Robert Hofstra

List of Publications by Citations

185 papers	10,925 citations	55 h-index	99 g-index
193 ext. papers	12,295 ext. citations	7.1 avg, IF	5.44 L-index

#	Paper	IF	Citations
185	A mutation in the RET proto-oncogene associated with multiple endocrine neoplasia type 2B and sporadic medullary thyroid carcinoma. *Nature*, **1994**, 367, 375-6	50.4	975
184	Evidence based selection of housekeeping genes. *PLoS ONE*, **2007**, 2, e898	3.7	527
183	Familial endometrial cancer in female carriers of MSH6 germline mutations. *Nature Genetics*, **1999**, 23, 142-4	36.3	342
182	C. elegans model identifies genetic modifiers of alpha-synuclein inclusion formation during aging. *PLoS Genetics*, **2008**, 4, e1000027	6	295
181	Plakophilin-2 mutations are the major determinant of familial arrhythmogenic right ventricular dysplasia/cardiomyopathy. *Circulation*, **2006**, 113, 1650-8	16.7	276
180	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, 1199-207	12.3	270
179	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	6	261
178	RET as a diagnostic and therapeutic target in sporadic and hereditary endocrine tumors. *Endocrine Reviews*, **2006**, 27, 535-60	27.2	261
177	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	11	246
176	Segregation at three loci explains familial and population risk in Hirschsprung disease. *Nature Genetics*, **2002**, 31, 89-93	36.3	223
175	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	11	198
174	Mutations in a TGF-□ligand, TGFB3, cause syndromic aortic aneurysms and dissections. *Journal of the American College of Cardiology*, **2015**, 65, 1324-1336	15.1	184
173	Histone methyltransferase gene SETD2 is a novel tumor suppressor gene in clear cell renal cell carcinoma. *Cancer Research*, **2010**, 70, 4287-91	10.1	178
172	Mutation update on the CHD7 gene involved in CHARGE syndrome. *Human Mutation*, **2012**, 33, 1149-60	4.7	170
171	Germline hypermethylation of MLH1 and EPCAM deletions are a frequent cause of Lynch syndrome. *Genes Chromosomes and Cancer*, **2009**, 48, 737-44	5	160
170	Genetics, Clinical Features, and Long-Term□Outcome of Noncompaction□Cardiomyopathy. *Journal of the American College of Cardiology*, **2018**, 71, 711-722	15.1	158
169	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	36.3	158

168	Survival-related profile, pathways, and transcription factors in ovarian cancer. *PLoS Medicine*, **2009**, 6, e24	11.6	133
167	Homozygous nonsense mutations in KIAA1279 are associated with malformations of the central and enteric nervous systems. *American Journal of Human Genetics*, **2005**, 77, 120-6	11	129
166	BRAF-V600E is not involved in the colorectal tumorigenesis of HNPCC in patients with functional MLH1 and MSH2 genes. *Oncogene*, **2005**, 24, 3995-8	9.2	128
165	High yield of LMNA mutations in patients with dilated cardiomyopathy and/or conduction disease referred to cardiogenetics outpatient clinics. *American Heart Journal*, **2007**, 154, 1130-9	4.9	124
164	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, 1574-83	6.7	117
163	TBX4 mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension. *Journal of Medical Genetics*, **2013**, 50, 500-6	5.8	115
162	Building a brain in the gut: development of the enteric nervous system. *Clinical Genetics*, **2013**, 83, 307-16	4	113
161	Toward new strategies to select young endometrial cancer patients for mismatch repair gene mutation analysis. *Journal of Clinical Oncology*, **2003**, 21, 4364-70	2.2	112
160	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, 5635-43	2.2	111
159	Distinct patterns of KRAS mutations in colorectal carcinomas according to germline mismatch repair defects and hMLH1 methylation status. *Human Molecular Genetics*, **2004**, 13, 2303-11	5.6	102
158	A genetic variants database for arrhythmogenic right ventricular dysplasia/cardiomyopathy. *Human Mutation*, **2009**, 30, 1278-83	4.7	94
157	Current concepts in RET-related genetics, signaling and therapeutics. *Trends in Genetics*, **2006**, 22, 627-36	8.5	91
156	Contribution of rare and common variants determine complex diseases-Hirschsprung disease as a model. *Developmental Biology*, **2013**, 382, 320-9	3.1	90
155	Assessment of functional effects of unclassified genetic variants. *Human Mutation*, **2008**, 29, 1314-26	4.7	85
154	The role of maternal-fetal cholesterol transport in early fetal life: current insights. *Biology of Reproduction*, **2013**, 88, 24	3.9	83
153	Identification of MOAG-4/SERF as a regulator of age-related proteotoxicity. *Cell*, **2010**, 142, 601-12	56.2	83
152	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, 581-96	11	82
151	RET-familial medullary thyroid carcinoma mutants Y791F and S891A activate a Src/JAK/STAT3 pathway, independent of glial cell line-derived neurotrophic factor. *Cancer Research*, **2005**, 65, 1729-37	10.1	79

