Claudio Pignata

List of Publications by Year in Descending Order

209 papers	5,973 citations	42 h-index	69 g-index
226 ext. papers	6,912 ext. citations	5 avg, IF	4.97 L-index

#	Paper	IF	Citations
209	T Cell Immunodeficiency, Congenital Alopecia, and Nail Dystrophy **2022**, 1-6		
208	DiGeorge Syndrome **2022**, 1-7		
207	Clinical Manifestations of 22q11.2 Deletion Syndrome. *Heart Failure Clinics*, **2022**, 18, 155-164	3.3	4
206	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination.. *Frontiers in Immunology*, **2022**, 13, 845496	8.4	0
205	Progressive Depletion of B and T Lymphocytes in Patients with Ataxia Telangiectasia: Results of the Italian Primary Immunodeficiency Network.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
204	Mechanisms of immune tolerance breakdown in inborn errors of immunity **2022**, 73-95		
203	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. *Archivos De Bronconeumologia*, **2021**, 57, 712-714	0.7	
202	Clinical outcome, incidence, and SARS-CoV-2 infection-fatality rates in Italian patients with inborn errors of immunity. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 2904-2906.e2	5.4	24
201	In Ataxia-Telangiectasia, Oral Betamethasone Administration Ameliorates Lymphocytes Functionality through Modulation of the IL-7/IL-7R Axis Paralleling the Neurological Behavior: A Comparative Report of Two Cases. *Immunological Investigations*, **2021**, 50, 295-303	2.9	2
200	Complement system network in cell physiology and in human diseases. *International Reviews of Immunology*, **2021**, 40, 159-170	4.6	4
199	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. *Archivos De Bronconeumologia*, **2021**, 57, 712-712	0.7	1
198	Growth Hormone Receptor (Ghr) 6 Pseudoexon Activation: A Novel Cause Of Severe Growth Hormone Insensitivity (Ghi). *Journal of Clinical Endocrinology and Metabolism*, **2021**,	5.6	2
197	SARS-CoV-2 Infection in the Immunodeficient Host: Necessary and Dispensable Immune Pathways. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 3237-3248	5.4	1
196	Simple Measurement of IgA Predicts Immunity and Mortality in Ataxia-Telangiectasia. *Journal of Clinical Immunology*, **2021**, 41, 1878-1892	5.7	2
195	Expanding the Nude SCID/CID Phenotype Associated with FOXN1 Homozygous, Compound Heterozygous, or Heterozygous Mutations. *Journal of Clinical Immunology*, **2021**, 41, 756-768	5.7	4
194	Long-term follow-up of 168 patients with X-linked agammaglobulinemia reveals increased morbidity and mortality. *Journal of Allergy and Clinical Immunology*, **2020**, 146, 429-437	11.5	35
193	Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1

192	Clinical Phenotype, Immunological Abnormalities, and Genomic Findings in Patients with DiGeorge Spectrum Phenotype without 22q11.2 Deletion. *Journal of Allergy and Clinical Immunology: in Practice*, **2020**, 8, 3112-3120	5.4	4
191	Health-Related Quality of Life and Emotional Difficulties in Chronic Granulomatous Disease: Data on Adult and Pediatric Patients from Italian Network for Primary Immunodeficiency (IPINet). *Journal of Clinical Immunology*, **2020**, 40, 289-298	5.7	7
190	The Italian Registry for Primary Immunodeficiencies (Italian Primary Immunodeficiency Network; IPINet): Twenty Years of Experience (1999-2019). *Journal of Clinical Immunology*, **2020**, 40, 1026-1037	5.7	6
189	T-Cell Immunodeficiencies With Congenital Alterations of Thymic Development: Genes Implicated and Differential Immunological and Clinical Features. *Frontiers in Immunology*, **2020**, 11, 1837	8.4	9
188	Consensus of the Italian Primary Immunodeficiency Network on transition management from pediatric to adult care in patients affected with childhood-onset inborn errors of immunity. *Journal of Allergy and Clinical Immunology*, **2020**, 146, 967-983	11.5	0
187	Clinical, Immunological, and Molecular Features of Typical and Atypical Severe Combined Immunodeficiency: Report of the Italian Primary Immunodeficiency Network. *Frontiers in Immunology*, **2019**, 10, 1908	8.4	19
