Isabella Ceccherini

List of Publications by Citations

242 papers	14,021 citations	50 h-index	115 g-index
270 ext. papers	15,889 ext. citations	5.8 avg, IF	5.45 L-index

#	Paper	IF	Citations
242	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). *Autophagy*, **2016**, 12, 1-222	10.2	3838
241	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
240	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. *Nature Genetics*, **1998**, 18, 171-3	36.3	659
239	Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. *Nature*, **1994**, 367, 377-8	50.4	659
238	Hirschsprung disease, associated syndromes and genetics: a review. *Journal of Medical Genetics*, **2008**, 45, 1-14	5.8	622
237	An official ATS clinical policy statement: Congenital central hypoventilation syndrome: genetic basis, diagnosis, and management. *American Journal of Respiratory and Critical Care Medicine*, **2010**, 181, 626-44	10.2	348
236	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, 373-80	5.8	205
235	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
234	Classification criteria for autoinflammatory recurrent fevers. *Annals of the Rheumatic Diseases*, **2019**, 78, 1025-1032	2.4	159
233	RET mutations in human disease. *Trends in Genetics*, **1996**, 12, 138-44	8.5	158
232	Stable length polymorphism of up to 260 kb at the tip of the short arm of human chromosome 16. *Cell*, **1991**, 64, 595-606	56.2	151
231	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	5.1	147
230	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	2.4	139
229	Adult-onset Alexander disease: a series of eleven unrelated cases with review of the literature. *Brain*, **2008**, 131, 2321-31	11.2	138
228	A diagnostic score for molecular analysis of hereditary autoinflammatory syndromes with periodic fever in children. *Arthritis and Rheumatism*, **2008**, 58, 1823-32		135
227	Guidelines for the genetic diagnosis of hereditary recurrent fevers. *Annals of the Rheumatic Diseases*, **2012**, 71, 1599-605	2.4	132
226	Clinical presentation and pathogenesis of cold-induced autoinflammatory disease in a family with recurrence of an NLRP12 mutation. *Arthritis and Rheumatism*, **2011**, 63, 830-9		131

225	Frequency of RET mutations in long- and short-segment Hirschsprung disease. *Human Mutation*, **1997**, 9, 243-9	4.7	125
224	Neutrophils from patients with TNFRSF1A mutations display resistance to tumor necrosis factor-induced apoptosis: pathogenetic and clinical implications. *Arthritis and Rheumatism*, **2006**, 54, 998-1008		121
223	Differentiating PFAPA syndrome from monogenic periodic fevers. *Pediatrics*, **2009**, 124, e721-8	7.4	109
222	Autoinflammation in pyoderma gangrenosum and its syndromic form (pyoderma gangrenosum, acne and suppurative hidradenitis). *British Journal of Dermatology*, **2017**, 176, 1588-1598	4	103
221	Distinct pathogenetic mechanisms for PHOX2B associated polyalanine expansions and frameshift mutations in congenital central hypoventilation syndrome. *Human Molecular Genetics*, **2005**, 14, 1815-24	5.6	99
220	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, 301-8	6.5	95
219	Nuclear run-on assay using biotin labeling, magnetic bead capture and analysis by fluorescence-based RT-PCR. *BioTechniques*, **2000**, 29, 1012-4, 1016-7	2.5	91
218	Contribution of rare and common variants determine complex diseases-Hirschsprung disease as a model. *Developmental Biology*, **2013**, 382, 320-9	3.1	90
217	Pathogenesis of Hirschsprung's disease. *Journal of Pediatric Surgery*, **2000**, 35, 1017-25	2.6	88
216	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	5.6	88
215	Congenital central hypoventilation syndrome from past to future: model for translational and transitional autonomic medicine. *Pediatric Pulmonology*, **2009**, 44, 521-35	3.5	87
214	Long-term clinical profile of children with the low-penetrance R92Q mutation of the TNFRSF1A gene. *Arthritis and Rheumatism*, **2011**, 63, 1141-50		85
213	Association of pyoderma gangrenosum, acne, and suppurative hidradenitis (PASH) shares genetic and cytokine profiles with other autoinflammatory diseases. *Medicine (United States)*, **2014**, 93, e187	1.8	83
212	Double heterozygosity for a RET substitution interfering with splicing and an EDNRB missense mutation in Hirschsprung disease. *American Journal of Human Genetics*, **1999**, 64, 1216-21	11	83
211	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
210	Can MR imaging diagnose adult-onset Alexander disease?. *American Journal of Neuroradiology*, **2008**, 29, 1190-6	4.4	80
209	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, O24	3.5	78
208	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,	3.5	78

