Isabella Ceccherini

List of Publications by Year
in descending order

258	17,352	34105 52	15732 125
papers	citations	h-index	g-index

270	270	270	23234
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	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
3	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	3.2	848
4	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	21.4	733
5	Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 377-378.	27.8	722
6	An Official ATS Clinical Policy Statement: Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2010, 181, 626-644.	5.6	433
7	Classification criteria for autoinflammatory recurrent fevers. Annals of the Rheumatic Diseases, 2019, 78, 1025-1032.	0.9	300
8	PHOX2B mutations and polyalanine expansions correlate with the severity of the respiratory phenotype and associated symptoms in both congenital and late onset Central Hypoventilation syndrome. Journal of Medical Genetics, 2004, 41, 373-380.	3.2	248
9	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
10	ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study. Annals of the Rheumatic Diseases, 2017, 76, 1648-1656.	0.9	199
11	Pyogenic Arthritis, Pyoderma Gangrenosum, Acne, and Hidradenitis Suppurativa (PAPASH): A New Autoinflammatory Syndrome Associated With a Novel Mutation of the PSTPIP1 Gene. JAMA Dermatology, 2013, 149, 762.	4.1	183
12	RET mutations in human disease. Trends in Genetics, 1996, 12, 138-144.	6.7	175
13	Stable length polymorphism of up to 260 kb at the tip of the short arm of human chromosome 16. Cell, 1991, 64, 595-606.	28.9	169
14	Adult-onset Alexander disease: a series of eleven unrelated cases with review of the literature. Brain, 2008, 131, 2321-2331.	7.6	169
15	A diagnostic score for molecular analysis of hereditary autoinflammatory syndromes with periodic fever in children. Arthritis and Rheumatism, 2008, 58, 1823-1832.	6.7	165
16	Clinical presentation and pathogenesis of cold-induced autoinflammatory disease in a family with recurrence of an NLRP12 mutation. Arthritis and Rheumatism, 2011, 63, 830-839.	6.7	162
17	Guidelines for the genetic diagnosis of hereditary recurrent fevers. Annals of the Rheumatic Diseases, 2012, 71, 1599-1605.	0.9	160
18	Autoinflammation in pyoderma gangrenosum and its syndromic form (pyoderma gangrenosum, acne) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.5	151

#	Article	IF	Citations
19	Frequency of RET mutations in long- and short-segment Hirschsprung disease. Human Mutation, 1997, 9, 243-249.	2.5	138
20	Neutrophils from patients withTNFRSF1A mutations display resistance to tumor necrosis factorâ€“induced apoptosis: Pathogenetic and clinical implications. Arthritis and Rheumatism, 2006, 54, 998-1008.	6.7	138
21	Differentiating PFAPA Syndrome From Monogenic Periodic Fevers. Pediatrics, 2009, 124, e721-e728.	2.1	138
22	Contribution of rare and common variants determine complex diseasesâ€”Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119
23	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
24	New workflow for classification of genetic variantsâ€™ pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). Journal of Medical Genetics, 2018, 55, 530-537.	3.2	117
25	Multiple endocrine neoplasia type 2 syndromes (MEN 2): results from the ItaMEN network analysis on the prevalence of different genotypes and phenotypes. European Journal of Endocrinology, 2010, 163, 301-308.	3.7	111
26	Association of Pyoderma Gangrenosum, Acne, and Suppurative Hidradenitis (PASH) Shares Genetic and Cytokine Profiles With Other Autoinflammatory Diseases. Medicine (United States), 2014, 93, e187.	1.0	108
27	Distinct pathogenetic mechanisms for PHOX2B associated polyalanine expansions and frameshift mutations in congenital central hypoventilation syndrome. Human Molecular Genetics, 2005, 14, 1815-1824.	2.9	106
28	Special basic science review. Journal of Pediatric Surgery, 2000, 35, 1017-1025.	1.6	105
29	Somatic mutations of the ret protooncogene in sporadic medullary thyroid carcinoma are not restricted to exon 16 and are associated with tumor recurrence. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 1619-1622.	3.6	103
30	Nuclear Run-On Assay Using Biotin Labeling, Magnetic Bead Capture and Analysis by Fluorescence-Based RT-PCR. BioTechniques, 2000, 29, 1012-1017.	1.8	102
31	Can MR Imaging Diagnose Adult-Onset Alexander Disease?. American Journal of Neuroradiology, 2008, 29, 1190-1196.	2.4	99
32	Congenital central hypoventilation syndrome from past to future: Model for translational and transitional autonomic medicine. Pediatric Pulmonology, 2009, 44, 521-535.	2.0	99
33	Longâ€term clinical profile of children with the lowâ€penetrance R92Q mutation of the TNFRSF1A gene. Arthritis and Rheumatism, 2011, 63, 1141-1150.	6.7	99
34	Betaine, Dimethyl Sulfoxide, and 7-Deaza-dGTP, a Powerful Mixture for Amplification of GC-Rich DNA Sequences. Journal of Molecular Diagnostics, 2006, 8, 544-550.	2.8	92
35	Close linkage with the RET protooncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. Human Molecular Genetics, 1993, 2, 1803-1808.	2.9	88
36	Double Heterozygosity for a RET Substitution Interfering with Splicing and an EDNRB Missense Mutation in Hirschsprung Disease. American Journal of Human Genetics, 1999, 64, 1216-1221.	6.2	88