186	Heterozygous FOXN1 Variants Cause Low TRECs and Severe T Cell Lymphopenia, Revealing a Crucial Role of FOXN1 in Supporting Early Thymopoiesis. *American Journal of Human Genetics*, **2019** , 105, 549-561	11	28
185	Lentiviral gene therapy corrects platelet phenotype and function in patients with Wiskott-Aldrich syndrome. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 825-838	11.5	29
184	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. *Clinical Genetics*, **2019**, 96, 169-175	4	16
183	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. *Frontiers in Immunology*, **2019**, 10, 316	8.4	22
182	X-linked agammaglobulinemia (XLA):Phenotype, diagnosis, and therapeutic challenges around the world. *World Allergy Organization Journal*, **2019**, 12, 100018	5.2	53
181	Immunophenotype Anomalies Predict the Development of Autoimmune Cytopenia in 22q11.2 Deletion Syndrome. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 2369-2376	5.4	22
180	Autosomal-dominant hyper-IgE syndrome is associated with appearance of infections early in life and/or neonatal rash: Evidence from the Italian cohort of 61 patients with elevated IgE. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 2072-2075.e4	5.4	4
179	Subcutaneous Immunoglobulin Twenty Percent Every Two Weeks in Pediatric Patients with Primary Immunodeficiencies: Subcohort Analysis of the IBIS Study. *Pediatric, Allergy, Immunology, and Pulmonology*, **2019**, 32, 70-75	0.8	2
178	Oral Thrush and Onychomycosis **2019**, 371-376		
177	Recurrent Cold Suppurative Granulomatous Lymphadenitis **2019**, 347-352		
176	A case of incontinentia pigmenti associated with congenital absence of portal vein system and nodular regenerative hyperplasia. *British Journal of Dermatology*, **2019**, 180, 674-675	4	1
175	Follow-up and outcome of symptomatic partial or absolute IgA deficiency in children. *European Journal of Pediatrics*, **2019**, 178, 51-60	4.1	10

174	Minimum effective betamethasone dosage on the neurological phenotype in patients with ataxia-telangiectasia: a multicenter observer-blind study. *European Journal of Neurology*, **2018**, 25, 833-840	6	9
173	Impaired platelet activation in patients with hereditary deficiency of p47. *British Journal of Haematology*, **2018**, 180, 454-456	4.5	5
172	Neutralizing Anti-Cytokine Autoantibodies Against Interferon-□n Immunodysregulation Polyendocrinopathy Enteropathy X-Linked. *Frontiers in Immunology*, **2018**, 9, 544	8.4	19
171	Biweekly Hizentra□ in Primary Immunodeficiency: a Multicenter, Observational Cohort Study (IBIS). *Journal of Clinical Immunology*, **2018**, 38, 602-609	5.7	7
170	Otolaryngological features in a cohort of patients affected with 22q11.2 deletion syndrome: A monocentric survey. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2128-2134	2.5	6
169	Direct and Indirect Costs of Immunoglobulin Replacement Therapy in Patients with Common Variable Immunodeficiency (CVID) and X-Linked Agammaglobulinemia (XLA) in Italy. *Clinical Drug Investigation*, **2018**, 38, 955-965	3.2	1
168	Impaired natural killer cell functions in patients with signal transducer and activator of transcription 1 (STAT1) gain-of-function mutations. *Journal of Allergy and Clinical Immunology*, **2017** , 140, 553-564.e4	11.5	33
167	DiGeorge-like syndrome in a child with a 3p12.3 deletion involving MIR4273 gene born to a mother with gestational diabetes mellitus. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1913-1918	2.5	6
166	Recommendations regarding splenectomy in hereditary hemolytic anemias. *Haematologica*, **2017**, 102, 1304-1313	6.6	77
165	Abnormal cell-clearance and accumulation of autophagic vesicles in lymphocytes from patients affected with Ataxia-Teleangiectasia. *Clinical Immunology*, **2017**, 175, 16-25	9	16
164	NADPH Oxidase Deficiency: A Multisystem Approach. *Oxidative Medicine and Cellular Longevity*, **2017**, 2017, 4590127	6.7	21
163	FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. *Journal of Clinical Immunology*, **2017**, 37, 751-758	5.7	17
