

Mutation analysis of the RET receptor tyrosine kinase in

Human Molecular Genetics

4, 821-830

DOI: [10.1093/hmg/4.5.821](https://doi.org/10.1093/hmg/4.5.821)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Intestinal neuronal dysplasia. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 1995, 426, 537-9.	1.4	37
2	GDNF-induced Activation of the Ret Protein Tyrosine Kinase Is Mediated by GDNFR- β , a Novel Receptor for GDNF. Cell, 1996, 85, 1113-1124.	13.5	1,128
3	RET mutations in human disease. Trends in Genetics, 1996, 12, 138-144.	2.9	175
4	RET oncogene. Current Opinion in Genetics and Development, 1996, 6, 82-86.	1.5	21
5	RET protooncogene mutational analysis in multiple endocrine neoplasia syndrome type 2B. Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics, 1996, 82, 288-294.	1.6	16
6	Unravelling the pathogenesis and molecular genetics of Hirschsprung's disease. Seminars in Fetal and Neonatal Medicine, 1996, 1, 211-217.	2.8	0
7	Novel mutations of the endothelin-B receptor gene in isolated patients with Hirschsprung's disease. Human Molecular Genetics, 1996, 5, 347-349.	1.4	110
8	Molecular heterogeneity of RET loss of function in Hirschsprung's disease.. EMBO Journal, 1996, 15, 2717-2725.	3.5	109
9	The Men II Syndromes and the Role of the ret Proto-oncogene. Advances in Cancer Research, 1996, 70, 179-222.	1.9	56
10	Congenital central hypoventilation syndrome: Mutation analysis of the receptor tyrosine kinase RET. , 1996, 63, 603-609.		45
11	Frequent loss of heterozygosity for markers on chromosome arm 10q in chondrosarcomas. , 1996, 16, 138-143.		28
12	A homozygous mutation in the endothelin-3 gene associated with a combined Waardenburg type 2 and Hirschsprung phenotype (Shah-Waardenburg syndrome). Nature Genetics, 1996, 12, 445-447.	9.4	296
13	Germline mutations in glial cell line-derived neurotrophic factor (GDNF) and RET in a Hirschsprung disease patient. Nature Genetics, 1996, 14, 341-344.	9.4	269
14	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	9.4	203
15	De novo mutation of GDNF, ligand for the RET/GDNFR-alpha receptor complex, in Hirschsprung disease. Human Molecular Genetics, 1996, 5, 2023-2026.	1.4	118
16	Endothelin-B receptor mutations in patients with isolated Hirschsprung disease from a non-inbred population. Human Molecular Genetics, 1996, 5, 351-354.	1.4	106
17	The RET Proto-Oncogene in Multiple Endocrine Neoplasia Type 2 and Hirschsprung's Disease. New England Journal of Medicine, 1996, 335, 943-951.	13.9	317
18	Mechanism of ret dysfunction by Hirschsprung mutations affecting its extracellular domain. Human Molecular Genetics, 1996, 5, 1577-1580.	1.4	87

#	ARTICLE	IF	CITATIONS
19	The Relationship Between Specific RET Proto-oncogene Mutations and Disease Phenotype in Multiple Endocrine Neoplasia Type 2. <i>JAMA - Journal of the American Medical Association</i> , 1996, 276, 1575.	3.8	690
20	Mutation Analysis of Glial Cell Line-Derived Neurotrophic Factor, a Ligand for an RET/Coreceptor Complex, in Multiple Endocrine Neoplasia Type 2 and Sporadic Neuroendocrine Tumors. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1997, 82, 3025-3028.	1.8	26
21	Genetic predisposition to pheochromocytoma: analysis of candidate genes GDNF, RET and VHL. <i>Human Molecular Genetics</i> , 1997, 6, 1051-1056.	1.4	102
22	Human Cancer Syndromes: Clues to the Origin and Nature of Cancer. <i>Science</i> , 1997, 278, 1043-1050.	6.0	528
23	Germline mutation of the RET proto-oncogene in children with total intestinal aganglionosis. <i>Journal of Pediatric Surgery</i> , 1997, 32, 498-500.	0.8	16
24	Mutation analysis of the RET, the endothelin-B receptor, and the endothelin-3 genes in sporadic cases of Hirschsprung's disease. <i>Journal of Pediatric Surgery</i> , 1997, 32, 501-504.	0.8	60
25	Hirschsprung's disease as a neurochristopathy. <i>Pediatric Surgery International</i> , 1997, 12, 2-10.	0.6	20
26	The RET proto-oncogene: A challenge to our understanding of disease pathogenesis. <i>Pediatric Surgery International</i> , 1997, 12, 11-18.	0.6	29
27	Does Phaster Mean Better?. <i>Clinical Chemistry</i> , 1997, 43, 424-426.	1.5	4
28	Glial cell line-derived neurotrophic factor-dependent RET activation can be mediated by two different cell-surface accessory proteins. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997, 94, 6238-6243.	3.3	285
29	Rapid, nonradioactive screening for mutations in exons 10, 11, and 16 of the RET protooncogene associated with inherited medullary thyroid carcinoma. <i>Clinical Chemistry</i> , 1997, 43, 453-457.	1.5	25
30	The RET proto-oncogene in medullary and papillary thyroid carcinoma. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 1997, 431, 1-9.	1.4	59
31	Ret in human development and oncogenesis. <i>BioEssays</i> , 1997, 19, 389-395.	1.2	74
32	Magnetic resonance imaging abnormalities of the brain in Goldberg-Shprintzen syndrome (Hirschsprung disease, microcephaly, and iris coloboma). <i>American Journal of Medical Genetics Part A</i> , 1997, 73, 230-232.	2.4	19
33	Mutations of the RET proto-oncogene in the multiple endocrine neoplasia type 2 syndromes, related sporadic tumours, and Hirschsprung disease. , 1997, 9, 97-109.		180
34	Frequency of RET mutations in long- and short-segment Hirschsprung disease. <i>Human Mutation</i> , 1997, 9, 243-249.	1.1	138
35	Dual effect on the RET receptor of MEN α 2 mutations affecting specific extracytoplasmic cysteines. <i>Oncogene</i> , 1998, 17, 2851-2861.	2.6	97
36	Genomic structure of the gene for the SH2 and pleckstrin homology domain-containing protein GRB10 and evaluation of its role in Hirschsprung disease. <i>Oncogene</i> , 1998, 17, 3065-3070.	2.6	27