207	Betaine, dimethyl sulfoxide, and 7-deaza-dGTP, a powerful mixture for amplification of GC-rich DNA sequences. *Journal of Molecular Diagnostics*, **2006**, 8, 544-50	5.1	78
206	Close linkage with the RET protooncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. *Human Molecular Genetics*, **1993**, 2, 1803-8	5.6	77
205	New workflow for classification of genetic variants' pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). *Journal of Medical Genetics*, **2018**, 55, 530-537	5.8	73
204	Interstitial deletion of the endothelin-B receptor gene in the spotting lethal (sl) rat. *Human Molecular Genetics*, **1995**, 4, 2089-96	5.6	72
203	Exon structure and flanking intronic sequences of the human RET proto-oncogene. *Biochemical and Biophysical Research Communications*, **1993**, 196, 1288-95	3.4	70
202	Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. *Journal of Internal Medicine*, **1998**, 243, 515-20	10.8	66
201	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, 201-4	5.2	66
200	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). *Annals of the Rheumatic Diseases*, **2013**, 72, 1044-52	2.4	62
199	Incidence of RET mutations in patients with Hirschsprung's disease. *Journal of Pediatric Surgery*, **2000**, 35, 139-42; discussion 142-3	2.6	61
198	MVK mutations and associated clinical features in Italian patients affected with autoinflammatory disorders and recurrent fever. *European Journal of Human Genetics*, **2005**, 13, 314-20	5.3	58
197	Clinical and genetic characterization of Italian patients affected by CINCA syndrome. *Rheumatology*, **2007**, 46, 473-8	3.9	56
196	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. *Genome Biology*, **2017**, 18, 48	18.3	55
195	Hirschsprung disease and congenital anomalies of the kidney and urinary tract (CAKUT): a novel syndromic association. *Medicine (United States)*, **2009**, 88, 83-90	1.8	53
194	Heterogeneity and low detection rate of RET mutations in Hirschsprung disease. *European Journal of Human Genetics*, **1994**, 2, 272-80	5.3	53
193	Clinical impact of MEFV mutations in children with periodic fever in a prevalent western European Caucasian population. *Annals of the Rheumatic Diseases*, **2012**, 71, 1961-5	2.4	51
192	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
191	A common haplotype at the 5' end of the RET proto-oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. *Human Mutation*, **2005**, 25, 189-95	4.7	44
190	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

189	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, 714-8	5.8	43
188	A web-based collection of genotype-phenotype associations in hereditary recurrent fevers from the Eurofever registry. *Orphanet Journal of Rare Diseases*, **2017**, 12, 167	4.2	42
187	Parental origin and somatic mosaicism of PHOX2B mutations in Congenital Central Hypoventilation Syndrome. *Human Mutation*, **2008**, 29, 206	4.7	42
186	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
185	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
184	Next-generation sequencing and its initial applications for molecular diagnosis of systemic auto-inflammatory diseases. *Annals of the Rheumatic Diseases*, **2016**, 75, 1550-7	2.4	41
183	Congenital central hypoventilation syndrome (CCHS) and sudden infant death syndrome (SIDS): kindred disorders of autonomic regulation. *Respiratory Physiology and Neurobiology*, **2008**, 164, 38-48	2.8	41
182	Genomic structure of the human lysosomal alpha-mannosidase gene (MANB). *Genomics*, **1997**, 42, 200-7	4.3	40
181	PHOX2B mutations and genetic predisposition to neuroblastoma. *Oncogene*, **2005**, 24, 3050-3	9.2	40
180	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
179	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		39
178	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, 205-9	5.8	38
177	The TLX2 homeobox gene is a transcriptional target of PHOX2B in neural-crest-derived cells. *Biochemical Journal*, **2006**, 395, 355-61	3.8	35
176	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	2.2	35
175	Specific haplotypes of the RET proto-oncogene are over-represented in patients with sporadic papillary thyroid carcinoma. *Journal of Medical Genetics*, **2002**, 39, 260-5	5.8	34
174	Gender-dependent disease severity in autosomal polycystic kidney disease of rats. *Kidney International*, **1995**, 48, 496-500	9.9	34
173	A common variant located in the 3'UTR of the RET gene is associated with protection from Hirschsprung disease. *Human Mutation*, **2007**, 28, 168-76	4.7	33
172	Molecular mechanisms of RET-induced Hirschsprung pathogenesis. *Annals of Medicine*, **2006**, 38, 11-9	1.5	33