162	Brain abscesses in children: an Italian multicentre study. *Epidemiology and Infection*, **2017**, 145, 2848-2855	4.3	13
161	Two Brothers with Atypical Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. *Frontiers in Immunology*, **2017**, 8, 1892	8.4	6
160	Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. *International Reviews of Immunology*, **2016**, 35, 25-38	4.6	6
159	Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease. *Data in Brief*, **2016**, 7, 311-5	1.2	9
158	Clinical heterogeneity of dominant chronic mucocutaneous candidiasis disease: presenting as treatment-resistant candidiasis and chronic lung disease. *Clinical Immunology*, **2016**, 164, 1-9	9	22
157	Long-term effects of growth hormone (GH) replacement therapy on hematopoiesis in a large cohort of children with GH deficiency. *Endocrine*, **2016**, 53, 192-8	4	8

156	A Bronchovascular Anomaly in a Patient With 22q11.2 Deletion Syndrome. *Journal of Investigational Allergology and Clinical Immunology*, **2016**, 26, 390-392	2.3	2
155	Wiskott-Aldrich Syndrome: A Retrospective Study on 575 Patients Analyzing the Impact of Splenectomy, Stem Cell Transplantation, or No Definitive Treatment on Frequency of Disease-Related Complications and Physician-Perceived Quality of Life. *Blood*, **2016**, 128, 366-366	2.2	1
154	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. *Frontiers in Immunology*, **2016**, 7, 466	8.4	44
153	Novel Findings into AIRE Genetics and Functioning: Clinical Implications. *Frontiers in Pediatrics*, **2016** , 4, 86	3.4	21
152	Unbalanced Immune System: Immunodeficiencies and Autoimmunity. *Frontiers in Pediatrics*, **2016**, 4, 107	3.4	16
151	Novel STAT1 gain-of-function mutation and suppurative infections. *Pediatric Allergy and Immunology*, **2016**, 27, 220-3	4.2	11
150	Targeted next-generation sequencing revealed MYD88 deficiency in a child with chronic yersiniosis and granulomatous lymphadenitis. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 1591-1595.e4	11.5	7
149	A mutation in caspase-9 decreases the expression of BAFFR and ICOS in patients with immunodeficiency and lymphoproliferation. *Genes and Immunity*, **2015**, 16, 151-61	4.4	4
148	Thymic stromal alterations and genetic disorders of immune system. *Frontiers in Immunology*, **2015**, 6, 81	8.4	4
147	Severe combined immunodeficiency--an update. *Annals of the New York Academy of Sciences*, **2015**, 1356, 90-106	6.5	54
146	Nutritional Status in Agammaglobulinemia: An Italian Multicenter Study. *Journal of Clinical Immunology*, **2015**, 35, 595-7	5.7	1
145	B cells from nuclear factor kB essential modulator deficient patients fail to differentiate to antibody secreting cells in response to TLR9 ligand. *Clinical Immunology*, **2015**, 161, 131-5	9	4
144	Clinical features and follow-up in patients with 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2014**, 164, 1475-80.e2	3.6	83
143	The PedPAD study: boys predominate in the hypogammaglobulinaemia registry of the ESID online database. *Clinical and Experimental Immunology*, **2014**, 176, 387-93	6.2	11
142	FOXN1 in organ development and human diseases. *International Reviews of Immunology*, **2014**, 33, 83-93	4.6	35
141	Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. *Scandinavian Journal of Gastroenterology*, **2014**, 49, 274-9	2.4	25
140	Insight into IKBKG/NEMO locus: report of new mutations and complex genomic rearrangements leading to incontinentia pigmenti disease. *Human Mutation*, **2014**, 35, 165-77	4.7	57
139	Different degrees of NADPH oxidase 2 regulation and in vivo platelet activation: lesson from chronic granulomatous disease. *Journal of the American Heart Association*, **2014**, 3, e000920	6	28

138	Cutaneous vasculitis in patients with autoimmune polyendocrine syndrome type 1: report of a case and brief review of the literature. *BMC Pediatrics*, **2014**, 14, 272	2.6	9
137	High-content cytometry and transcriptomic biomarker profiling of human B-cell activation. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 172-80.e1-10	11.5	11