#	ARTICLE	IF	CITATIONS
37	Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. <i>Journal of Internal Medicine</i> , 1998, 243, 515-520.	2.7	74
38	Ku70. <i>Molecular Cell</i> , 1998, 2, 1-8.	4.5	217
39	Occurrence of MEN 2a in familial hirschsprung's disease: A new indication for genetic testing of the RET proto-oncogene. <i>Journal of Pediatric Surgery</i> , 1998, 33, 207-214.	0.8	58
40	GDNF and ET-3 Differentially Modulate the Numbers of Avian Enteric Neural Crest Cells and Enteric Neurons in Vitro. <i>Developmental Biology</i> , 1998, 197, 93-105.	0.9	214
41	HumanGFRA1: Cloning, Mapping, Genomic Structure, and Evaluation as a Candidate Gene for Hirschsprung Disease Susceptibility. <i>Genomics</i> , 1998, 48, 354-362.	1.3	58
42	Novel Mutations of the Endothelin B Receptor Gene in Patients with Hirschsprung's Disease and Their Characterization. <i>Journal of Biological Chemistry</i> , 1998, 273, 11378-11383.	1.6	81
43	Oncological implications of RET gene mutations in Hirschsprung's disease. <i>Gut</i> , 1998, 43, 542-547.	6.1	59
44	Molecular Analysis of the RET and GDNF Genes in a Family with Multiple Endocrine Neoplasia Type 2A and Hirschsprung Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1998, 83, 3361-3364.	1.8	61
45	Total Colonic Aganglionosis. <i>Seminars in Pediatric Surgery</i> , 1998, 7, 174-180.	0.5	34
46	Genetic Aspects of Hirschsprung's Disease. <i>Seminars in Pediatric Surgery</i> , 1998, 7, 148-155.	0.5	32
47	Hirschsprung Disease in MEN 2A: Increased Spectrum of RET Exon 10 Genotypes and Strong Genotype-Phenotype Correlation. <i>Human Molecular Genetics</i> , 1998, 7, 129-134.	1.4	112
48	Hirschsprung's disease genes and the development of the enteric nervous system. <i>Annals of Medicine</i> , 1998, 30, 66-74.	1.5	25
49	Mutations in the Extracellular Domain Cause RET Loss of Function by a Dominant Negative Mechanism. <i>Molecular and Cellular Biology</i> , 1998, 18, 3321-3329.	1.1	54
50	A Novel 9-Base Pair Duplication in RET Exon 8 in Familial Medullary Thyroid Carcinoma. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 1700-1704.	1.8	69
51	Specific polymorphisms in the RET proto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression. <i>Journal of Medical Genetics</i> , 1999, 36, 771-774.	1.5	142
52	Autonomic dysfunction in children with Hirschsprung's disease. <i>Digestive Diseases and Sciences</i> , 1999, 44, 960-965.	1.1	36
53	Double Heterozygosity for a RET Substitution Interfering with Splicing and an EDNRB Missense Mutation in Hirschsprung Disease. <i>American Journal of Human Genetics</i> , 1999, 64, 1216-1221.	2.6	88
54	Association of RET Protooncogene Codon 45 Polymorphism with Hirschsprung Disease. <i>American Journal of Human Genetics</i> , 1999, 65, 1469-1473.	2.6	83