150	Candidate driver genes in microsatellite-unstable colorectal cancer. *International Journal of Cancer*, **2012**, 130, 1558-66	7.5	78
149	Studying the genetics of Hirschsprung disease: unraveling an oligogenic disorder. *Clinical Genetics*, **2005**, 67, 6-14	4	77
148	White paper on guidelines concerning enteric nervous system stem cell therapy for enteric neuropathies. *Developmental Biology*, **2016**, 417, 229-51	3.1	77
147	MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis **1997**, 18, 269-278		76
146	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, 29230-40	5.4	76
145	MEN2A-RET-induced cellular transformation by activation of STAT3. *Oncogene*, **2001**, 20, 5350-8	9.2	76
144	Inclusion of malignant fibrous histiocytoma in the tumour spectrum associated with hereditary non-polyposis colorectal cancer. *Genes Chromosomes and Cancer*, **2000**, 29, 353-5	5	71
143	Calibration of multiple in silico tools for predicting pathogenicity of mismatch repair gene missense substitutions. *Human Mutation*, **2013**, 34, 255-65	4.7	70
142	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, 1972-81	8.7	69
141	A novel point mutation in the intracellular domain of the ret protooncogene in a family with medullary thyroid carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **1997**, 82, 4176-8	5.6	68
140	A DGGE system for comprehensive mutation screening of BRCA1 and BRCA2: application in a Dutch cancer clinic setting. *Human Mutation*, **2006**, 27, 654-66	4.7	68
139	Clinical relevance of 18F-FDG PET and 18F-DOPA PET in recurrent medullary thyroid carcinoma. *Journal of Nuclear Medicine*, **2012**, 53, 1863-71	8.9	67
138	Severe myocardial fibrosis caused by a deletion of the 5 end of the lamin A/C gene. *Journal of the American College of Cardiology*, **2007**, 49, 2430-9	15.1	67
137	The effects of four different tyrosine kinase inhibitors on medullary and papillary thyroid cancer cells. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E991-5	5.6	66
136	MEIS and PBX homeobox proteins in ovarian cancer. *European Journal of Cancer*, **2007**, 43, 2495-505	7.5	66
135	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, 914-23	8.1	65
134	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. *Human Mutation*, **2011**, 32, 1100-7	4.7	63
133	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	11.5	62