136	Intergenerational and intrafamilial phenotypic variability in 22q11.2 deletion syndrome subjects. *BMC Medical Genetics*, **2014**, 15, 1	2.1	28
135	The R156H variation in IL-12R is not a mutation. *Italian Journal of Pediatrics*, **2013**, 39, 12	3.2	1
134	Hypertransaminasemia and fatal lung disease: a case report. *Italian Journal of Pediatrics*, **2013**, 39, 9	3.2	0
133	Non-autoimmune subclinical hypothyroidism due to a mutation in TSH receptor: report on two brothers. *Italian Journal of Pediatrics*, **2013**, 39, 5	3.2	5
132	Treatment of children with chronic viral hepatitis: what is available and what is in store. *World Journal of Pediatrics*, **2013**, 9, 212-20	4.6	5
131	Steroid treatment in Ataxia-Telangiectasia induces alterations of functional magnetic resonance imaging during prono-supination task. *European Journal of Paediatric Neurology*, **2013**, 17, 135-40	3.8	18
130	Human skin-derived keratinocytes and fibroblasts co-cultured on 3D poly caprolactone scaffold support in vitro HSC differentiation into T-lineage committed cells. *International Immunology*, **2013**, 25, 703-14	4.9	14
129	Non invasive assessment of lung disease in ataxia telangiectasia by high-field magnetic resonance imaging. *Journal of Clinical Immunology*, **2013**, 33, 1185-91	5.7	18
128	Betamethasone therapy in ataxia telangiectasia: unraveling the rationale of this serendipitous observation on the basis of the pathogenesis. *European Journal of Neurology*, **2013**, 20, 740-7	6	16
127	Does NADPH oxidase deficiency cause artery dilatation in humans?. *Antioxidants and Redox Signaling*, **2013**, 18, 1491-6	8.4	25
126	Reduced atherosclerotic burden in subjects with genetically determined low oxidative stress. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2013**, 33, 406-12	9.4	46
125	Networking Between and GH-R Signaling in the Control of Cell Growth. *Current Signal Transduction Therapy*, **2013**, 8, 67-73	0.8	
124	New strategies for the treatment of lysosomal storage diseases (review). *International Journal of Molecular Medicine*, **2013**, 31, 11-20	4.4	71
123	FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. *Frontiers in Immunology*, **2013**, 4, 187	8.4	51
122	Alterations of the autoimmune regulator transcription factor and failure of central tolerance: APECED as a model. *Expert Review of Clinical Immunology*, **2013**, 9, 43-51	5.1	11
121	Phenotypic characterization and outcome of paediatric patients affected with haemophagocytic syndrome of unknown genetic cause. *British Journal of Haematology*, **2013**, 162, 713-7	4.5	1

120	APECED: A Paradigm of Complex Interactions between Genetic Background and Susceptibility Factors. *Frontiers in Immunology*, **2013**, 4, 331	8.4	25
119	Molecular evidence for a thymus-independent partial T cell development in a FOXN1-/- athymic human fetus. *PLoS ONE*, **2013**, 8, e81786	3.7	5
118	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy from the pediatric perspective. *Journal of Endocrinological Investigation*, **2013**, 36, 903-12	5.2	13
117	Severe combined immunodeficiences: new and old scenarios. *International Reviews of Immunology*, **2012**, 31, 43-65	4.6	37
116	De novo 13q12.3-q14.11 deletion involving BRCA2 gene in a patient with developmental delay, elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an A-T like phenotype. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2571-6	2.5	6
115	Altered regulatory mechanisms governing cell survival in children affected with clustering of autoimmune disorders. *Italian Journal of Pediatrics*, **2012**, 38, 42	3.2	
114	Hyper IgM syndrome presenting as chronic suppurative lung disease. *Italian Journal of Pediatrics*, **2012**, 38, 45	3.2	4
113	Interleukin 12 receptor deficiency in a child with recurrent bronchopneumonia and very high IgE levels. *Italian Journal of Pediatrics*, **2012**, 38, 46	3.2	4
112	A case of galactosemia misdiagnosed as cow milk intolerance. *Italian Journal of Pediatrics*, **2012**, 38, 47	3.2	2