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37	Exon Structure and Flanking Intronic Sequences of the Human RET Proto-oncogene. Biochemical and Biophysical Research Communications, 1993, 196, 1288-1295.	2.1	82
38	Interstitial deletion of the Endothelin-B receptor gene in the spotting lethal (sl) rat. Human Molecular Genetics, 1995, 4, 2089-2096.	2.9	79
39	Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. Journal of Internal Medicine, 1998, 243, 515-520.	6.0	74
40	Guidelines for diagnosis and management of congenital central hypoventilation syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 252.	2.7	74
41	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
42	Identification of the Cys634->Tyr mutation of the RET proto-oncogene in a pedigree with multiple endocrine neoplasia type 2A and localized cutaneous lichen amyloidosis. Journal of Endocrinological Investigation, 1994, 17, 201-204.	3.3	71
43	MVK mutations and associated clinical features in Italian patients affected with autoinflammatory disorders and recurrent fever. European Journal of Human Genetics, 2005, 13, 314-320.	2.8	71
44	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). Annals of the Rheumatic Diseases, 2013, 72, 1044-1052.	0.9	69
45	Clinical and genetic characterization of Italian patients affected by CINCA syndrome. Rheumatology, 2007, 46, 473-478.	1.9	68
46	Incidence of RET mutations in patients with Hirschsprung's disease. Journal of Pediatric Surgery, 2000, 35, 139-143.	1.6	67
47	Clinical impact ofMEFVmutations in children with periodic fever in a prevalent western European Caucasian population. Annals of the Rheumatic Diseases, 2012, 71, 1961-1965.	0.9	65
48	Hirschsprung Disease and Congenital Anomalies of the Kidney and Urinary Tract (CAKUT). Medicine (United States), 2009, 88, 83-90.	1.0	62
49	Heterogeneity and Low Detection Rate of RET Mutations in Hirschsprung Disease. European Journal of Human Genetics, 1994, 2, 272-280.	2.8	60
50	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	2.5	57
51	Next-generation sequencing and its initial applications for molecular diagnosis of systemic auto-inflammatory diseases. Annals of the Rheumatic Diseases, 2016, 75, 1550-1557.	0.9	57
52	Parental origin and somatic mosaicism of PHOX2B mutations in Congenital Central Hypoventilation Syndrome. Human Mutation, 2008, 29, 206-206.	2.5	52
53	A web-based collection of genotype-phenotype associations in hereditary recurrent fevers from the Eurofever registry. Orphanet Journal of Rare Diseases, 2017, 12, 167.	2.7	52
54	Congenital central hypoventilation syndrome (CCHS) and sudden infant death syndrome (SIDS): Kindred disorders of autonomic regulation. Respiratory Physiology and Neurobiology, 2008, 164, 38-48.	1.6	51