132	Genotype-phenotype correlations in L1 syndrome: a guide for genetic counselling and mutation analysis. *Journal of Medical Genetics*, **2010**, 47, 169-75	5.8	61
131	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, 1059-62	4.7	57
130	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. *Genome Biology*, **2017**, 18, 48	18.3	55
129	Paediatric intestinal cancer and polyposis due to bi-allelic PMS2 mutations: case series, review and follow-up guidelines. *European Journal of Cancer*, **2011**, 47, 965-82	7.5	54
128	Pathological assessment of mismatch repair gene variants in Lynch syndrome: past, present, and future. *Human Mutation*, **2012**, 33, 1617-25	4.7	52
127	A novel classification system to predict the pathogenic effects of CHD7 missense variants in CHARGE syndrome. *Human Mutation*, **2012**, 33, 1251-60	4.7	52
126	Tumor characteristics as an analytic tool for classifying genetic variants of uncertain clinical significance. *Human Mutation*, **2008**, 29, 1292-303	4.7	51
125	Interaction between a chromosome 10 RET enhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. *Human Mutation*, **2009**, 30, 771-5	4.7	50
124	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, 6415-24	5.4	48
123	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. *American Journal of Human Genetics*, **2017**, 101, 123-129	11	47
122	A database to support the interpretation of human mismatch repair gene variants. *Human Mutation* , **2008**, 29, 1337-41	4.7	47
121	Functional analysis helps to clarify the clinical importance of unclassified variants in DNA mismatch repair genes. *Human Mutation*, **2007**, 28, 1047-54	4.7	46
120	Identifying candidate Hirschsprung disease-associated RET variants. *American Journal of Human Genetics*, **2005**, 76, 850-8	11	45
119	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. *Human Mutation*, **2004**, 23, 57-66	4.7	45
118	Review: Clinical aspects of hereditary DNA Mismatch repair gene mutations. *DNA Repair*, **2016**, 38, 155-163	4.3	43
117	CLMP is required for intestinal development, and loss-of-function mutations cause congenital short-bowel syndrome. *Gastroenterology*, **2012**, 142, 453-462.e3	13.3	43
116	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, 604-12	5.3	42
115	Concomitant RASSF1A hypermethylation and KRAS/BRAF mutations occur preferentially in MSI sporadic colorectal cancer. *Oncogene*, **2005**, 24, 7630-4	9.2	42

114	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, 1646-53	10.3	42
113	A single-nucleotide polymorphic variant of the RET proto-oncogene is underrepresented in sporadic Hirschsprung disease. *European Journal of Human Genetics*, **2000**, 8, 721-4	5.3	42
112	Cardiac Phenotypes, Genetics, and Risks□n Familial Noncompaction Cardiomyopathy. *Journal of the American College of Cardiology*, **2019**, 73, 1601-1611	15.1	41
111	Paroxysmal exercise-induced dystonia within the phenotypic spectrum of ECHS1 deficiency. *Movement Disorders*, **2016**, 31, 1041-8	7	41
110	Biallelic Truncating Mutations in ALPK3 Cause Severe Pediatric Cardiomyopathy. *Journal of the American College of Cardiology*, **2016**, 67, 515-25	15.1	41
109	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, 17292-302	5.4	41
108	An updated and upgraded L1CAM mutation database. *Human Mutation*, **2010**, 31, E1102-9	4.7	41
107	MSI-L gastric carcinomas share the hMLH1 methylation status of MSI-H carcinomas but not their clinicopathological profile. *Laboratory Investigation*, **2000**, 80, 1915-23	5.9	41
106	ACTG2 variants impair actin polymerization in sporadic Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. *Human Molecular Genetics*, **2016**, 25, 571-83	5.6	40
105	The cardiac phenotype in patients with a CHD7 mutation. *Circulation: Cardiovascular Genetics*, **2013**, 6, 248-54		39
104	Locus-specific databases and recommendations to strengthen their contribution to the classification of variants in cancer susceptibility genes. *Human Mutation*, **2008**, 29, 1273-81	4.7	39
103	ABCD syndrome is caused by a homozygous mutation in the EDNRB gene. *American Journal of Medical Genetics Part A*, **2002**, 108, 223-5		39
102	A rare haplotype of the RET proto-oncogene is a risk-modifying allele in hirschsprung disease. *American Journal of Human Genetics*, **2002**, 71, 969-74	11	39
101	RET/PTC rearrangement is prevalent in follicular H□thle cell carcinomas. *Histopathology*, **2012**, 61, 833-43	7.3	37
100	A rapid and cell-free assay to test the activity of lynch syndrome-associated MSH2 and MSH6 missense variants. *Human Mutation*, **2012**, 33, 488-94	4.7	37
99	Combined adverse effects of maternal smoking and high body mass index on heart development in offspring: evidence for interaction?. *Heart*, **2012**, 98, 474-9	5.1	37
98	Hydrocephalus and intestinal aganglionosis: is L1CAM a modifier gene in Hirschsprung disease?. *American Journal of Medical Genetics Part A*, **2002**, 108, 51-6		37
97	Variants in RET associated with Hirschsprung□ disease affect binding of transcription factors and gene expression. *Gastroenterology*, **2011**, 140, 572-582.e2	13.3	35