111	Acute adrenal failure as the presenting feature of primary antiphospholipid syndrome in a child. *Italian Journal of Pediatrics*, **2012**, 38, 49	3.2	1
110	Precocious puberty in Turner Syndrome: report of a case and review of the literature. *Italian Journal of Pediatrics*, **2012**, 38, 54	3.2	17
109	Therapeutic options in pediatric non alcoholic fatty liver disease: current status and future directions. *Italian Journal of Pediatrics*, **2012**, 38, 55	3.2	28
108	Bone health in children with long-term idiopathic subclinical hypothyroidism. *Italian Journal of Pediatrics*, **2012**, 38, 56	3.2	18
107	Genetic basis of altered central tolerance and autoimmune diseases: a lesson from AIRE mutations. *International Reviews of Immunology*, **2012**, 31, 344-62	4.6	14
106	Human FOXN1-deficiency is associated with double-negative and FoxP3+ T-cell expansions that are distinctly modulated upon thymic transplantation. *PLoS ONE*, **2012**, 7, e37042	3.7	28
105	Noonan-like syndrome with loose anagen hair associated with growth hormone insensitivity and atypical neurological manifestations. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 856-60	2.5	13
104	Magnetic resonance imaging is an accurate and reliable method to evaluate non-cystic fibrosis paediatric lung disease. *Respirology*, **2012**, 17, 87-91	3.6	51
103	Patient-centred screening for primary immunodeficiency, a multi-stage diagnostic protocol designed for non-immunologists: 2011 update. *Clinical and Experimental Immunology*, **2012**, 167, 108-19	6.2	106

102	The European internet-based patient and research database for primary immunodeficiencies: update 2011. *Clinical and Experimental Immunology*, **2012**, 167, 479-91	6.2	79
101	Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy: insights into genotype-phenotype correlation. *International Journal of Endocrinology*, **2012**, 2012, 353250	2.7	35
100	From murine to human nude/SCID: the thymus, T-cell development and the missing link. *Clinical and Developmental Immunology*, **2012**, 2012, 467101		32
99	Role of the common □chain in cell cycle progression of human malignant cell lines. *International Immunology*, **2012**, 24, 159-67	4.9	8
98	Clinical Heterogeneity in two patients with Noonan-like Syndrome associated with the same SHOC2 mutation. *Italian Journal of Pediatrics*, **2012**, 38, 48	3.2	15
97	Current Role of Leukotriene Receptor Antagonists in Preschool Asthma. *Current Respiratory Medicine Reviews*, **2012**, 8, 391-395	0.3	
96	Molecular background and genotype-phenotype correlation in autoimmune-polyendocrinopathy-candidiasis-ectodermal-distrophy patients from Campania and in their relatives. *Journal of Endocrinological Investigation*, **2012**, 35, 169-73	5.2	18
95	High intrafamilial variability in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy: a case study. *Journal of Endocrinological Investigation*, **2012**, 35, 77-81	5.2	14
94	SCID-like phenotype associated with an inhibitory autoreactive immunoglobulin. *Journal of Investigational Allergology and Clinical Immunology*, **2012**, 22, 67-70	2.3	2
93	Chronic granulomatous disease with gastrointestinal presentation: diagnostic pitfalls and novel ultrastructural findings. *Journal of Investigational Allergology and Clinical Immunology*, **2012**, 22, 527-9	2.3	1
92	Intergenerational anticipation of disease onset in people with multiple autoimmune syndrome. *Diabetes Research and Clinical Practice*, **2011**, 94, e37-9	7.4	4
91	Involvement of nitro-compounds in the mutagenicity of urban Pm2.5 and Pm10 in Turin. *Mutation Research - Genetic Toxicology and Environmental Mutagenesis*, **2011**, 726, 54-9	3	20
90	X-linked lymphoproliferative disease due to SAP/SH2D1A deficiency: a multicenter study on the manifestations, management and outcome of the disease. *Blood*, **2011**, 117, 53-62	2.2	231
89	Immunodeficiency diagnosis: a Mondrian or Pollock scenario?. *Blood*, **2011**, 118, 5714-6	2.2	1
88	Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. *European Journal of Neurology*, **2011**, 18, 564-70	6	55