171	PHOX2B-mediated regulation of ALK expression: in vitro identification of a functional relationship between two genes involved in neuroblastoma. *PLoS ONE*, **2010**, 5, e13108	3.7	32
170	In vitro treatments with ceftriaxone promote elimination of mutant glial fibrillary acidic protein and transcription down-regulation. *Experimental Cell Research*, **2010**, 316, 2152-65	4.2	29
169	Geldanamycin promotes nuclear localisation and clearance of PHOX2B misfolded proteins containing polyalanine expansions. *International Journal of Biochemistry and Cell Biology*, **2007**, 39, 327-39	5.6	29
168	Genetics of enteric neuropathies. *Developmental Biology*, **2016**, 417, 198-208	3.1	29
167	Low amounts of PHOX2B expanded alleles in asymptomatic parents suggest unsuspected recurrence risk in congenital central hypoventilation syndrome. *Journal of Molecular Medicine*, **2011** , 89, 505-13	5.5	28
166	Location of the first genetic locus, PKDr1, controlling autosomal dominant polycystic kidney disease in Han:SPRD cy/+ rat. *Human Molecular Genetics*, **1997**, 6, 609-13	5.6	28
165	Somatic in frame deletions not involving juxtamembranous cysteine residues strongly activate the RET proto-oncogene. *Oncogene*, **1997**, 14, 2609-12	9.2	28
164	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, 1211-5	5.3	27
163	In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins. *Neurobiology of Disease*, **2012**, 45, 508-18	7.5	27
162	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, 104-8	11.5	27
161	A prospective observational study of associated anomalies in Hirschsprung's disease. *Orphanet Journal of Rare Diseases*, **2013**, 8, 184	4.2	26
160	Autosomal dominant polycystic kidney disease: prenatal diagnosis by DNA analysis and sonography at 14 weeks. *Prenatal Diagnosis*, **1989**, 9, 751-8	3.2	25
159	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	7.5	24
158	Genomics approach to the analysis of bacterial communities dynamics in Hirschsprung's disease-associated enterocolitis: a pilot study. *Pediatric Surgery International*, **2010**, 26, 465-71	2.1	24
157	Diagnostic and therapeutic approach to multiple endocrine neoplasia type 2B in pediatric patients. *Pediatric Surgery International*, **2002**, 18, 378-83	2.1	24
156	Adult-onset Alexander disease : report on a family. *Journal of Neurology*, **2008**, 255, 24-30	5.5	23
155	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, 6719-30	4.8	23
154	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. *Human Molecular Genetics*, **2016**, 25, 5265-5275	5.6	23