#	ARTICLE	IF	CITATIONS
55	Development of the human gastrointestinal tract: Twenty years of progress. <i>Gastroenterology</i> , 1999, 116, 702-731.	0.6	275
56	Sympathoadrenal Hyperplasia Causes Renal Malformations in RetMEN2B-Transgenic Mice. <i>American Journal of Pathology</i> , 1999, 155, 2167-2179.	1.9	20
57	<i>RET</i> Proto-Oncogene in the Development of Human Cancer. <i>Journal of Clinical Oncology</i> , 1999, 17, 380-380.	0.8	314
58	Genetics of Hirschsprung disease. <i>Current Opinion in Pediatrics</i> , 2000, 12, 610-617.	1.0	217
59	RET andGDNF gene scanning in Hirschsprung patients using two dual denaturing gel systems. , 2000, 15, 418-429.		86
60	Mutational analysis ofRET/GDNF/NTN genes in children with total colonic aganglionosis with small bowel involvement. <i>American Journal of Medical Genetics Part A</i> , 2000, 93, 278-284.	2.4	35
61	Familial form of Hirschsprung disease: Nucleotide sequence studies reveal point mutations in the RET proto-oncogene in two of six families but not in other candidate genes. <i>American Journal of Medical Genetics Part A</i> , 2000, 94, 19-27.	2.4	15
62	Immunolocalization of the HNK-1 epitope in the autonomic innervation to the liver and upper digestive tract of the developing rat embryo. <i>The Histochemical Journal</i> , 2000, 32, 439-446.	0.6	7
63	Idiopathic slow-transit constipation is not associated with mutations of the RET proto-oncogene or GDNF. <i>Diseases of the Colon and Rectum</i> , 2000, 43, 851-857.	0.7	14
64	Japanese patients with sporadic Hirschsprung: mutation analysis of the receptor tyrosine kinase proto-oncogene, endothelin-B receptor, endothelin-3, glial cell line-derived neurotrophic factor and neurturin genes: a comparison with similar studies. <i>European Journal of Pediatrics</i> , 2000, 159, 160-167.	1.3	27
65	A human model for multigenic inheritance: Phenotypic expression in Hirschsprung disease requires both the RET gene and a new 9q31 locus. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2000, 97, 268-273.	3.3	203
66	Evaluation of the HOX11L1 gene as a candidate for congenital disorders of intestinal innervation. <i>Journal of Medical Genetics</i> , 2000, 37, 9e-9.	1.5	36
67	RET genotypes comprising specific haplotypes of polymorphic variants predispose to isolated Hirschsprung disease. <i>Journal of Medical Genetics</i> , 2000, 37, 572-578.	1.5	93
68	Sacral Neural Crest Cells Colonise Aganglionic Hindgut in Vivo but Fail to Compensate for Lack of Enteric Ganglia. <i>Developmental Biology</i> , 2000, 219, 30-43.	0.9	149
69	Special basic science review. <i>Journal of Pediatric Surgery</i> , 2000, 35, 1017-1025.	0.8	105
70	Incidence of RET mutations in patients with Hirschsprung's disease. <i>Journal of Pediatric Surgery</i> , 2000, 35, 139-143.	0.8	67
71	Multiple Endocrine Neoplasias. <i>Annual Review of Physiology</i> , 2000, 62, 377-411.	5.6	82
72	Functional Analysis of RET With Hirschsprung Mutations Affecting Its Kinase Domain. <i>Gastroenterology</i> , 2001, 121, 24-33.	0.6	61