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55	Single nucleotide polymorphic alleles in the 5' region of the RET proto-oncogene define a risk haplotype in Hirschsprung's disease. Journal of Medical Genetics, 2003, 40, 714-718.	3.2	50
56	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.	3.2	50
57	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
58	Molecular analysis of mutations in the hprt gene in circulating lymphocytes from normal and DNA-repair-deficient donors. Mutation Research DNA Repair, 1993, 294, 29-41.	3.7	47
59	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
60	A common haplotype at the 5â€² end of the RET protoâ€oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. Human Mutation, 2005, 25, 189-195.	2.5	46
61	Recent advances in the developmental origin of neuroblastoma: an overview. Journal of Experimental and Clinical Cancer Research, 2022, 41, 92.	8.6	46
62	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
63	PHOX2B mutations and genetic predisposition to neuroblastoma. Oncogene, 2005, 24, 3050-3053.	5.9	45
64	Gender-dependent disease severity in autosomal polycystic kidney disease of rats. Kidney International, 1995, 48, 496-500.	5.2	44
65	Genetics of enteric neuropathies. Developmental Biology, 2016, 417, 198-208.	2.0	44
66	Variants of the ACTG2 gene correlate with degree of severity and presence of megacystis in chronic intestinal pseudo-obstruction. European Journal of Human Genetics, 2016, 24, 1211-1215.	2.8	43
67	ISSAID/EMQN Best Practice Guidelines for the Genetic Diagnosis of Monogenic Autoinflammatory Diseases in the Next-Generation Sequencing Era. Clinical Chemistry, 2020, 66, 525-536.	3.2	43
68	Genomic Structure of the Human Lysosomal Î±-Mannosidase Gene (MANB). Genomics, 1997, 42, 200-207.	2.9	42
69	Location of the first genetic locus, PKDr1, controlling autosomal dominant polycystic kidney disease in Han:SPRD cy/+ rat. Human Molecular Genetics, 1997, 6, 609-613.	2.9	41
70	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
71	The TLX2 homeobox gene is a transcriptional target of PHOX2B in neural-crest-derived cells. Biochemical Journal, 2006, 395, 355-361.	3.7	41
72	Specific haplotypes of the RET proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma. Journal of Medical Genetics, 2002, 39, 260-265.	3.2	40

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73	PHOX2B-Mediated Regulation of ALK Expression: In Vitro Identification of a Functional Relationship between Two Genes Involved in Neuroblastoma. PLoS ONE, 2010, 5, e13108.	2.5	40
74	Causative and common PHOX2B variants define a broad phenotypic spectrum. Clinical Genetics, 2020, 97, 103-113.	2.0	39
75	Haplotypes of the Human RET Proto-oncogene Associated with Hirschsprung Disease in the Italian Population Derive from a Single Ancestral Combination of Alleles. Annals of Human Genetics, 2006, 70, 12-26.	0.8	38
76	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	2.9	38
77	Low amounts of PHOX2B expanded alleles in asymptomatic parents suggest unsuspected recurrence risk in congenital central hypoventilation syndrome. Journal of Molecular Medicine, 2011, 89, 505-513.	3.9	37
78	Somatic in frame deletions not involving juxtamembranous cysteine residues strongly activate the RET proto-oncogene. Oncogene, 1997, 14, 2609-2612.	5.9	36
79	Molecular mechanisms ofRETâ€induced Hirschsprung pathogenesis. Annals of Medicine, 2006, 38, 11-19.	3.8	36
80	Genomics approach to the analysis of bacterial communities dynamics in Hirschsprungâ€™s disease-associated enterocolitis: a pilot study. Pediatric Surgery International, 2010, 26, 465-471.	1.4	36
81	In vitro treatments with ceftriaxone promote elimination of mutant glial fibrillary acidic protein and transcription down-regulation. Experimental Cell Research, 2010, 316, 2152-2165.	2.6	36
82	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	1.9	36
83	A common variant located in the 3â€²UTR of theRET gene is associated with protection from Hirschsprung disease. Human Mutation, 2007, 28, 168-176.	2.5	35
84	Geldanamycin promotes nuclear localisation and clearance of PHOX2B misfolded proteins containing polyalanine expansions. International Journal of Biochemistry and Cell Biology, 2007, 39, 327-339.	2.8	34
85	A prospective observational study of associated anomalies in Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 184.	2.7	33
86	In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins. Neurobiology of Disease, 2012, 45, 508-518.	4.4	32
87	Clinical Characteristics of Patients Carrying the Q703K Variant of the NLRP3 Gene: A 10-year Multicentric National Study. Journal of Rheumatology, 2016, 43, 1093-1100.	2.0	31
88	CD70 Deficiency due to a Novel Mutation in a Patient with Severe Chronic EBV Infection Presenting As a Periodic Fever. Frontiers in Immunology, 2017, 8, 2015.	4.8	31
89	Construction of a map of chromosome 16 by using radiation hybrids.. Proceedings of the National Academy of Sciences of the United States of America, 1992, 89, 104-108.	7.1	30
90	Diagnostic and therapeutic approach to multiple endocrine neoplasia type 2B in pediatric patients. Pediatric Surgery International, 2002, 18, 378-383.	1.4	29