153	GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. *Clinical Genetics*, **2007**, 72, 427-33	4	22
152	Clinical Characteristics of Patients Carrying the Q703K Variant of the NLRP3 Gene: A 10-year Multicentric National Study. *Journal of Rheumatology*, **2016**, 43, 1093-100	4.1	22
151	Structural and functional differences in PHOX2B frameshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. *Human Mutation*, **2018**, 39, 219-236	4.7	22
150	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	6.9	21
149	FAS-mediated apoptosis impairment in patients with ALPS/ALPS-like phenotype carrying variants on CASP10 gene. *British Journal of Haematology*, **2019**, 187, 502-508	4.5	21
148	An in vitro approach to test the possible role of candidate factors in the transcriptional regulation of the RET proto-oncogene. *Gene Expression*, **2005**, 12, 137-49	3.4	21
147	Guidelines for diagnosis and management of congenital central hypoventilation syndrome. *Orphanet Journal of Rare Diseases*, **2020**, 15, 252	4.2	21
146	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. *International Journal of Molecular Medicine*, **2002**, 10, 101-6	4.4	21
145	The involvement of the RET variant G691S in medullary thyroid carcinoma enlightened by a meta-analysis study. *International Journal of Cancer*, **2013**, 132, 2808-19	7.5	20
144	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, 483-90	5.3	20
143	Brainstem anomalies in two patients affected by congenital central hypoventilation syndrome. *American Journal of Respiratory and Critical Care Medicine*, **2006**, 174, 706-9	10.2	20
142	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, 1844-54	4.2	19
141	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	4.1	19
140	Glomerulocystic kidney disease in a family. *Nephrology Dialysis Transplantation*, **2002**, 17, 813-8	4.3	19
139	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. *Rheumatology*, **2020**, 59, 344-360	3.9	19
138	Causative and common PHOX2B variants define a broad phenotypic spectrum. *Clinical Genetics*, **2020**, 97, 103-113	4	19
137	Chromosome 21 scan in Down syndrome reveals DSCAM as a predisposing locus in Hirschsprung disease. *PLoS ONE*, **2013**, 8, e62519	3.7	18
136	Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A and PHOX2B genes. *European Journal of Human Genetics*, **2007**, 15, 848-55	5.3	18

135	Brainstem signs with progressing atrophy of medulla oblongata and upper cervical spinal cord. *Lancet Neurology, The*, **2007**, 6, 562-70	24.1	18
134	Genetic mapping of the RET protooncogene on rat chromosome 4. *Mammalian Genome*, **1995**, 6, 433-5	3.2	18
133	High-dose ustekinumab for severe childhood deficiency of interleukin-36 receptor antagonist (DITRA). *Annals of the Rheumatic Diseases*, **2018**, 77, 1241-1243	2.4	18
132	miR-204 mediates post-transcriptional down-regulation of PHOX2B gene expression in neuroblastoma cells. *Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms*, **2015**, 1849, 1057-65	6	17
131	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	5.5	17
130	Tumor necrosis factor receptor-associated periodic syndrome as a model linking autophagy and inflammation in protein aggregation diseases. *Journal of Molecular Medicine*, **2014**, 92, 583-94	5.5	17
129	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	4.2	17
128	Cell-line specific chromatin acetylation at the Sox10-Pax3 enhancer site modulates the RET proto-oncogene expression. *FEBS Letters*, **2002**, 523, 123-7	3.8	17
127	Genetic and epigenetic factors affect RET gene expression in breast cancer cell lines and influence survival in patients. *Oncotarget*, **2016**, 7, 26465-79	3.3	17
126	Congenital central hypoventilation syndrome: genotype-phenotype correlation in parents of affected children carrying a PHOX2B expansion mutation. *Clinical Genetics*, **2010**, 78, 289-93	4	16
125	Mutational analysis of the RNX gene in congenital central hypoventilation syndrome. *American Journal of Medical Genetics Part A*, **2002**, 113, 178-82		16
124	Induction of RET dependent and independent pro-inflammatory programs in human peripheral blood mononuclear cells from Hirschsprung patients. *PLoS ONE*, **2013**, 8, e59066	3.7	16
123	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	8.4	15
122	A novel mutation in the GFAP gene in a familial adult onset Alexander disease. *Journal of Neurology* , **2007**, 254, 1278-80	5.5	15
121	Rescue of human RET gene expression by sodium butyrate: a novel powerful tool for molecular studies in Hirschsprung disease. *Gut*, **2003**, 52, 1154-8	19.2	15
120	Caffeine post-treatment causes a shift in the chromosome aberration types induced by mitomycin C, suggesting a caffeine-sensitive mechanism of DNA repair in G2. *Mutagenesis*, **1988**, 3, 39-44	2.8	15
119	Pathways systematically associated to Hirschsprung's disease. *Orphanet Journal of Rare Diseases*, **2013**, 8, 187	4.2	14
118	Mutations in cause a recessive form of central hypoventilation with autonomic dysfunction. *Journal of Medical Genetics*, **2017**, 54, 754-761	5.8	14