#	ARTICLE	IF	CITATIONS
73	The GDNF/RET signaling pathway and human diseases. <i>Cytokine and Growth Factor Reviews</i> , 2001, 12, 361-373.	3.2	387
74	Multiple Endocrine Neoplasia Type 2: Molecular Aspects. , 2001, 28, 81-102.		5
75	Lower vagal tone associated with anxiety disorders in women with irritable bowel syndrome. <i>Gastroenterology</i> , 2001, 120, A638-A638.	0.6	0
76	EDNRB/EDN3 and Hirschsprung Disease Type II. <i>Pigment Cell & Melanoma Research</i> , 2001, 14, 161-169.	4.0	97
77	Case/control family study of autonomic nervous system dysfunction in idiopathic congenital central hypoventilation syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 237-245.	2.4	83
78	Genetic segregation analysis of autonomic nervous system dysfunction in families of probands with idiopathic congenital central hypoventilation syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 229-236.	2.4	49
79	Novel RET mutations in Hirschsprung's disease patients from the diverse South African population. <i>European Journal of Human Genetics</i> , 2001, 9, 419-423.	1.4	22
80	Analysis of the RET, GDNF, EDN3, and EDNRB genes in patients with intestinal neuronal dysplasia and Hirschsprung disease. <i>Gut</i> , 2001, 48, 671-675.	6.1	73
81	The Sensitivity of Activated Cys Ret Mutants to Glial Cell Line-Derived Neurotrophic Factor Is Mandatory To Rescue Neuroectodermic Cells from Apoptosis. <i>Molecular and Cellular Biology</i> , 2001, 21, 6719-6730.	1.1	28
82	A Homozygous Missense Mutation in The Tyrosine Kinase Domain of The Ret Proto-Oncogene in An Infant With Total Intestinal Aganglionosis. <i>American Journal of Gastroenterology</i> , 2001, 96, 1286-1291.	0.2	19
83	Submicroscopic subtelomeric 1qter deletions: a recognisable phenotype?. <i>Journal of Medical Genetics</i> , 2001, 38, 175-178.	1.5	60
84	Hirschsprung disease, associated syndromes, and genetics: a review. <i>Journal of Medical Genetics</i> , 2001, 38, 729-739.	1.5	394
85	Congenital Central Hypoventilation Syndrome Associated with Hirschsprung's Disease: Mutation Analysis of the RET and Endothelin-Signaling Pathways. <i>European Journal of Pediatric Surgery</i> , 2001, 11, 335-337.	0.7	28
86	Nitroergic innervation of the normal gut and in motility disorders of childhood. <i>Journal of Pediatric Surgery</i> , 2002, 37, 551-567.	0.8	54
87	Gdnf Haploinsufficiency Causes Hirschsprung-Like Intestinal Obstruction and Early-Onset Lethality in Mice. <i>American Journal of Human Genetics</i> , 2002, 70, 435-447.	2.6	73
88	RET mutation profile and variable clinical manifestations in a family with multiple endocrine neoplasia type 2A and Hirschsprung's disease. <i>Surgery</i> , 2002, 131, 373-381.	1.0	38
89	Hirschsprung disease: A component of the familial cancer syndrome multiple endocrine neoplasia type 2a. <i>Newborn and Infant Nursing Reviews</i> , 2002, 2, 221-227.	0.4	0
90	Colonic inertia disorders in pediatrics. <i>Current Problems in Surgery</i> , 2002, 39, 671-730.	0.6	6

#	ARTICLE	IF	CITATIONS
91	Idiopathic congenital central hypoventilation syndrome: Evaluation of brain-derived neurotrophic factor genomic DNA sequence variation. American Journal of Medical Genetics Part A, 2002, 107, 306-310.	2.4	63
92	Molecular etiology of gut malformations and diseases. American Journal of Medical Genetics Part A, 2002, 115, 221-230.	2.4	62
93	Enteric Nervous System: Development and Developmental Disturbancesâ€™Part 1. Pediatric and Developmental Pathology, 2002, 5, 224-247.	0.5	182
94	PMX2B , a new candidate gene for Hirschsprung's disease. Clinical Genetics, 2003, 64, 204-209.	1.0	53
95	Cell signalling and gene expression mediated by RET tyrosine kinase. Journal of Internal Medicine, 2003, 253, 627-633.	2.7	28
96	Is there a role for the IHH gene in Hirschsprung's disease?. Neurogastroenterology and Motility, 2003, 15, 663-668.	1.6	8
97	Allelic loss of DNA locus of the RET proto-oncogene in small cell lung cancer. Cancer Letters, 2003, 195, 59-65.	3.2	7
98	A Founding Locus within the RET Proto-Oncogene May Account for a Large Proportion of Apparently Sporadic Hirschsprung Disease and a Subset of Cases of Sporadic Medullary Thyroid Carcinoma. American Journal of Human Genetics, 2003, 72, 88-100.	2.6	100
99	Intrinsic susceptibility to misfolding of a hot-spot for Hirschsprung disease mutations in the ectodomain of RET. Human Molecular Genetics, 2003, 12, 2133-2144.	1.4	56
100	Investigation of germline GFRA4 mutations and evaluation of the involvement of GFRA1, GFRA2, GFRA3, and GFRA4 sequence variants in Hirschsprung disease. Journal of Medical Genetics, 2003, 40, 18e-18.	1.5	34
101	Association of germline mutations and polymorphisms of the RET proto-oncogene with idiopathic congenital central hypoventilation syndrome in 33 patients. Journal of Medical Genetics, 2003, 40, 10e-10.	1.5	26
102	Multiple Endocrine Neoplasia Type 2. Archives of Surgery, 2003, 138, 409.	2.3	196
103	Chinese patients with sporadic Hirschsprung's disease are predominantly represented by a single RET haplotype. Journal of Medical Genetics, 2003, 40, 122e-122.	1.5	45
104	The soluble ectodomain of RetC634Y inhibits both the wild-type and the constitutively active Ret. Biochemical Journal, 2003, 372, 897-903.	1.7	24
106	Genetics of Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2004, 170, 16-21.	2.5	76
107	Interactions between Sox10 and EdnrB modulate penetrance and severity of aganglionosis in the Sox10Dom mouse model of Hirschsprung disease. Human Molecular Genetics, 2004, 13, 2289-2301.	1.4	97
108	Highly Recurrent RET Mutations and Novel Mutations in Genes of the Receptor Tyrosine Kinase and Endothelin Receptor B Pathways in Chinese Patients with Sporadic Hirschsprung Disease. Clinical Chemistry, 2004, 50, 93-100.	1.5	55
109	Interaction ofRETproto-oncogene codon 609 germline mutations with RET haplotypes characterized by c.135G>A alleles modifying MEN 2A or HSCR phenotypes. , 2004, 129A, 323-325.		6