87	□Chain transducing element: a shared pathway between endocrine and immune system. *Cellular Immunology*, **2011**, 269, 10-5	4.4	4
86	Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. *Clinical Immunology*, **2011**, 139, 6-11	9	43
85	Effectiveness of immunoglobulin replacement therapy on clinical outcome in patients with primary antibody deficiencies: results from a multicenter prospective cohort study. *Journal of Clinical Immunology*, **2011**, 31, 315-22	5.7	203

84	First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases. *Blood*, **2011**, 117, 688-96	2.2	88
83	FOXN1 mutation abrogates prenatal T-cell development in humans. *Journal of Medical Genetics*, **2011**, 48, 413-6	5.8	26
82	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. *Journal of Allergy and Clinical Immunology*, **2010**, 125, 424-432.e8	11.5	200
81	Brain alteration in a Nude/SCID fetus carrying FOXN1 homozygous mutation. *Journal of the Neurological Sciences*, **2010**, 298, 121-3	3.2	11
80	Altered signaling through IL-12 receptor in children with very high serum IgE levels. *Cellular Immunology*, **2010**, 265, 74-9	4.4	1
79	Hereditary deficiency of gp91(phox) is associated with enhanced arterial dilatation: results of a multicenter study. *Circulation*, **2009**, 120, 1616-22	16.7	113
78	The cellular amount of the common gamma-chain influences spontaneous or induced cell proliferation. *Journal of Immunology*, **2009**, 182, 3304-9	5.3	28
77	The quality of life of children and adolescents with X-linked agammaglobulinemia. *Journal of Clinical Immunology*, **2009**, 29, 501-7	5.7	30
76	The European internet-based patient and research database for primary immunodeficiencies: results 2006-2008. *Clinical and Experimental Immunology*, **2009**, 157 Suppl 1, 3-11	6.2	142
75	In ataxia-teleangiectasia betamethasone response is inversely correlated to cerebellar atrophy and directly to antioxidative capacity. *European Journal of Neurology*, **2009**, 16, 755-9	6	38
74	Molecular characterization of a large cohort of patients with Chronic Granulomatous Disease and identification of novel CYBB mutations: an Italian multicenter study. *Molecular Immunology*, **2009**, 46, 1935-41	4.3	32
73	Human ClinicalPhenotype Associated with FOXN1 Mutations. *Advances in Experimental Medicine and Biology*, **2009**, 195-206	3.6	17
72	Assessment of chest high-field magnetic resonance imaging in children and young adults with noncystic fibrosis chronic lung disease: comparison to high-resolution computed tomography and correlation with pulmonary function. *Investigative Radiology*, **2009**, 44, 532-8	10.1	44
71	Human clinical phenotype associated with FOXN1 mutations. *Advances in Experimental Medicine and Biology*, **2009**, 665, 195-206	3.6	6
70	Steroid-induced improvement of neurological signs in ataxia-telangiectasia patients. *European Journal of Neurology*, **2008**, 15, 223-8	6	49
69	FOXN1 homozygous mutation associated with anencephaly and severe neural tube defect in human athymic Nude/SCID fetus. *Clinical Genetics*, **2008**, 73, 380-4	4	50
68	Posterior reversible encephalopathy syndrome in a child during an accelerated phase of a severe APECED phenotype due to an uncommon mutation of AIRE. *Clinical Endocrinology*, **2008**, 69, 511-3	3.4	16
67	Clinical features, long-term follow-up and outcome of a large cohort of patients with Chronic Granulomatous Disease: an Italian multicenter study. *Clinical Immunology*, **2008**, 126, 155-64	9	242

66	A prospective study on children with initial diagnosis of transient hypogammaglobulinemia of infancy: results from the Italian Primary Immunodeficiency Network. *International Journal of Immunopathology and Pharmacology*, **2008**, 21, 343-52	3	53
65	Chemical characteristics and mutagenic activity of PM10 in Torino, a northern Italian city. *Science of the Total Environment*, **2007**, 385, 97-107	10.2	27
64	Long-term follow-up and outcome of a large cohort of patients with common variable immunodeficiency. *Journal of Clinical Immunology*, **2007**, 27, 308-16	5.7	380