117	The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome-associated polyalanine-expanded PHOX2B. *Journal of Molecular Medicine*, **2012**, 90, 1025-35	5.5	14
116	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, 462-70	5.3	14
115	Dysregulation in B-cell responses and T follicular helper cell function in ADA2 deficiency patients. *European Journal of Immunology*, **2021**, 51, 206-219	6.1	14
114	Novel spondyloepimetaphyseal dysplasia due to UFSP2 gene mutation. *Clinical Genetics*, **2018**, 93, 671-674	4	13
113	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	3.5	13
112	Search for pathogenetic variants of the SPRY2 gene in intestinal innervation defects. *Internal Medicine Journal*, **2009**, 39, 335-7	1.6	13
111	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, 1378-84	4.1	13
110	Oligogenic inheritance in neuroblastoma. *Cancer Letters*, **2005**, 228, 65-9	9.9	13
109	A novel polymorphic AP-1 binding element of the GFAP promoter is associated with different allelic transcriptional activities. *Annals of Human Genetics*, **2010**, 74, 506-15	2.2	12
108	Ceftriaxone has a therapeutic role in Alexander disease. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2010**, 34, 416-7	5.5	12
107	The 423Q polymorphism of the X-linked inhibitor of apoptosis gene influences monocyte function and is associated with periodic fever. *Arthritis and Rheumatism*, **2009**, 60, 3476-84		12
106	A novel missense mutation in the PHOX2B gene is associated with late onset central hypoventilation syndrome. *Pediatric Pulmonology*, **2008**, 43, 1036-9	3.5	12
105	A young female with early onset arthritis, uveitis, hepatic, and renal granulomas: a clinical tryst with Blau syndrome over 20 years and case-based review. *Rheumatology International*, **2021**, 41, 173-181	3.6	12
104	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	2.2	12
103	Recurrence of CCHS associated PHOX2B poly-alanine expansion mutation due to maternal mosaicism. *Pediatric Pulmonology*, **2014**, 49, E45-7	3.5	11
102	Megacystis, megacolon, and malrotation: a new syndromic association?. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1798-802	2.5	11
101	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, 879-82		11
100	Human lymphocytes assay: cyclophosphamide metabolic activation by S9 system with low cytotoxicity. *Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology*, **1984**, 130, 411-6		11

99	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	3.4	10
98	An autoinflammatory neurological disease due to interleukin 6 hypersecretion. *Journal of Neuroinflammation*, **2013**, 10, 29	10.1	10
97	Nuclear factor Y drives basal transcription of the human TLX3, a gene overexpressed in T-cell acute lymphocytic leukemia. *Molecular Cancer Research*, **2006**, 4, 635-43	6.6	10
96	Analysis of candidate genes for intrinsic neuropathy in a family with chronic idiopathic intestinal pseudo-obstruction. *Clinical Genetics*, **2001**, 59, 131-3	4	10
95	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	5.3	9
94	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	4.2	9
93	Expression variability and function of the RET gene in adult peripheral blood mononuclear cells. *Journal of Cellular Physiology*, **2014**, 229, 2027-37	7	9
92	Uncommon association of germline mutations of RET proto-oncogene and CDKN2A gene. *European Journal of Endocrinology*, **2008**, 158, 417-22	6.5	9
91	Hirschsprung associated GDNF mutations do not prevent RET activation. *European Journal of Human Genetics*, **2002**, 10, 183-7	5.3	9
90	RAG deficiency with ALPS features successfully treated with TCR$\alpha\beta$/CD19 cell depleted haploidentical stem cell transplant. *Clinical Immunology*, **2018**, 187, 102-103	9	9
89	ADA2 deficiency due to a novel structural variation in 22q11.1. *Clinical Genetics*, **2019**, 95, 732-733	4	8
88	Desogestrel down-regulates PHOX2B and its target genes in progesterone responsive neuroblastoma cells. *Experimental Cell Research*, **2018**, 370, 671-679	4.2	8
87	Ceftriaxone for Alexander's Disease: A Four-Year Follow-Up. *JIMD Reports*, **2013**, 9, 67-71	1.9	8
86	cDNA sequence and genomic structure of the rat RET proto-oncogene. *DNA Sequence*, **2000**, 11, 405-17		8
85	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. *Brain*, **2021**, 144, 1451-1466	11.2	8
84	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	1.7	7
83	The ocular motor features of adult-onset alexander disease: a case and review of the literature. *Journal of Neuro-Ophthalmology*, **2011**, 31, 155-9	2.6	7
82	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