#	ARTICLE	IF	CITATIONS
112	Molecular genetics of Hirschsprung's disease. <i>Seminars in Pediatric Surgery</i> , 2004, 13, 236-248.	0.5	30
113	Developmental biology of the enteric nervous system: Pathogenesis of Hirschsprung's disease and other congenital dysmotilities. <i>Seminars in Pediatric Surgery</i> , 2004, 13, 224-235.	0.5	71
115	Low RET mutation frequency and polymorphism analysis of the RET and EDNRB genes in patients with Hirschsprung disease in Taiwan. <i>Journal of Human Genetics</i> , 2005, 50, 168-174.	1.1	19
116	A novel susceptibility locus for Hirschsprung's disease maps to 4q31.3-q32.3. <i>Journal of Medical Genetics</i> , 2005, 43, e35-e35.	1.5	21
117	Homozygosity for a frequent and weakly penetrant predisposing allele at the RET locus in sporadic Hirschsprung disease. <i>Journal of Medical Genetics</i> , 2005, 42, e18-e18.	1.5	23
118	Ancestral RET haplotype associated with Hirschsprung's disease shows linkage disequilibrium breakpoint at -1249. <i>Journal of Medical Genetics</i> , 2005, 42, 322-327.	1.5	26
119	RET Proto-Oncogene: A Review and Update of Genotype-Phenotype Correlations in Hereditary Medullary Thyroid Cancer and Associated Endocrine Tumors. <i>Thyroid</i> , 2005, 15, 531-544.	2.4	269
120	Medullary Thyroid Cancer in a Patient with Hirschsprung Disease with a C609Y Germline RET-mutation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2005, 40, 226-229.	0.9	3
121	The combination of Hirschsprung's disease and achalasia. <i>Journal of Pediatric Surgery</i> , 2005, 40, E28-E30.	0.8	4
122	A novel Czech kindred with familial medullary thyroid carcinoma and Hirschsprung's disease. <i>Journal of Pediatric Surgery</i> , 2005, 40, e1-e6.	0.8	15
123	RET tyrosine kinase signaling in development and cancer. <i>Cytokine and Growth Factor Reviews</i> , 2005, 16, 441-467.	3.2	397
124	Neurotrophic Factor Expression in Three Infants With Ondine's Curse. <i>Pediatric Neurology</i> , 2005, 33, 331-336.	1.0	12
125	Proteome analysis of isolated myenteric plexus reveals significant changes in protein expression during postnatal development. <i>Autonomic Neuroscience: Basic and Clinical</i> , 2005, 122, 1-8.	1.4	12
126	Phox2b function in the enteric nervous system is conserved in zebrafish and is sox10-dependent. <i>Mechanisms of Development</i> , 2005, 122, 659-669.	1.7	126
127	Molecular mechanisms of RET-induced Hirschsprung pathogenesis. <i>Annals of Medicine</i> , 2006, 38, 11-19.	1.5	36
128	A De Novo Novel Mutation of the EDNRB Gene in a Taiwanese Boy with Hirschsprung Disease. <i>Journal of the Formosan Medical Association</i> , 2006, 105, 349-354.	0.8	10
129	Novel mutations of RET gene in Korean patients with sporadic Hirschsprung's disease. <i>Journal of Pediatric Surgery</i> , 2006, 41, 1250-1254.	0.8	13
130	New variations of the EDNRB gene and its association with sporadic Hirschsprung's disease in Korea. <i>Journal of Pediatric Surgery</i> , 2006, 41, 1708-1712.	0.8	6