96	High frequency of RPL22 mutations in microsatellite-unstable colorectal and endometrial tumors. *Human Mutation*, **2014**, 35, 1442-5	4.7	34
95	KBP interacts with SCG10, linking Goldberg-Shprintzen syndrome to microtubule dynamics and neuronal differentiation. *Human Molecular Genetics*, **2010**, 19, 3642-51	5.6	33
94	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.e19-25	6.4	32
93	PMS2 involvement in patients suspected of Lynch syndrome. *Genes Chromosomes and Cancer*, **2009**, 48, 322-9	5	31
92	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, 247-9	11	30
91	Recurrent and founder mutations in the Netherlands : Plakophilin-2 p.Arg79X mutation causing arrhythmogenic right ventricular cardiomyopathy/dysplasia. *Netherlands Heart Journal*, **2010**, 18, 583-91	2.2	29
90	Genetics of enteric neuropathies. *Developmental Biology*, **2016**, 417, 198-208	3.1	29
89	Two mismatch repair gene mutations found in a colon cancer patient--which one is pathogenic?. *Human Genetics*, **2003**, 112, 105-9	6.3	28
88	Acceptable age for prophylactic surgery in children with multiple endocrine neoplasia type 2a. *European Journal of Surgical Oncology*, **2003**, 29, 331-5	3.6	28
87	Nuclear localization of human DNA mismatch repair protein exonuclease 1 (hEXO1). *Nucleic Acids Research*, **2007**, 35, 2609-19	20.1	27
86	MUTYH and the mismatch repair system: partners in crime?. *Human Genetics*, **2006**, 119, 206-11	6.3	27
85	What monozygotic twins discordant for phenotype illustrate about mechanisms influencing genetic forms of neurodegeneration. *Clinical Genetics*, **2012**, 81, 325-33	4	26
84	Congenital short bowel syndrome as the presenting symptom in male patients with FLNA mutations. *Genetics in Medicine*, **2013**, 15, 310-3	8.1	26
83	Novel no-stop FLNA mutation causes multi-organ involvement in males. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2376-84	2.5	26
82	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, 2352-61	6.9	25
81	Common arterial trunk and ventricular non-compaction in Lrp2 knockout mice indicate a crucial role of LRP2 in cardiac development. *DMM Disease Models and Mechanisms*, **2016**, 9, 413-25	4.1	25
80	The entire miR-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, 165-73	5	25
79	Two cases of the caudal duplication anomaly including a discordant monozygotic twin. *American Journal of Medical Genetics Part A*, **2002**, 112, 390-3		25