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91	GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. Clinical Genetics, 2007, 72, 427-433.	2.0	29
92	The involvement of the RET variant G691S in medullary thyroid carcinoma enlightened by a metaâ€analysis study. International Journal of Cancer, 2013, 132, 2808-2819.	5.1	29
93	Transcriptional dysregulation and impairment of PHOX2B auto-regulatory mechanism induced by polyalanine expansion mutations associated with congenital central hypoventilation syndrome. Neurobiology of Disease, 2013, 50, 187-200.	4.4	29
94	FASâ€mediated apoptosis impairment in patients with ALPS/ALPSâ€like phenotype carrying variants on CASP10 gene. British Journal of Haematology, 2019, 187, 502-508.	2.5	29
95	Dysregulation in Bâ€cell responses and T follicular helper cell function in ADA2 deficiency patients. European Journal of Immunology, 2021, 51, 206-219.	2.9	29
96	The Sensitivity of Activated Cys Ret Mutants to Glial Cell Line-Derived Neurotrophic Factor Is Mandatory To Rescue Neuroectodermic Cells from Apoptosis. Molecular and Cellular Biology, 2001, 21, 6719-6730.	2.3	28
97	Adult-onset Alexander disease. Journal of Neurology, 2008, 255, 24-30.	3.6	28
98	Cryopyrin-associated Periodic Syndromes in Italian Patients: Evaluation of the Rate of Somatic NLRP3 Mosaicism and Phenotypic Characterization. Journal of Rheumatology, 2017, 44, 1667-1673.	2.0	28
99	Structural and functional differences inPHOX2Bframeshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. Human Mutation, 2018, 39, 219-236.	2.5	28
100	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	7.6	28
101	Autosomal dominant polycystic kidney disease: Prenatal diagnosis by DNA analysis and sonography at 14 weeks. Prenatal Diagnosis, 1989, 9, 751-758.	2.3	26
102	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
103	Beneficial effects of curcumin on GFAP filament organization and down-regulation of GFAP expression in an in vitro model of Alexander disease. Experimental Cell Research, 2012, 318, 1844-1854.	2.6	26
104	Novel spondyloepimetaphyseal dysplasia due to UFSP2 gene mutation. Clinical Genetics, 2018, 93, 671-674.	2.0	26
105	miR-204 mediates post-transcriptional down-regulation of PHOX2B gene expression in neuroblastoma cells. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2015, 1849, 1057-1065.	1.9	25
106	Common PHOX2B poly-alanine contractions impair RET gene transcription, predisposing to Hirschsprung disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1770-1777.	3.8	25
107	Whole exome sequencing approach to childhood onset familial erythrodermic psoriasis unravels a novel mutation of CARD14 requiring unusual high doses of ustekinumab. Pediatric Rheumatology, 2019, 17, 38.	2.1	25
108	Glomerulocystic kidney disease in a family. Nephrology Dialysis Transplantation, 2002, 17, 813-818.	0.7	24

