: partners in crime?. Human Genetics, 2006, 119, 206-211.	3.8	32
104	Congenital short bowel syndrome as the presenting symptom in male patients with FLNA mutations. Genetics in Medicine, 2013, 15, 310-313.	2.4	32
105	Regulators of gene expression in Enteric Neural Crest Cells are putative Hirschsprung disease genes. Developmental Biology, 2016, 416, 255-265.	2.0	31
106	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
107	Nuclear localization of human DNA mismatch repair protein exonuclease 1 (hEXO1). Nucleic Acids Research, 2007, 35, 2609-2619.	14.5	30
108	Two cases of the caudal duplication anomaly including a discordant monozygotic twin. American Journal of Medical Genetics Part A, 2002, 112, 390-393.	2.4	29

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109	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
110	Comprehensive TP53-Denaturing Gradient Gel Electrophoresis Mutation Detection Assay Also Applicable to Archival Paraffin-Embedded Tissue. Diagnostic Molecular Pathology, 1999, 8, 2-10.	2.1	28
111	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
112	Mutation analysis of the RET proto-oncogene in Dutch families with MEN 2A, MEN 2B and FMTC: two novel mutations and one de novo mutation for MEN 2A. Human Genetics, 1996, 97, 11-14.	3.8	27
113	Colorectal cancer and theCHEK2 1100delC mutation. Genes Chromosomes and Cancer, 2005, 43, 377-382.	2.8	27
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	Natural Gene Therapy in Dystrophic Epidermolysis Bullosa. Archives of Dermatology, 2012, 148, 213.	1.4	27
116	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
117	Zebrafish: A Model Organism for Studying Enteric Nervous System Development and Disease. Frontiers in Cell and Developmental Biology, 2020, 8, 629073.	3.7	27
118	No mutations found byRET mutation scanning in sporadic and hereditary neuroblastoma. Human Genetics, 1996, 97, 362-364.	3.8	26
119	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
120	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
121	Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours. Human Molecular Genetics, 2010, 19, 697-706.	2.9	26
122	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
123	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
124	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
125	Coexistent Hirschsprung's disease and esophageal achalasia in male siblings. Journal of Pediatric Surgery, 1997, 32, 1809-1811.	1.6	24
126	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Scientific Reports, 2015, 5, 16286.	3.3	24

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127	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	8.2	24
128	Constipation as the Presenting Symptom in De Novo Multiple Endocrine Neoplasia Type 2B. Pediatrics, 1998, 102, 405-407.	2.1	22
129	Mononucleotide precedes dinucleotide repeat instability during colorectal tumour development in Lynch syndrome patients. Journal of Pathology, 2009, 219, 96-102.	4.5	22
130	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
131	ENDOCRINE TUMOURS: Progressive metastatic medullary thyroid carcinoma: first- and second-line strategies. European Journal of Endocrinology, 2015, 172, R241-R251.	3.7	22
132	Brush border myosin Ia inactivation in gastric but not endometrial tumors. International Journal of Cancer, 2013, 132, 1790-1799.	5.1	21
133	Prognostic factors in ovarian cancer: current evidence and future prospects. European Journal of Cancer, Supplement, 2003, 1, 127-145.	2.2	18
134	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
135	A "late-but-fitter revertant cell" explains the high frequency of revertant mosaicism in epidermolysis bullosa. PLoS ONE, 2018, 13, e0192994.	2.5	18
136	Suspected Lynch syndrome associated MSH6 variants: A functional assay to determine their pathogenicity. PLoS Genetics, 2017, 13, e1006765.	3.5	18
137	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
138	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
139	Investigation of the genes for RET and its ligand complex, GDNF/GFRÎ±-1, in small cell lung carcinoma. , 1998, 21, 326-332.		16
140	MBD4 mutations are rare in gastric carcinomas with microsatellite instability. Cancer Genetics and Cytogenetics, 2003, 145, 103-107.	1.0	16
141	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
142	No association between two MLH3 variants (S845G and P844L)and colorectal cancer risk. Cancer Genetics and Cytogenetics, 2004, 152, 70-71.	1.0	15
143	Estrogens, MSI and Lynch syndrome-associated tumors. Biochimica Et Biophysica Acta: Reviews on Cancer, 2009, 1796, 194-200.	7.4	14
144	TP53 germline mutations in Portugal and genetic modifiers of age at cancer onset. Familial Cancer, 2009, 8, 383-390.	1.9	14