78	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, 296-8		25
77	Colorectal cancer and the CHEK2 1100delC mutation. *Genes Chromosomes and Cancer*, **2005**, 43, 377-82	5	25
76	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, 11-4	6.3	25
75	Comprehensive TP53-denaturing gradient gel electrophoresis mutation detection assay also applicable to archival paraffin-embedded tissue. *Diagnostic Molecular Pathology*, **1999**, 8, 2-10		24
74	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. *Human Molecular Genetics*, **2016**, 25, 5265-5275	5.6	23
73	Regulators of gene expression in Enteric Neural Crest Cells are putative Hirschsprung disease genes. *Developmental Biology*, **2016**, 416, 255-265	3.1	22
72	Natural gene therapy in dystrophic epidermolysis bullosa. *Archives of Dermatology*, **2012**, 148, 213-6		22
71	No mutations found by RET mutation scanning in sporadic and hereditary neuroblastoma. *Human Genetics*, **1996**, 97, 362-4	6.3	22
70	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, 4128-33	11.5	21
69	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. *Scientific Reports*, **2015**, 5, 16286	4.9	21
68	Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours. *Human Molecular Genetics*, **2010**, 19, 697-706	5.6	21
67	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, 160-7	5.8	21
66	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, 110-4		21
65	Constipation as the presenting symptom in de novo multiple endocrine neoplasia type 2B. *Pediatrics*, **1998**, 102, 405-8	7.4	21
64	Brush border myosin Ia inactivation in gastric but not endometrial tumors. *International Journal of Cancer*, **2013**, 132, 1790-9	7.5	20
63	Complex pathogenesis of Hirschsprung 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, 483-90	5.2	20
62	Long-term follow-up of patients with recessive dystrophic epidermolysis bullosa in the Netherlands: expansion of the mutation database and unusual phenotype-genotype correlations. *Journal of Dermatological Science*, **2009**, 56, 9-18	4.3	20
61	Coexistent Hirschsprung disease and esophageal achalasia in male siblings. *Journal of Pediatric Surgery*, **1997**, 32, 1809-11	2.6	20

60	Chromosome 21 scan in Down syndrome reveals DSCAM as a predisposing locus in Hirschsprung disease. *PLoS ONE*, **2013**, 8, e62519	3.7	18
59	Mononucleotide precedes dinucleotide repeat instability during colorectal tumour development in Lynch syndrome patients. *Journal of Pathology*, **2009**, 219, 96-102	9.4	18
58	Endocrine tumours: progressive metastatic medullary thyroid carcinoma: first- and second-line strategies. *European Journal of Endocrinology*, **2015**, 172, R241-51	6.5	17
57	Identification of Variants in RET and IHH Pathway Members in a Large Family With History of Hirschsprung Disease. *Gastroenterology*, **2018**, 155, 118-129.e6	13.3	17
56	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	4.5	16
55	Biochemical characterization of MLH3 missense mutations does not reveal an apparent role of MLH3 in Lynch syndrome. *Genes Chromosomes and Cancer*, **2009**, 48, 340-50	5	15
54	MBD4 mutations are rare in gastric carcinomas with microsatellite instability. *Cancer Genetics and Cytogenetics*, **2003**, 145, 103-7		15
53	A "late-but-fitter revertant cell" explains the high frequency of revertant mosaicism in epidermolysis bullosa. *PLoS ONE*, **2018**, 13, e0192994	3.7	14
52	No association between two MLH3 variants (S845G and P844L)and colorectal cancer risk. *Cancer Genetics and Cytogenetics*, **2004**, 152, 70-1		14
51	Prognostic factors in ovarian cancer: current evidence and future prospects. *European Journal of Cancer, Supplement*, **2003**, 1, 127-145	1.6	14
50	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	11	14
49	TP53 germline mutations in Portugal and genetic modifiers of age at cancer onset. *Familial Cancer*, **2009**, 8, 383-90	3	12
48	A new perspective on transcriptional system regulation (TSR): towards TSR profiling. *PLoS ONE*, **2008**, 3, e1656	3.7	11
47	Suspected Lynch syndrome associated MSH6 variants: A functional assay to determine their pathogenicity. *PLoS Genetics*, **2017**, 13, e1006765	6	11
46	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	3.7	11
45	Do microsatellite instability profiles really differ between colorectal and endometrial tumors?. *Genes Chromosomes and Cancer*, **2009**, 48, 552-7	5	10
44	Investigation of the genes for RET and its ligand complex, GDNF/GFR1, in small cell lung carcinoma **1998**, 21, 326-332		9
43	Getting rid of the PMS2 pseudogenes: mission impossible?. *Human Mutation*, **2007**, 28, 414; author reply 415	4.7	9