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109	An In Vitro Approach to Test the Possible Role of Candidate Factors in the Transcriptional Regulation of the <I>RET</I> Proto-Oncogene. Gene Expression, 2005, 12, 137-149.	1.2	24
110	Induction of RET Dependent and Independent Pro-Inflammatory Programs in Human Peripheral Blood Mononuclear Cells from Hirschsprung Patients. PLoS ONE, 2013, 8, e59066.	2.5	24
111	Tumor necrosis factor receptor-associated periodic syndrome as a model linking autophagy and inflammation in protein aggregation diseases. Journal of Molecular Medicine, 2014, 92, 583-594.	3.9	23
112	Genetic and epigenetic factors affect RET gene expression in breast cancer cell lines and influence survival in patients. Oncotarget, 2016, 7, 26465-26479.	1.8	23
113	Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A and PHOX2B genes. European Journal of Human Genetics, 2007, 15, 848-855.	2.8	22
114	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
115	A young female with early onset arthritis, uveitis, hepatic, and renal granulomas: a clinical tryst with Blau syndrome over 20Âyears and case-based review. Rheumatology International, 2021, 41, 173-181.	3.0	22
116	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
117	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. International Journal of Molecular Medicine, 2002, 10, 101-6.	4.0	22
118	Brainstem Anomalies in Two Patients Affected by Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 706-709.	5.6	21
119	Brainstem signs with progressing atrophy of medulla oblongata and upper cervical spinal cord. Lancet Neurology, The, 2007, 6, 562-570.	10.2	21
120	Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. Orphanet Journal of Rare Diseases, 2013, 8, 66.	2.7	21
121	Mutations inMYO1Hcause a recessive form of central hypoventilation with autonomic dysfunction. Journal of Medical Genetics, 2017, 54, 754-761.	3.2	21
122	High-dose ustekinumab for severe childhood deficiency of interleukin-36 receptor antagonist (DITRA). Annals of the Rheumatic Diseases, 2018, 77, annrheumdis-2017-211805.	0.9	21
123	Mild functional effects of a novel GFAP mutant allele identified in a familial case of adult-onset Alexander disease. European Journal of Human Genetics, 2008, 16, 462-470.	2.8	20
124	A novel mutation in the GFAP gene in a familial adult onset Alexander disease. Journal of Neurology, 2007, 254, 1278-1280.	3.6	19
125	Congenital central hypoventilation syndrome: genotypeâ€“phenotype correlation in parents of affected children carrying a PHOX2B expansion mutation. Clinical Genetics, 2010, 78, 289-293.	2.0	19
126	A Metagenomics Study on Hirschsprung's Disease Associated Enterocolitis: Biodiversity and Gut Microbial Homeostasis Depend on Resection Length and Patient's Clinical History. Frontiers in Pediatrics, 2019, 7, 326.	1.9	19

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127	Genetic mapping of the RET protooncogene on rat Chromosome 4. Mammalian Genome, 1995, 6, 433-435.	2.2	18
128	Late-onset and long-lasting autoimmune neutropenia: an analysis from the Italian Neutropenia Registry. Blood Advances, 2020, 4, 5644-5649.	5.2	18
129	Cell-line specific chromatin acetylation at the Sox10-Pax3 enhancer site modulates theRETproto-oncogene expression. FEBS Letters, 2002, 523, 123-127.	2.8	17
130	Mutational analysis of theRNX gene in congenital central hypoventilation syndrome. American Journal of Medical Genetics Part A, 2002, 113, 178-182.	2.4	17
131	Rescue of human RET gene expression by sodium butyrate: a novel powerful tool for molecular studies in Hirschsprung disease. Gut, 2003, 52, 1154-1158.	12.1	17
132	Candidate Genes in Patients with Autoinflammatory Syndrome Resembling Tumor Necrosis Factor Receptor-associated Periodic Syndrome Without Mutations in the TNFRSF1A Gene. Journal of Rheumatology, 2011, 38, 1378-1384.	2.0	17
133	The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome-associated polyalanine-expanded PHOX2B. Journal of Molecular Medicine, 2012, 90, 1025-1035.	3.9	17
134	Pathways systematically associated to Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 187.	2.7	17
135	Recessive NLRC4-Autoinflammatory Disease Reveals an Ulcerative Colitis Locus. Journal of Clinical Immunology, 2022, 42, 325-335.	3.8	17
136	Ceftriaxone has a therapeutic role in Alexander disease. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 416-417.	4.8	16
137	A case report of a novel compound heterozygous mutation in a Brazilian patient with deficiency of Interleukin-1 receptor antagonist (DIRA). Pediatric Rheumatology, 2020, 18, 67.	2.1	16
138	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotypeâ€“phenotype correlation in congenital central hypoventilation syndrome (CCHS). Genetics in Medicine, 2021, 23, 1656-1663.	2.4	16
139	A Focus on Regulatory Networks Linking MicroRNAs, Transcription Factors and Target Genes in Neuroblastoma. Cancers, 2021, 13, 5528.	3.7	16
140	Gene expression profile in TNF receptor-associated periodic syndrome reveals constitutively enhanced pathways and new players in the underlying inflammation. Clinical and Experimental Rheumatology, 2016, 34, S121-S128.	0.8	16
141	Caffeine post-treatment causes a shift in the chromosome aberration types induced by mitomycin C, suggesting a caffeinesensitive mechanism of DNA repair in G2. Mutagenesis, 1988, 3, 39-44.	2.6	15
142	Oligogenic inheritance in neuroblastoma. Cancer Letters, 2005, 228, 65-69.	7.2	15
143	ABCC6 mutations and early onset stroke: Two cases of a typical Pseudoxanthoma Elasticum. European Journal of Paediatric Neurology, 2018, 22, 725-728.	1.6	15
144	Search for pathogenetic variants of the SPRY2 gene in intestinal innervation defects. Internal Medicine Journal, 2009, 39, 335-337.	0.8	14