63	The mutagenic hazards of environmental PM2.5 in Turin. *Environmental Research*, **2007**, 103, 168-75	7.9	52
62	Airborne particulate matter: Ionic species role in different Italian sites. *Environmental Research*, **2007**, 103, 1-8	7.9	25
61	A91V perforin variation in healthy subjects and FHLH patients. *International Journal of Immunogenetics*, **2006**, 33, 123-5	2.3	26
60	Functional interaction of common gamma-chain and growth hormone receptor signaling apparatus. *Journal of Immunology*, **2006**, 177, 6889-95	5.3	16
59	Cutaneous manifestations as presenting sign of autoimmune lymphoproliferative syndrome in childhood. *Dermatology*, **2005**, 210, 336-40	4.4	9
58	Primary immunodeficiencies: the tip of an iceberg?. *Blood*, **2005**, 105, 2626-2627	2.2	1
57	Nail dystrophy associated with a heterozygous mutation of the nude/SCID human FOXN1 (WHN) gene. *Archives of Dermatology*, **2005**, 141, 647-8		27
56	Allogeneic bone marrow transplantation restores IGF-I production and linear growth in a gamma-SCID patient with abnormal growth hormone receptor signaling. *Bone Marrow Transplantation*, **2004**, 33, 773-5	4.4	6
55	Ancestral founder mutation of the nude (FOXN1) gene in congenital severe combined immunodeficiency associated with alopecia in southern Italy population. *Annals of Human Genetics*, **2004**, 68, 265-8	2.2	66
54	Search for poliovirus long-term excretors among patients affected by agammaglobulinemia. *Clinical Immunology*, **2004**, 111, 98-102	9	23
53	A91V is a polymorphism in the perforin gene not causative of an FHLH phenotype. *Blood*, **2004**, 104, 1909; author reply 1910	2.2	52
52	Atypical features of familial hemophagocytic lymphohistiocytosis. *Blood*, **2004**, 103, 4610-2	2.2	53
51	Atypical X-linked SCID phenotype associated with growth hormone hyporesponsiveness. *Clinical and Experimental Immunology*, **2002**, 129, 502-9	6.2	17
50	A lesson for unraveling complex aspects of novel immunodeficiencies from the human equivalent of the nude/SCID phenotype. *Journal of Hematotherapy and Stem Cell Research*, **2002**, 11, 409-14		7
49	Clinical, immunological, and molecular analysis in a large cohort of patients with X-linked agammaglobulinemia: an Italian multicenter study. *Clinical Immunology*, **2002**, 104, 221-30	9	248

48	The structure of human STAT5A and B genes reveals two regions of nearly identical sequence and an alternative tissue specific STAT5B promoter. *Gene*, **2002**, 285, 311-8	3.8	39
47	Clinical and molecular analysis of patients with defects in heavy chain gene. *Journal of Clinical Investigation*, **2002**, 110, 1029-1035	15.9	51
46	Clinical and molecular analysis of patients with defects in micro heavy chain gene. *Journal of Clinical Investigation*, **2002**, 110, 1029-35	15.9	17
45	Defective surface expression of attractin on T cells in patients with common variable immunodeficiency (CVID). *Clinical and Experimental Immunology*, **2001**, 123, 99-104	6.2	24
44	Defective function of Fas in patients with type 1 diabetes associated with other autoimmune diseases. *Diabetes*, **2001**, 50, 483-8	0.9	41
43	Abnormal GH receptor signaling in children with idiopathic short stature. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 3882-8	5.6	51
42	Human equivalent of the mouse Nude/SCID phenotype: long-term evaluation of immunologic reconstitution after bone marrow transplantation. *Blood*, **2001**, 97, 880-5	2.2	52
41	Clustering of distinct autoimmune diseases associated with functional abnormalities of T cell survival in children. *Clinical and Experimental Immunology*, **2000**, 121, 53-8	6.2	8
40	Simultaneous peripubertal onset of multireactive autoimmune diseases with an unusual long-lasting remission of type 1 diabetes mellitus. *Clinical Endocrinology*, **2000**, 53, 649-53	3.4	4
39	Multisystem involvement in congenital insensitivity to pain with anhidrosis (CIPA), a nerve growth factor receptor(Trk A)-related disorder. *Neuropediatrics*, **2000**, 31, 39-41	1.6	42
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