81	Comparative genomic sequence analysis coupled to chromatin immunoprecipitation: a screening procedure applied to search for regulatory elements at the RET locus. *Physiological Genomics*, **2005**, 23, 269-74	3.6	7
80	Late-onset and long-lasting autoimmune neutropenia: an analysis from the Italian Neutropenia Registry. *Blood Advances*, **2020**, 4, 5644-5649	7.8	7
79	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. *Brain*, **2021**, 144, 1422-1434	11.2	7
78	When neonatal inflammation does not mean infection: an early-onset mevalonate kinase deficiency with interstitial lung disease. *Clinical Immunology*, **2019**, 205, 25-28	9	6
77	gene mutation in a patient with overlapping neutrophilic disease (pyoderma gangrenosum and aseptic abscess syndrome). *JAAD Case Reports*, **2018**, 4, 120-122	1.4	6
76	ABCC6 mutations and early onset stroke: Two cases of a typical Pseudoxanthoma Elasticum. *European Journal of Paediatric Neurology*, **2018**, 22, 725-728	3.8	6
75	Magnetic resonance imaging "tigroid pattern" in Alexander disease. *Neuropediatrics*, **2013**, 44, 174-6	1.6	6
74	Failure of tocilizumab treatment in a CINCA patient: clinical and pathogenic implications. *Rheumatology*, **2013**, 52, 1731-2	3.9	6
73	Treatment of preclinical medullary thyroid carcinoma in MEN 2A gene carrier. *Lancet, The*, **1994**, 344, 1084-5	40	6
72	Kinetics of chromosomal aberrations and first mitosis division in human lymphocytes exposed to mitomycin C. *Mutation Research-Fundamental and Molecular Mechanisms of Mutagenesis*, **1988**, 208, 183-8		6
71	Targeting of expression allows the identification of drugs effective in counteracting neuroblastoma cell growth. *Oncotarget*, **2017**, 8, 72133-72146	3.3	6
70	A Focus on Regulatory Networks Linking MicroRNAs, Transcription Factors and Target Genes in Neuroblastoma. *Cancers*, **2021**, 13,	6.6	6
69	A Common 3'UTR Variant of the Gene Is Associated With Infant Life-Threatening and Sudden Death Events in the Italian Population. *Frontiers in Neurology*, **2021**, 12, 642735	4.1	6
68	Toward a therapeutic strategy for polyalanine expansions disorders: in vivo and in vitro models for drugs analysis. *European Journal of Paediatric Neurology*, **2011**, 15, 449-52	3.8	5
67	The Ret proto-oncogene in the WAG/Rij rat strain: an animal model for inherited C-cell carcinoma?. *Laboratory Animals*, **2003**, 37, 215-21	2.6	5
66	The Molecular Genetics of Hirschsprung□ Disease **2008**, 63-78		5
65	Novel ACTG2 variants disclose allelic heterogeneity and bi-allelic inheritance in pediatric chronic intestinal pseudo-obstruction. *Clinical Genetics*, **2021**, 99, 430-436	4	5
64	PAPA and FMF in two siblings: possible amplification of clinical presentation? A case report. *Italian Journal of Pediatrics*, **2019**, 45, 111	3.2	4