42	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	2.4	9
41	Re: Role of the oxidative DNA damage repair gene OGG1 in colorectal tumorigenesis. *Journal of the National Cancer Institute*, **2014**, 106,	9.7	8
40	New target genes in endometrial tumors show a role for the estrogen-receptor pathway in microsatellite-unstable cancers. *Human Mutation*, **2014**, 35, 1514-23	4.7	8
39	Perspectives for tailored chemoprevention and treatment of colorectal cancer in Lynch syndrome. *Critical Reviews in Oncology/Hematology*, **2011**, 80, 264-77	7	8
38	Yield of Lynch Syndrome Surveillance for Patients With Pathogenic Variants in DNA Mismatch Repair Genes. *Clinical Gastroenterology and Hepatology*, **2020**, 18, 1112-1120.e1	6.9	8
37	Zebrafish: A Model Organism for Studying Enteric Nervous System Development and Disease. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 629073	5.7	8
36	Estrogens, MSI and Lynch syndrome-associated tumors. *Biochimica Et Biophysica Acta: Reviews on Cancer*, **2009**, 1796, 194-200	11.2	7
35	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. *European Journal of Human Genetics*, **2012**, 20, 917-20	5.3	7
34	Do RET somatic mutations play a role in Hirschsprung disease?. *Genetics in Medicine*, **2018**, 20, 1477-1478	8.1	6
33	The human leukocyte antigen region and colorectal cancer risk. *Diseases of the Colon and Rectum*, **2005**, 48, 303-6	3.1	6
32	The MLH1 c.1852_1853delinsGC (p.K618A) variant in colorectal cancer: genetic association study in 18,723 individuals. *PLoS ONE*, **2014**, 9, e95022	3.7	6
31	Loss of enteric neuronal Ndrg4 promotes colorectal cancer via increased release of Nid1 and Fbln2. *EMBO Reports*, **2021**, 22, e51913	6.5	6
30	Correspondence: SEMA4A variation and risk of colorectal cancer. *Nature Communications*, **2016**, 7, 10611	17.4	6
29	Haplotype sharing test maps genes for familial cardiomyopathies. *Clinical Genetics*, **2011**, 79, 459-67	4	5
28	Mutations in SCG10 are not involved in Hirschsprung disease. *PLoS ONE*, **2010**, 5, e15144	3.7	5
27	A substantial proportion of microsatellite-unstable colon tumors carry TP53 mutations while not showing chromosomal instability. *Genes Chromosomes and Cancer*, **2005**, 43, 194-201	5	5
26	Inhibition of ROCK signaling pathway accelerates enteric neural crest cell-based therapy after transplantation in a rat hypoganglionic model. *Neurogastroenterology and Motility*, **2020**, 32, e13895	4	4
25	Goldberg-Shprintzen syndrome is determined by the absence, or reduced expression levels, of KIFBP. *Human Mutation*, **2020**, 41, 1906-1917	4.7	4

24	Routine Molecular Analysis for Lynch Syndrome Among Adenomas or Colorectal Cancer Within a National Screening Program. *Gastroenterology*, **2018**, 155, 1410-1415	13.3	4
23	MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis **1997**, 18, 269		4
22	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	3.7	3
21	Functional analyses of RET mutations in Chinese Hirschsprung disease patients. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2012**, 94, 47-51		3
20	Screening for germline DND1 mutations in testicular cancer patients. *Familial Cancer*, **2010**, 9, 439-42	3	3
19	Using Out-of-Batch Reference Populations to Improve Untargeted Metabolomics for Screening Inborn Errors of Metabolism. *Metabolites*, **2020**, 11,	5.6	3
18	Three-step site-directed mutagenesis screen identifies pathogenic variants associated with Lynch syndrome. *Journal of Medical Genetics*, **2020**, 57, 308-315	5.8	3
17	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. *Journal of Clinical Investigation*, **2021**, 131,	15.9	3
16	Biallelic Variants in , Encoding a Cytosolic Targeting Factor of Tail-Anchored Proteins, Cause Rapidly Progressive Pediatric Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2019**, 12, 397-406	5.2	2
15	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. *European Journal of Human Genetics*, **2018**, 26, 1603-1610	5.3	2
14	A novel MSH2 germline mutation in a Druze HNPCC family. *Familial Cancer*, **2008**, 7, 135-9	3	2
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