#	ARTICLE	IF	CITATIONS
131	Haplotypes of the Human RET Proto-oncogene Associated with Hirschsprung Disease in the Italian Population Derive from a Single Ancestral Combination of Alleles. <i>Annals of Human Genetics</i> , 2006, 70, 12-26.	0.3	38
132	Single nucleotide polymorphisms in the RET gene and their correlations with Hirschsprung disease phenotype. <i>Journal of Applied Genetics</i> , 2006, 47, 261-267.	1.0	3
133	A complex additive model of inheritance for Hirschsprung disease is supported by both RET mutations and predisposing RET haplotypes. <i>Genetics in Medicine</i> , 2006, 8, 704-710.	1.1	29
134	Hirschsprung disease, associated syndromes and genetics: a review. <i>Journal of Medical Genetics</i> , 2007, 45, 1-14.	1.5	848
135	Gastrointestinal Motility Disorders in Adolescent Patients: Transitioning to Adult Care. <i>Gastroenterology Clinics of North America</i> , 2007, 36, 749-763.	1.0	9
136	Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2007, 15, 242-245.	1.4	13
137	Prokineticin-1 modulates proliferation and differentiation of enteric neural crest cells. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2007, 1773, 536-545.	1.9	39
138	RET polymorphisms and the risk of Hirschsprung's disease in a Chinese population. <i>Journal of Human Genetics</i> , 2008, 53, 825-833.	1.1	8
139	The nervous system and gastrointestinal function. <i>Developmental Disabilities Research Reviews</i> , 2008, 14, 87-95.	2.9	40
140	Low frequency of <i>RET</i> mutations in Hirschsprung disease in Sweden. <i>Clinical Genetics</i> , 1998, 54, 39-44.	1.0	41
141	Prokineticin-1 (Prok-1) works coordinately with glial cell line-derived neurotrophic factor (GDNF) to mediate proliferation and differentiation of enteric neural crest cells. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2008, 1783, 467-478.	1.9	32
142	Functional Anatomy of the Enteric Nervous System. , 2008, , 21-49.		17
143	Development of the Enteric Nervous System. , 2008, , 13-20.		4
144	Management of Medullary Thyroid Carcinoma. <i>Endocrinology and Metabolism Clinics of North America</i> , 2008, 37, 481-496.	1.2	75
145	Pediatric Anorectal Disorders. <i>Gastroenterology Clinics of North America</i> , 2008, 37, 709-730.	1.0	12
146	Hirschsprung's Disease, One of the Most Difficult Diagnoses in Pediatric Surgery: A Review of the Problems from Clinical Practice to the Bench. <i>European Journal of Pediatric Surgery</i> , 2008, 18, 140-149.	0.7	91
147	Genetic basis of Hirschsprung's disease. <i>Pediatric Surgery International</i> , 2009, 25, 543-558.	0.6	117
148	Analyses of PRMT1 proteins in human colon tissues from Hirschsprung disease patients. <i>Neurogastroenterology and Motility</i> , 2010, 22, 984.	1.6	5

#	ARTICLE	IF	CITATIONS
149	Genetic background impacts developmental potential of enteric neural crest-derived progenitors in the Sox10Dom model of Hirschsprung disease. <i>Human Molecular Genetics</i> , 2010, 19, 4353-4372.	1.4	46
150	Neural Precursor Death Is Central to the Pathogenesis of Intestinal Aganglionosis in Ret Hypomorphic Mice. <i>Journal of Neuroscience</i> , 2010, 30, 5211-5218.	1.7	44
152	Common alleles of predisposition in endocrine neoplasia. <i>Current Opinion in Genetics and Development</i> , 2010, 20, 251-256.	1.5	7
153	Developmental determinants of the independence and complexity of the enteric nervous system. <i>Trends in Neurosciences</i> , 2010, 33, 446-456.	4.2	137
154	Genetics of Hirschsprung disease and anorectal malformations. <i>Seminars in Pediatric Surgery</i> , 2010, 19, 107-117.	0.5	50
155	Polymorphisms of the RET Gene in Hirschsprung Disease, Anorectal Malformation and Intestinal Pseudo-obstruction in Taiwan. <i>Journal of the Formosan Medical Association</i> , 2010, 109, 32-38.	0.8	12
156	Principles of Clinical Cancer Genetics. , 2010, , .		3
157	RET and GDNF mutations are rare in fetuses with renal agenesis or other severe kidney development defects. <i>Journal of Medical Genetics</i> , 2011, 48, 497-504.	1.5	60
158	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2012, 20, 917-920.	1.4	8
159	A Sonic hedgehog (Shh) response deficit in trisomic cells may be a common denominator for multiple features of Down syndrome. <i>Progress in Brain Research</i> , 2012, 197, 223-236.	0.9	33
160	Hirschsprung's disease and variants in genes that regulate enteric neural crest cell proliferation, migration and differentiation. <i>Journal of Human Genetics</i> , 2012, 57, 485-493.	1.1	30
161	Intronic RET gene variants in Down syndrome-associated Hirschsprung disease in an African population. <i>Journal of Pediatric Surgery</i> , 2012, 47, 299-302.	0.8	21
163	Advances in Molecular Genetics of Hirschsprung's Disease. <i>Anatomical Record</i> , 2012, 295, 1628-1638.	0.8	25
164	Downregulation of Notch-1/Jagged-2 in human colon tissues from Hirschsprung disease patients. <i>International Journal of Colorectal Disease</i> , 2012, 27, 37-41.	1.0	5
165	Contribution of rare and common variants determine complex diseases—Hirschsprung disease as a model. <i>Developmental Biology</i> , 2013, 382, 320-329.	0.9	119
166	Inhibition of RET Activated Pathways: Novel Strategies for Therapeutic Intervention in Human Cancers. <i>Current Pharmaceutical Design</i> , 2013, 19, 864-882.	0.9	5
167	A novel stop mutation in the EDNRB gene in a family with Hirschsprung's disease associated with Multiple Sclerosis. <i>Journal of Pediatric Surgery</i> , 2014, 49, 622-625.	0.8	9
168	2013 William Allan Award Introduction: Aravinda Chakravarti. <i>American Journal of Human Genetics</i> , 2014, 94, 324-325.	2.6	0