63	Custom Array Comparative Genomic Hybridization: the Importance of DNA Quality, an Expert Eye, and Variant Validation. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	4
62	Allele-specific expression at the RET locus in blood and gut tissue of individuals carrying risk alleles for Hirschsprung disease. *Human Mutation*, **2013**, 34, 754-62	4.7	4
61	Safe drugs to fight mutant protein overload and alpha-1-antitrypsin deficiency. *Journal of Hepatology*, **2011**, 55, 949-50	13.4	4
60	Unusual Late-onset Enteropathy in a Patient With Lipopolysaccharide-responsive Beige-like Anchor Protein Deficiency. *Journal of Pediatric Hematology/Oncology*, **2020**, 42, e768-e771	1.2	4
59	Targeted re-sequencing in pediatric and perinatal stroke. *European Journal of Medical Genetics*, **2020**, 63, 104030	2.6	4
58	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotype-phenotype correlation in congenital central hypoventilation syndrome (CCHS). *Genetics in Medicine*, **2021**, 23, 1656-1663	8.1	4
57	Recent advances in the developmental origin of neuroblastoma: an overview.. *Journal of Experimental and Clinical Cancer Research*, **2022**, 41, 92	12.8	4
56	Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease. *Neurological Sciences*, **2016**, 37, 973-7	3.5	3
55	An intronic nucleotide variant of the RET proto-oncogene causes Hirschsprung disease by interfering with RNA splicing. *Gene Function & Disease*, **2000**, 1, 184-188		3
54	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	3.5	3
53	The challenge of early diagnosis of autoimmune lymphoproliferative syndrome in children with suspected autoinflammatory/autoimmune disorders. *Rheumatology*, **2021**,	3.9	3
52	Genetic screening of children with marrow failure. The role of primary Immunodeficiencies. *American Journal of Hematology*, **2021**, 96, 1077-1086	7.1	3
51	FRI0568 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
50	Thrombotic thrombocytopenic purpura and defective apoptosis due to CASP8/10 mutations: the role of mycophenolate mofetil. *Blood Advances*, **2019**, 3, 3432-3435	7.8	3
49	Copy number variations in candidate genomic regions confirm genetic heterogeneity and parental bias in Hirschsprung disease. *Orphanet Journal of Rare Diseases*, **2019**, 14, 270	4.2	3
48	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	11.5	3
47	Widening the Neuroimaging Features of Adenosine Deaminase 2 Deficiency. *American Journal of Neuroradiology*, **2021**, 42, 975-979	4.4	3
46	Genetic Aspects of Investigating and Understanding Autoinflammation **2019**, 19-48		2

45	The involvement of the RET variant G691S in medullary thyroid carcinoma: conflicting results of meta-analyses need to be reconciled. *International Journal of Cancer*, **2013**, 133, 1760-1	7.5	2
44	Correspondence regarding: Alexander disease mutant glial fibrillary acidic protein compromises glutamate transport in astrocytes. *Journal of Neuropathology and Experimental Neurology*, **2010**, 69, 1270; author reply 1270-1	3.1	2
43	Incorporating prior biological information in linkage studies increases power and limits multiple testing. *BMC Proceedings*, **2007**, 1 Suppl 1, S89	2.3	2
42	Replication pattern of the pericentromeric region of chromosome 10q and expression of the RET protooncogene. *Experimental Cell Research*, **2004**, 298, 602-10	4.2	2
41	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. *PLoS Genetics*, **2020**, 16, e1009106	6	2
40	Recessive NLRC4-Autoinflammatory Disease Reveals an Ulcerative Colitis Locus. *Journal of Clinical Immunology*, **2021**, 1	5.7	2
39	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	8.4	2
38	RET in breast cancer: pathogenic implications and mechanisms of drug resistance. **2019**, 2, 1136-1152		2
37	Underlying CTLA4 Deficiency in a Patient With Juvenile Idiopathic Arthritis and Autoimmune Lymphoproliferative Syndrome Features Successfully Treated With Abatacept-A Case Report. *Journal of Pediatric Hematology/Oncology*, **2021**, 43, e1168-e1172	1.2	2
36	Chronic intestinal pseudo-obstruction in a child harboring a founder Hirschsprung RET mutation. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2400-3	2.5	2
35	Neonatal-Onset Urticaria and Fever. *Journal of Pediatrics*, **2016**, 177, 329-329.e1	3.6	1
34	Severe erytrodermic psoriasis and arthritis as clinical presentation of a CARD14-mediated psoriasis (CAMPS). *Pediatric Rheumatology*, **2015**, 13,	3.5	1
33	Recurrent arthritis as a unique manifestation of hyperimmunoglobulinaemia D. *Rheumatology*, **2009**, 48, 199-201	3.9	1
32	The immediate upstream sequence of the mouse Ret gene controls tissue-specific expression in transgenic mice. *International Journal of Molecular Medicine*, **2006**, 18, 601	4.4	1
31	Cell-line specific transcription rates of the RET gene and functional domains in its minimal promoter. *Gene Function & Disease*, **2001**, 1, 145-153		1
30	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. *International Journal of Molecular Medicine*, **2002**, 10, 101	4.4	1
29	Strategies for the Identification of Intron-Exon Boundaries and Point Mutations: The Example of the RET Proto-Oncogene. *Methods*, **1996**, 9, 98-105	4.6	1
28	Radiation hybrids for mapping and cloning DNA sequences of distal 16p. *Somatic Cell and Molecular Genetics*, **1992**, 18, 319-24		1

