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	Recessive NLRC4-Autoinflammatory Disease Reveals an Ulcerative Colitis Locus. Journal of Clinical Immunology, 2022, 42, 325-335.	3.8	17
2	Recent advances in the developmental origin of neuroblastoma: an overview. Journal of Experimental and Clinical Cancer Research, 2022, 41, 92.	8.6	46
3	Generation of two hiPSC lines (UMILi027-A and UMILi028-A) from early and late-onset Congenital Central hypoventilation Syndrome (CCHS) patients carrying a polyalanine expansion mutation in the PHOX2B gene. Stem Cell Research, 2022, 61, 102781.	0.7	0
4	Progression of nonâ€hematologic manifestations in SAMD9Lâ€associated autoinflammatory disease (SAAD) after hematopoietic stem cell transplantation. Pediatric Allergy and Immunology, 2022, 33, .	2.6	4
5	Case Report: Atypical Manifestations Associated With FOXP3 Mutations. The â€œFil Rougeâ€•of Treg Between IPEX Features and Other Clinical Entities?. Frontiers in Immunology, 2022, 13, 854749.	4.8	6
6	Underlying Inborn Errors of Immunity in Patients With Evans Syndrome and Multilineage Cytopenias: A Single-Centre Analysis. Frontiers in Immunology, 2022, 13, .	4.8	7
7	Genotype-Phenotype Correlation and Functional Insights for Two Monoallelic TREX1 Missense Variants Affecting the Catalytic Core. Genes, 2022, 13, 1179.	2.4	2
8	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
9	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
10	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
11	Hemolysis and Neurologic Impairment in PAMI Syndrome: Novel Characteristics of an Elusive Disease. Pediatrics, 2021, 147, e20200784.	2.1	3
12	Widening the Neuroimaging Features of Adenosine Deaminase 2 Deficiency. American Journal of Neuroradiology, 2021, 42, 975-979.	2.4	10
13	Spectrum of Systemic Auto-Inflammatory Diseases in India: A Multi-Centric Experience. Frontiers in Immunology, 2021, 12, 630691.	4.8	11
14	Underlying CTLA4 Deficiency in a Patient With Juvenile Idiopathic Arthritis and Autoimmune Lymphoproliferative Syndrome Features Successfully Treated With Abataceptâ€”A Case Report. Journal of Pediatric Hematology/Oncology, 2021, 43, e1168-e1172.	0.6	5
15	A Common 3â€²UTR Variant of the PHOX2B Gene Is Associated With Infant Life-Threatening and Sudden Death Events in the Italian Population. Frontiers in Neurology, 2021, 12, 642735.	2.4	10
16	Congenital anomalies of the kidney and urinary tract in a cohort of 280 consecutive patients with Hirschsprung disease. Pediatric Nephrology, 2021, 36, 3151-3158.	1.7	8
17	The challenge of early diagnosis of autoimmune lymphoproliferative syndrome in children with suspected autoinflammatory/autoimmune disorders. Rheumatology, 2021, , .	1.9	4
18	The OSMR Gene Is Involved in Hirschsprung Associated Enterocolitis Susceptibility through an Altered Downstream Signaling. International Journal of Molecular Sciences, 2021, 22, 3831.	4.1	6

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19	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	7.6	28
20	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
21	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
22	Genetic screening of children with marrow failure. The role of primary Immunodeficiencies. American Journal of Hematology, 2021, 96, 1077-1086.	4.1	12
23	Targeted NGS Yields Plentiful Ultra-Rare Variants in Inborn Errors of Immunity Patients. Genes, 2021, 12, 1299.	2.4	8
24	Type I interferon activation in RAS-associated autoimmune leukoproliferative disease (RALD). Clinical Immunology, 2021, 231, 108837.	3.2	4
25	The Association among Pyoderma Gangrenosum, Ulcerative Colitis, and Hidradenitis Suppurativa and the Syndromic Hidradenitis Suppurativa Network: A Case Report. Skin Appendage Disorders, 2021, 7, 227-230.	1.0	1
26	Multidisciplinary study of sudden unexpected infant death in Liguria (Italy): a nine-year report. Minerva Pediatrics, 2021, 73, 435-443.	0.4	1
27	Case Report: Deficiency of Adenosine Deaminase 2 Presenting With Overlapping Features of Autoimmune Lymphoproliferative Syndrome and Bone Marrow Failure. Frontiers in Immunology, 2021, 12, 754029.	4.8	11
28	A Focus on Regulatory Networks Linking MicroRNAs, Transcription Factors and Target Genes in Neuroblastoma. Cancers, 2021, 13, 5528.	3.7	16
29	Alexander disease evolution over time: data from an Italian cohort of pediatric-onset patients. Molecular Genetics and Metabolism, 2021, 134, 353-358.	1.1	6
30	Multidisciplinary study of sudden unexpected infant death in Liguria (Italy): a nine-year report. Minerva Pediatrics, 2021, 73, .	0.4	1
31	Parental Somatic Mosaicism Uncovers Inheritance of an Apparently De Novo GFAP Mutation. Frontiers in Genetics, 2021, 12, 744068.	2.3	0
32	Beneficial Effect of Phenytoin and Carbamazepine on GFAP Gene Expression and Mutant GFAP Folding in a Cellular Model of Alexanderâ€™s Disease. Frontiers in Pharmacology, 2021, 12, 723218.	3.5	2
33	The Genetic Landscape of Patent Foramen Ovale: A Systematic Review. Genes, 2021, 12, 1953.	2.4	5
34	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	1.9	36
35	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
36	Causative and common PHOX2B variants define a broad phenotypic spectrum. Clinical Genetics, 2020, 97, 103-113.	2.0	39