#	ARTICLE	IF	CITATIONS
169	The association between Hirschsprungâ€™s disease and multiple endocrine neoplasia type 2a: a systematic review. <i>Pediatric Surgery International</i> , 2014, 30, 751-756.	0.6	36
170	2013 William Allan Award: My Multifactorial Journey. <i>American Journal of Human Genetics</i> , 2014, 94, 326-333.	2.6	2
171	Tissue specific somatic mutations and aganglionosis in Hirschsprung's disease. <i>Journal of Pediatric Surgery</i> , 2014, 49, 258-261.	0.8	14
172	Surgical Treatment of Colorectal Problems in Children. , 2015, , .		35
173	RET gene is a major risk factor for Hirschsprungâ€™s disease: a meta-analysis. <i>Pediatric Surgery International</i> , 2015, 31, 701-710.	0.6	49
174	Clinical and genetic correlations of familial Hirschsprungâ€™s disease. <i>Journal of Pediatric Surgery</i> , 2015, 50, 285-288.	0.8	12
175	Association Analysis of <i>SLC6A20</i> Polymorphisms With Hirschsprung Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2016, 62, 64-70.	0.9	7
176	2015 ASHG Awards and Addresses. <i>American Journal of Human Genetics</i> , 2016, 98, 405-406.	2.6	1
177	Cadherin-Related Diseases. , 2016, , 399-421.		1
178	The Cadherin Superfamily. , 2016, , .		1
179	Defects in the calcium-binding region drastically affect the cadherin-like domains of RET tyrosine kinase. <i>Physical Chemistry Chemical Physics</i> , 2016, 18, 8673-8681.	1.3	2
180	RET and EDNRB mutation screening in patients with Hirschsprung disease: Functional studies and its implications for genetic counseling. <i>European Journal of Human Genetics</i> , 2016, 24, 823-829.	1.4	20
181	Hirschsprungâ€™s Disease and Intestinal Neuronal Dysplasias. , 2016, , 261-268.		1
183	Defining the transcriptomic landscape of the developing enteric nervous system and its cellular environment. <i>BMC Genomics</i> , 2017, 18, 290.	1.2	38
184	Advances in understanding the association between Down syndrome and Hirschsprung disease (DSâ€™HSCR). <i>Pediatric Surgery International</i> , 2018, 34, 1127-1137.	0.6	18
185	Sequence characterization of RET in 117 Chinese Hirschsprung disease families identifies a large burden of de novo and parental mosaic mutations. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 237.	1.2	10
186	Familial Hirschsprungâ€™s Disease. , 2019, , 115-119.		2
187	Development of the Enteric Nervous System. , 2019, , 19-29.		1

#	ARTICLE	IF	CITATIONS
188	Functional Anatomy of the Enteric Nervous System. , 2019, , 31-76.		7
189	Profile of Aravinda Chakravarti. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 10608-10610.	3.3	0
190	What is new about the genetic background of Hirschsprung disease?. Clinical Genetics, 2020, 97, 114-124.	1.0	24
191	The overall risk of malignancies is not increased in patients with Hirschsprung disease. Pediatric Surgery International, 2020, 36, 471-475.	0.6	0
192	Kidney malformations and Hirschsprung's disease in carriers of cysteine mutations in exon 10 of the RET proto-oncogene. Endocrine, 2021, 73, 217-222.	1.1	1
193	The enteric nervous system in gastrointestinal disease etiology. Cellular and Molecular Life Sciences, 2021, 78, 4713-4733.	2.4	58
194	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	3.9	24
195	The Emerging Genetic Landscape of Hirschsprung Disease and Its Potential Clinical Applications. Frontiers in Pediatrics, 2021, 9, 638093.	0.9	25
196	The relationship between specific RET proto-oncogene mutations and disease phenotype in multiple endocrine neoplasia type 2. International RET mutation consortium analysis. JAMA - Journal of the American Medical Association, 1996, 276, 1575-1579.	3.8	516
197	Melanoblast Development and Associated Disorders. , 0, , 140-154.		4
198	Multiple endocrine neoplasia type II and familial medullary thyroid carcinoma Impact of genetic screening on management. Cancer Treatment and Research, 1997, 89, 421-441.	0.2	12
199	Hirschsprung's Disease. , 2015, , 397-434.		3
200	The Molecular Genetics of Hirschsprung's Disease. , 2008, , 63-78.		5
201	Mutations in Ret in MEN 2. Medical Intelligence Unit, 1996, , 21-35.	0.2	2
202	Hirschsprung's Disease and Related Neuromuscular Disorders of the Intestine. , 2006, , 1514-1559.		21
203	Development of the Enteric Nervous System. , 2006, , 499-521.		8
204	Genomic Variation in Multigenic Traits: Hirschsprung Disease. Cold Spring Harbor Symposia on Quantitative Biology, 2003, 68, 373-382.	2.0	30
205	A heterozygous endothelin 3 mutation in Waardenburg-Hirschsprung disease: is there a dosage effect of EDN3/EDNRB gene mutations on neurocristopathy phenotypes?. Journal of Medical Genetics, 2001, 38, 205-209.	1.5	50