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145	A Novel Polymorphic APâ€1 Binding Element of the GFAP Promoter is Associated with Different Allelic Transcriptional Activities. Annals of Human Genetics, 2010, 74, 506-515.	0.8	14
146	Recurrence of CCHS associated PHOX2B polyâ€alanine expansion mutation due to maternal mosaicism. Pediatric Pulmonology, 2014, 49, E45-7.	2.0	14
147	A novel knock-in mouse model of cryopyrin-associated periodic syndromes with development of amyloidosis: Therapeutic efficacy of proton pump inhibitors. Journal of Allergy and Clinical Immunology, 2020, 145, 368-378.e13.	2.9	14
148	Analysis of candidate genes for intrinsic neuropathy in a family with chronic idiopathic intestinal pseudo-obstruction. Clinical Genetics, 2001, 59, 131-133.	2.0	13
149	A novel missense mutation in the PHOX2Bgene is associated with late onset central hypoventilation syndrome. Pediatric Pulmonology, 2008, 43, 1036-1039.	2.0	13
150	The 423Q polymorphism of the Xâ€linked inhibitor of apoptosis gene influences monocyte function and is associated with periodic fever. Arthritis and Rheumatism, 2009, 60, 3476-3484.	6.7	13
151	Human lymphocytes assay: Cyclophosphamide metabolic activation by S9 system with low cytotoxicity. Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology, 1984, 130, 411-416.	0.4	12
152	Germ Line Mutations of the ret Proto-oncogene in Japanese Patients with Multiple Endocrine Neoplasia Type 2A and Type 2B. Japanese Journal of Cancer Research, 1994, 85, 879-882.	1.7	12
153	Nuclear Factor Y Drives Basal Transcription of the Human TLX3, a Gene Overexpressed in T-Cell Acute Lymphocytic Leukemia. Molecular Cancer Research, 2006, 4, 635-643.	3.4	12
154	Megacystis, megacolon, and malrotation: A new syndromic association?. American Journal of Medical Genetics, Part A, 2011, 155, 1798-1802.	1.2	12
155	Expression Variability and Function of the RET Gene in Adult Peripheral Blood Mononuclear Cells. Journal of Cellular Physiology, 2014, 229, 2027-2037.	4.1	12
156	Desogestrel down-regulates PHOX2B and its target genes in progesterone responsive neuroblastoma cells. Experimental Cell Research, 2018, 370, 671-679.	2.6	12
157	Novel ACTG2 variants disclose allelic heterogeneity and biâ€allelic inheritance in pediatric chronic intestinal pseudoâ€obstruction. Clinical Genetics, 2021, 99, 430-436.	2.0	12
158	Genetic screening of children with marrow failure. The role of primary Immunodeficiencies. American Journal of Hematology, 2021, 96, 1077-1086.	4.1	12
159	RAG deficiency with ALPS features successfully treated with TCRÎ±Î²/CD19 cell depleted haploidentical stem cell transplant. Clinical Immunology, 2018, 187, 102-103.	3.2	12
160	Ceftriaxone for Alexanderâ€™s Disease: A Four-Year Follow-Up. JIMD Reports, 2012, 9, 67-71.	1.5	11
161	An autoinflammatory neurological disease due to interleukin 6 hypersecretion. Journal of Neuroinflammation, 2013, 10, 29.	7.2	11
162	LPIN2 gene mutation in a patient with overlapping neutrophilic disease (pyoderma gangrenosum and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.8	11

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163	ADA2 deficiency due to a novel structural variation in 22q11.1. Clinical Genetics, 2019, 95, 732-733.	2.0	11
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