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37	Unusual Late-onset Enteropathy in a Patient With Lipopolysaccharide-responsive Beige-like Anchor Protein Deficiency. Journal of Pediatric Hematology/Oncology, 2020, 42, e768-e771.	0.6	8
38	Guidelines for diagnosis and management of congenital central hypoventilation syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 252.	2.7	74
39	Targeted re-sequencing in pediatric and perinatal stroke. European Journal of Medical Genetics, 2020, 63, 104030.	1.3	9
40	Late-onset and long-lasting autoimmune neutropenia: an analysis from the Italian Neutropenia Registry. Blood Advances, 2020, 4, 5644-5649.	5.2	18
41	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
42	Alexander Disease Modeling in Zebrafish: An In Vivo System Suitable to Perform Drug Screening. Genes, 2020, 11, 1490.	2.4	2
43	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
44	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
45	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
46	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
47	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
48	PAPA and FMF in two siblings: possible amplification of clinical presentation? A case report. Italian Journal of Pediatrics, 2019, 45, 111.	2.6	7
49	Current practices for the genetic diagnosis of autoinflammatory diseases: results of a European Molecular Genetics Quality Network Survey. European Journal of Human Genetics, 2019, 27, 1502-1508.	2.8	10
50	When neonatal inflammation does not mean infection: an early-onset mevalonate kinase deficiency with interstitial lung disease. Clinical Immunology, 2019, 205, 25-28.	3.2	10
51	Classification criteria for autoinflammatory recurrent fevers. Annals of the Rheumatic Diseases, 2019, 78, 1025-1032.	0.9	300
52	ADA2 deficiency due to a novel structural variation in 22q11.1. Clinical Genetics, 2019, 95, 732-733.	2.0	11
53	Genetic Aspects of Investigating and Understanding Autoinflammation. , 2019, , 19-48.		3
54	AB1063â€…INTERSTITIAL LUNG DISEASE IN A NEWBORN AFFECTED BY MEVALONIC ACIDURIA. , 2019, , .		0