27	Multidisciplinary study of sudden unexpected infant death in Liguria (Italy): a nine-year report. *Minerva Pediatrics*, **2021**, 73, 435-443	1.5	1
26	Spectrum of Systemic Auto-Inflammatory Diseases in India: A Multi-Centric Experience. *Frontiers in Immunology*, **2021**, 12, 630691	8.4	1
25	Targeted NGS Yields Plentiful Ultra-Rare Variants in Inborn Errors of Immunity Patients. *Genes*, **2021**, 12,	4.2	1
24	Type I interferon activation in RAS-associated autoimmune leukoproliferative disease (RALD). *Clinical Immunology*, **2021**, 231, 108837	9	1
23	Cell-line specific transcription rates of theRET gene and functional domains in its minimal promoter. *Gene Function & Disease*, **2000**, 1, 1-9		1
22	The Genetic Landscape of Patent Foramen Ovale: A Systematic Review.. *Genes*, **2021**, 12,	4.2	1
21	Congenital anomalies of the kidney and urinary tract in a cohort of 280 consecutive patients with Hirschsprung disease. *Pediatric Nephrology*, **2021**, 36, 3151-3158	3.2	0
20	A Novel Mutation of Causing Adult-Onset Alexander Disease. *Frontiers in Neurology*, **2019**, 10, 1124	4.1	0
19	Hemolysis and Neurologic Impairment in PAMI Syndrome: Novel Characteristics of an Elusive Disease. *Pediatrics*, **2021**, 147,	7.4	0
18	Progression of non-hematologic manifestations in SAMD9L-associated autoinflammatory disease (SAAD) after hematopoietic stem cell transplantation. *Pediatric Allergy and Immunology*, **2021**,	4.2	0
17	Beneficial Effect of Phenytoin and Carbamazepine on Gene Expression and Mutant GFAP Folding in a Cellular Model of Alexander's Disease.. *Frontiers in Pharmacology*, **2021**, 12, 723218	5.6	0
16	Case Report: Atypical Manifestations Associated With FOXP3 Mutations. The "Fil Rouge" of Treg Between IPEX Features and Other Clinical Entities?. *Frontiers in Immunology*, **2022**, 13, 854749	8.4	0
15	SAT0484 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	2.4	
14	SAT0001 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	2.4	
13	THU0528 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	2.4	
12	Assignment of mouse Gfra1, the homologue of a new human HSCR candidate gene, to the telomeric region of mouse chromosome 19. *Cytogenetic and Genome Research*, **1997**, 78, 291-4	1.9	
11	PHOX2A and PHOX2B genes are highly co-expressed in human neuroblastoma **1992**, 33, 985		
10	Molecular and cytogenetic characterization of radiation hybrids specific for human chromosome 16. *Cytogenetic and Genome Research*, **1992**, 61, 167-71	1.9	

9	In vitro studies of PHOX2B gene mutations in congenital central hypoventilation syndrome **2008**, 71-83	
8	FAS-Mediated Apoptosis Assay in Patients with ALPS-like Phenotype Carrying CASP10 Mutations. *Blood*, **2018**, 132, 4960-4960	2.2
7	Severe Chronic Neutropenia: Primary Immunodeficiency Mutations Are Frequent Causative Agents. *Blood*, **2018**, 132, 2402-2402	2.2
6	Secondary Autoimmune Neutropenia: Data from the Italian Neutropenia Registry. *Blood*, **2019**, 134, 3585-3585	2.2
5	Single-Lineage Bone Marrow Failure Driven By 2 Novel PI3KCD Mutations. *Blood*, **2016**, 128, 1347-1347	2.2
4	Functional characterization of a minimal sequence essential for the expression of human TLX2 gene. *BMB Reports*, **2009**, 42, 788-93	5.5
3	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	1.4
2	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	1.6
1	Parental Somatic Mosaicism Uncovers Inheritance of an Apparently Mutation.. *Frontiers in Genetics*, **2021**, 12, 744068	4.5