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145	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
146	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
147	Loss of enteric neuronal Ndrg4 promotes colorectal cancer via increased release of Nid1 and Fbln2. EMBO Reports, 2021, 22, e51913.	4.5	14
148	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
149	Using Out-of-Batch Reference Populations to Improve Untargeted Metabolomics for Screening Inborn Errors of Metabolism. Metabolites, 2021, 11, 8.	2.9	14
150	Intestinal multicellular organoids to study colorectal cancer. Biochimica Et Biophysica Acta: Reviews on Cancer, 2021, 1876, 188586.	7.4	13
151	Getting rid of thePMS2 pseudogenes: mission impossible?. Human Mutation, 2007, 28, 414-414.	2.5	12
152	A New Perspective on Transcriptional System Regulation (TSR): Towards TSR Profiling. PLoS ONE, 2008, 3, e1656.	2.5	11
153	Perspectives for tailored chemoprevention and treatment of colorectal cancer in Lynch syndrome. Critical Reviews in Oncology/Hematology, 2011, 80, 264-277.	4.4	11
154	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
155	Do microsatellite instability profiles really differ between colorectal and endometrial tumors?. Genes Chromosomes and Cancer, 2009, 48, 552-557.	2.8	10
156	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
157	Re: Role of the Oxidative DNA Damage Repair Gene OGG1 in Colorectal Tumorigenesis. Journal of the National Cancer Institute, 2014, 106, .	6.3	9
158	Routine Molecular Analysis for Lynch Syndrome Among Adenomas or Colorectal Cancer Within a National Screening Program. Gastroenterology, 2018, 155, 1410-1415.	1.3	9
159	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
160	The Human Leukocyte Antigen Region and Colorectal Cancer Risk. Diseases of the Colon and Rectum, 2005, 48, 303-306.	1.3	7
161	Correspondence: SEMA4A variation and risk of colorectal cancer. Nature Communications, 2016, 7, 10611.	12.8	7
162	Do RET somatic mutations play a role in Hirschsprung disease?. Genetics in Medicine, 2018, 20, 1477-1478.	2.4	7

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163	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
164	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
165	Mutations in SCG10 Are Not Involved in Hirschsprung Disease. PLoS ONE, 2010, 5, e15144.	2.5	6
166	Haplotype sharing test maps genes for familial cardiomyopathiesâ€. Clinical Genetics, 2011, 79, 459-467.	2.0	6
167	Hirschsprung Disease and Activation of Hedgehog Signaling via GLI1-3 Mutations. Gastroenterology, 2015, 149, 1672-1675.	1.3	6
168	Goldbergâ€“Shprintzen syndrome is determined by the absence, or reduced expression levels, of KIFBP. Human Mutation, 2020, 41, 1906-1917.	2.5	6
169	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
170	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
171	Screening for germline DND1 mutations in testicular cancer patients. Familial Cancer, 2010, 9, 439-442.	1.9	5
172	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
173	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
174	A novel MSH2 germline mutation in a Druze HNPCC family. Familial Cancer, 2008, 7, 135-139.	1.9	4
175	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1603-1610.	2.8	4
176	Germline genome editing: public dialogue is urgent but not self-evident. European Journal of Human Genetics, 2020, 28, 4-5.	2.8	4
177	MSH2 and MLH1 mutations in sporadic replication errorâ€positive colorectal carcinoma as assessed by twoâ€dimensional DNA electrophoresis. Genes Chromosomes and Cancer, 1997, 18, 269-278.	2.8	4
178	TFAP2B Haploinsufficiency Impacts Gastrointestinal Function and Leads to Pediatric Intestinal Pseudo-obstruction. Frontiers in Cell and Developmental Biology, 0, 10, .	3.7	4
179	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
180	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

#	Article	IF	Citations
181	The Somatic Mutation Paradigm in Congenital Malformations: Hirschsprung Disease as a Model. International Journal of Molecular Sciences, 2021, 22, 12354.	4.1	3
182	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
183	No mutations found by RET mutation scanning in sporadic and hereditary neuroblastoma. Human Genetics, 1996, 97, 362-364.	3.8	3
184	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
185	Infantile hypertrophic pyloric stenosis in patients with esophageal atresia. Birth Defects Research, 2020, 112, 670-687.	1.5	1
186	Ordering of markers in the pericentromeric region of chromosome 10. Human Genetics, 1995, 96, 116-118.	3.8	0
187	No association between the Arg201Gly polymorphism of the DCC gene and colorectal cancer. Digestive and Liver Disease, 2004, 36, 821-823.	0.9	0
188	Cell-free assay breakthrough forMLH1variants. Human Mutation, 2010, 31, v-v.	2.5	0