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55	THU0505â€…INTRINSIC AND EXTRINSIC B CELL DEFECT IN DADA2 PATIENTS. , 2019, , .		0
56	AB0974â€…A CASE OF ADENOSINE DEAMINASE 2 DEFICIENCY (DADA2) WITH AN UNCOMMON CLINICAL PRESENTATION AND RESPONSE TO IV IG. , 2019, , .		0
57	OP0106â€…A NOVEL KNOCK-IN MOUSE MODEL OF CAPS THAT DEVELOPS AMYLOIDOSIS: THERAPEUTIC EFFICACY OF PROTON PUMP INHIBITORS. , 2019, , .		0
58	FRI0568â€…THE USE OF NEXT GENERATION SEQUENCING PANEL IN UNDIFFERENTIATED AUTOINFLAMMATORY DISEASES IDENTIFY A SEPARATE SUBSET OF COLCHICINE-RESPONDER RECURRENT FEVERS DISTINCT FROM PFAPA SYNDROME. , 2019, , .		3
59	Thrombotic thrombocytopenic purpura and defective apoptosis due to CASP8/10 mutations: the role of mycophenolate mofetil. Blood Advances, 2019, 3, 3432-3435.	5.2	5
60	Copy number variations in candidate genomic regions confirm genetic heterogeneity and parental bias in Hirschsprung disease. Orphanet Journal of Rare Diseases, 2019, 14, 270.	2.7	3
61	A Novel Mutation of GFAP Causing Adult-Onset Alexander Disease. Frontiers in Neurology, 2019, 10, 1124.	2.4	7
62	RET in breast cancer: pathogenic implications and mechanisms of drug resistance. , 2019, 2, 1136-1152.		6
63	Secondary Autoimmune Neutropenia: Data from the Italian Neutropenia Registry. Blood, 2019, 134, 3585-3585.	1.4	0
64	LPIN2 gene mutation in a patient with overlapping neutrophilic disease (pyoderma gangrenosum and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.8	11
65	Medico-legal investigation in an explicable case of congenital central hypoventilation syndrome due to a rare variant of the PHOX2B gene. Journal of Clinical Forensic and Legal Medicine, 2018, 58, 1-5.	1.0	10
66	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
67	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
68	Novel spondyloepimetaphyseal dysplasia due to UFSP2 gene mutation. Clinical Genetics, 2018, 93, 671-674.	2.0	26
69	Structural and functional differences inPHOX2Bframeshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. Human Mutation, 2018, 39, 219-236.	2.5	28
70	Desogestrel down-regulates PHOX2B and its target genes in progesterone responsive neuroblastoma cells. Experimental Cell Research, 2018, 370, 671-679.	2.6	12
71	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
72	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

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73	FAS-Mediated Apoptosis Assay in Patients with ALPS-like Phenotype Carrying CASP10 Mutations. Blood, 2018, 132, 4960-4960.	1.4	0
74	Severe Chronic Neutropenia: Primary Immunodeficiency Mutations Are Frequent Causative Agents. Blood, 2018, 132, 2402-2402.	1.4	1
75	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
76	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
77	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
78	Autoinflammation in pyoderma gangrenosum and its syndromic form (pyoderma gangrenosum, acne) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.5	151
79	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
80	Mutations inMYO1Hcause a recessive form of central hypoventilation with autonomic dysfunction. Journal of Medical Genetics, 2017, 54, 754-761.	3.2	21
81	Custom Array Comparative Genomic Hybridization: the Importance of DNA Quality, an Expert Eye, and Variant Validation. International Journal of Molecular Sciences, 2017, 18, 609.	4.1	4
82	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
83	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
84	Targeting ofPHOX2Bexpression allows the identification of drugs effective in counteracting neuroblastoma cell growth. Oncotarget, 2017, 8, 72133-72146.	1.8	8
85	Chronic intestinal pseudoâ€obstruction in a child harboring a founder Hirschsprung RET mutation. American Journal of Medical Genetics, Part A, 2016, 170, 2400-2403.	1.2	5
86	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
87	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	2.9	38
88	Genetics of enteric neuropathies. Developmental Biology, 2016, 417, 198-208.	2.0	44
89	Neonatal-Onset Urticaria and Fever. Journal of Pediatrics, 2016, 177, 329-329.e1.	1.8	1
90	Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease. Neurological Sciences, 2016, 37, 973-977.	1.9	7

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91	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
92	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
93	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
94	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
95	Single-Lineage Bone Marrow Failure Driven By 2 Novel PI3KCD Mutations. Blood, 2016, 128, 1347-1347.	1.4	0
96	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
97	Prevalence of CECR1 mutations in pediatric patients with polyarteritis nodosa, livedo reticularis and/or stroke. Pediatric Rheumatology, 2015, 13, .	2.1	1
98	SAT0484â€...Prevalence of Cecr1 Mutations in Pediatric Patients with Polyarteritis Nodosa, Livedo Reticularis and/or Stroke. Annals of the Rheumatic Diseases, 2015, 74, 835.2-835.	0.9	1
99	A Next Generation Sequencing approach to the mutational screening of patients affected with systemic autoinflammatory disorders: diagnosis improvement and interpretation of complex clinical phenotypes. Pediatric Rheumatology, 2015, 13, .	2.1	0
100	SAT0001â€...Cryopyrin Associated Periodic Syndromes (CAPS): Investigations on Knock-In Mouse Model to Exploit Novel Approaches for the Modulation of the NLRP3 Inflammasome. Annals of the Rheumatic Diseases, 2015, 74, 650.1-650.	0.9	0
101	THU0528â€...Severe Erytrodermic Psoriasis and Arthritis as Clinical Presentation of a Card14-Mediated Pustular Psoriasis (CAMPS). Annals of the Rheumatic Diseases, 2015, 74, 391.2-391.	0.9	0
102	Cryopyrin associated periodic syndromes (CAPS): immunological characterization of knock-in mouse model to exploit novel approaches for the modulation of the NLRP3 inflammasome.. Pediatric Rheumatology, 2015, 13, .	2.1	0
103	Severe erytrodermic psoriasis and arthritis as clinical presentation of a CARD14-mediated psoriasis (CAMPS). Pediatric Rheumatology, 2015, 13, .	2.1	1
104	B cells characterization in ADA2 Deficiency patients. Pediatric Rheumatology, 2015, 13, .	2.1	1
105	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
106	Genetic and epigenetic factors affect RET gene expression in breast cancer cell lines and influence survival in patients. Annals of Oncology, 2015, 26, vi12.	1.2	0
107	210 Genetic and epigenetic factors affect RET gene expression in breast cancer cell lines and influence survival in patients. European Journal of Cancer, 2015, 51, S30.	2.8	0
108	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