#	ARTICLE	IF	CITATIONS
206	Prevalence and Parental Origin of de novo RET Mutations in Hirschsprung's Disease. European Journal of Human Genetics, 1996, 4, 356-358.	1.4	13
207	Various mechanisms cause RET-mediated signaling defects in Hirschsprung's disease.. Journal of Clinical Investigation, 1998, 101, 1415-1423.	3.9	99
208	Dosage Effects of Cohesin Regulatory Factor PDS5 on Mammalian Development: Implications for Cohesinopathies. PLoS ONE, 2009, 4, e5232.	1.1	74
209	Induction of RET Dependent and Independent Pro-Inflammatory Programs in Human Peripheral Blood Mononuclear Cells from Hirschsprung Patients. PLoS ONE, 2013, 8, e59066.	1.1	24
210	Roles of the <i>RET</i> Proto-oncogene in Cancer and Development. JMA Journal, 2020, 3, 175-181.	0.6	34
211	New techniques in the tissue diagnosis of gastrointestinal neuromuscular diseases. World Journal of Gastroenterology, 2009, 15, 192.	1.4	10
212	Tiermodelle in der biomedizinischen Forschung. , 2003, , 299-339.		0
213	Do RET and APC Crosstalk in Hirschsprungs Disease Pathogenesis?. Trends in Medical Research, 2008, 3, 31-35.	0.2	0
214	Multiple Endocrine Neoplasia Type 2. , 2010, , 2742-2758.		0
215	Multiple Endocrine Neoplasia. , 2010, , 145-162.		0
216	Neural Crest and Hirschsprungâ€™s Disease. , 2012, , 353-386.		1
217	The Contributions of RET Noncoding Variation to Hirschsprung Disease. , 2012, , 169-194.		0
218	RET. , 1997, , 341-349.		0
219	Hirschsprungâ€™s Disease. , 2017, , 1-20.		0
220	Hirschsprungâ€™s Disease. , 2019, , 1-20.		0
222	Hirschsprungâ€™s Disease. , 2020, , 1011-1030.		0
223	Multiple Endocrine Neoplasia Type 2. , 2006, , 393-399.		4
224	Molecular heterogeneity of RET loss of function in Hirschsprung's disease. EMBO Journal, 1996, 15, 2717-25.	3.5	30

#	ARTICLE	IF	CITATIONS
225	Hereditary multiple exostoses (EXT): mutational studies of familial EXT1 cases and EXT-associated malignancies. <i>American Journal of Human Genetics</i> , 1997, 60, 80-6.	2.6	99
226	The locus for a novel syndromic form of neuronal intestinal pseudoobstruction maps to Xq28. <i>American Journal of Human Genetics</i> , 1996, 58, 743-8.	2.6	66
228	A multi-enhancer <i>RET</i> regulatory code is disrupted in Hirschsprung disease. <i>Genome Research</i> , 2021, 31, 2199-2208.	2.4	10
229	Hirschsprung's Disease and Intestinal Neuronal Dysplasias. , 2022, , 305-312.		0
230	The <i>RET</i> gene encodes RET protein, which triggers intracellular signaling pathways for enteric neurogenesis, and <i>RET</i> mutation results in Hirschsprung's disease. <i>AIMS Neuroscience</i> , 2022, 9, 128-149.	1.0	5
232	<i>RET</i> Proto-Oncogene "Not Such an Obvious Starting Point in Cancer Therapy. <i>Cancers</i> , 2022, 14, 5298.	1.7	2
233	Association of rs2435357 and rs2506030 polymorphisms in <i>RET</i> with susceptibility to hirschsprung disease: A systematic review and meta-analysis. <i>Frontiers in Pediatrics</i> , 0, 10, .	0.9	0
234	Comprehensive characterization of the genetic landscape of familial Hirschsprung's disease. <i>World Journal of Pediatrics</i> , 2023, 19, 644-651.	0.8	5