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109	Identification of novel pathways and molecules able to down-regulate PHOX2B gene expression by in vitro drug screening approaches in neuroblastoma cells. Experimental Cell Research, 2015, 336, 43-57.	2.6	9
110	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
111	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
112	Recurrence of CCHS associated PHOX2B polyâ€alanine expansion mutation due to maternal mosaicism. Pediatric Pulmonology, 2014, 49, E45-7.	2.0	14
113	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
114	An autoinflammatory neurological disease due to interleukin 6 hypersecretion. Journal of Neuroinflammation, 2013, 10, 29.	7.2	11
115	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
116	Contribution of rare and common variants determine complex diseasesâ€"Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119
117	Pathways systematically associated to Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 187.	2.7	17
118	The involvement of the RET variant G691S in medullary thyroid carcinoma: Conflicting results of metaâ€analyses need to be reconciled. International Journal of Cancer, 2013, 133, 1760-1761.	5.1	2
119	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
120	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
121	A prospective observational study of associated anomalies in Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 184.	2.7	33
122	Magnetic Resonance Imaging â€œTigroid Patternâ€•in Alexander Disease. Neuropediatrics, 2013, 44, 174-176.	0.6	9
123	Failure of tocilizumab treatment in a CINCA patient: clinical and pathogenic implications. Rheumatology, 2013, 52, 1731-1732.	1.9	9
124	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
125	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). Annals of the Rheumatic Diseases, 2013, 72, 1044-1052.	0.9	69
126	Allele-Specific Expression at theRETLocus in Blood and Gut Tissue of Individuals Carrying Risk Alleles for Hirschsprung Disease. Human Mutation, 2013, 34, 754-762.	2.5	4

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127	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
128	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
129	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
130	Ceftriaxone for Alexanderâ€™s Disease: A Four-Year Follow-Up. JIMD Reports, 2012, 9, 67-71.	1.5	11
131	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
132	Guidelines for the genetic diagnosis of hereditary recurrent fevers. Annals of the Rheumatic Diseases, 2012, 71, 1599-1605.	0.9	160
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	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
135	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
136	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
137	Safe drugs to fight mutant protein overload and alpha-1-antitrypsin deficiency. Journal of Hepatology, 2011, 55, 949-950.	3.7	4
138	Toward a therapeutic strategy for polyalanine expansions disorders: In vivo and in vitro models for drugs analysis. European Journal of Paediatric Neurology, 2011, 15, 449-452.	1.6	6
139	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
140	Megacystis, megacolon, and malrotation: A new syndromic association?. American Journal of Medical Genetics, Part A, 2011, 155, 1798-1802.	1.2	12
141	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
142	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
143	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
144	The Ocular Motor Features of Adult-Onset Alexander Disease: A Case and Review of the Literature. Journal of Neuro-Ophthalmology, 2011, 31, 155-159.	0.8	9

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145	Correspondence Regarding: Alexander Disease Mutant Glial Fibrillary Acidic Protein Compromises Glutamate Transport in Astrocytes J Neuropathol Exp Neurol 2010;69:335â€“45. Journal of Neuropathology and Experimental Neurology, 2010, 69, 1270.1-1270.	1.7	2
146	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
147	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
148	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
149	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
150	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
151	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
152	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, 963.	3.7	1
153	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
154	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
155	Ceftriaxone has a therapeutic role in Alexander disease. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 416-417.	4.8	16
156	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	2.5	57
157	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
158	Congenital central hypoventilation syndrome from past to future: Model for translational and transitional autonomic medicine. Pediatric Pulmonology, 2009, 44, 521-535.	2.0	99
159	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
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