Claudio Pignata

List of Publications by Year in descending order

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217 papers	7,650 citations	53794 45 h-index	66911 78 g-index
229 all docs	229 docs citations	229 times ranked	8480 citing authors

#	Article	IF	Citations
1	Growth Hormone Receptor (GHR) 6Î© Pseudoexon Activation: A Novel Cause of Severe Growth Hormone Insensitivity. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e401-e416.	3.6	4
2	Vaccinations in Children and Adolescents Treated with Immune-Modifying Biologics: Update and Current Developments. Journal of Allergy and Clinical Immunology: in Practice, 2022, , .	3.8	0
3	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	2.4	8
4	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination. Frontiers in Immunology, 2022, 13, 845496.	4.8	13
5	Progressive Depletion of B and T Lymphocytes in Patients with Ataxia Telangiectasia: Results of the Italian Primary Immunodeficiency Network. Journal of Clinical Immunology, 2022, 42, 783-797.	3.8	5
6	Mechanisms of immune tolerance breakdown in inborn errors of immunity. , 2022, , 73-95.		0
7	The Impact of SARS-CoV-2 Infection in Patients with Inborn Errors of Immunity: the Experience of the Italian Primary Immunodeficiencies Network (IPINet). Journal of Clinical Immunology, 2022, 42, 935-946.	3.8	21
8	Follicular helper T cell signature of replicative exhaustion, apoptosis, and senescence in common variable immunodeficiency. European Journal of Immunology, 2022, 52, 1171-1189.	2.9	9
9	In Ataxia-Telangiectasia, Oral Betamethasone Administration Ameliorates Lymphocytes Functionality through Modulation of the IL-7/IL-7RÎ± Axis Paralleling the Neurological Behavior: A Comparative Report of Two Cases. Immunological Investigations, 2021, 50, 295-303.	2.0	3
10	Complement system network in cell physiology and in human diseases. International Reviews of Immunology, 2021, 40, 159-170.	3.3	10
11	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.	0.8	3
12	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.	3.8	56
13	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.	3.8	4
14	Simple Measurement of IgA Predicts Immunity and Mortality in Ataxia-Telangiectasia. Journal of Clinical Immunology, 2021, 41, 1878-1892.	3.8	9
15	Expanding the Nude SCID/CID Phenotype Associated with FOXN1 Homozygous, Compound Heterozygous, or Heterozygous Mutations. Journal of Clinical Immunology, 2021, 41, 756-768.	3.8	13
16	Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164.	2.1	15
17	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.	0.8	1
18	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.	3.8	11

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19	The Italian Registry for Primary Immunodeficiencies (Italian Primary Immunodeficiency Network;) Tj ETQq1 1 0.784314 rgBT /Overlock 1	3.8	15
20	T-Cell Immunodeficiencies With Congenital Alterations of Thymic Development: Genes Implicated and Differential Immunological and Clinical Features. Frontiers in Immunology, 2020, 11, 1837.	4.8	21
21	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.	2.9	12
22	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.	2.9	59
23	Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. Journal of Clinical Medicine, 2020, 9, 818.	2.4	4
24	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.	3.8	10
25	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.	3.8	38
26	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.	3.8	10
27	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	3
28	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.	4.8	41
29	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.	6.2	52
30	Lentiviral gene therapy corrects platelet phenotype and function in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2019, 144, 825-838.	2.9	50
31	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. Clinical Genetics, 2019, 96, 169-175.	2.0	27
32	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 316.	4.8	42
33	X-linked agammaglobulinemia (XLA): Phenotype, diagnosis, and therapeutic challenges around the world. World Allergy Organization Journal, 2019, 12, 100018.	3.5	83
34	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.	1.5	1
35	Follow-up and outcome of symptomatic partial or absolute IgA deficiency in children. European Journal of Pediatrics, 2019, 178, 51-60.	2.7	22
36	Oral Thrush and Onychomycosis. , 2019, , 371-376.		0

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37	Recurrent Cold Suppurative Granulomatous Lymphadenitis. , 2019, , 347-352.		0
38	Minimum effective betamethasone dosage on the neurological phenotype in patients with ataxiaâ€telangiectasia: a multicenter observerâ€blind study. European Journal of Neurology, 2018, 25, 833-840.	3.3	11
39	Impaired platelet activation in patients with hereditary deficiency of p47^{phox}. British Journal of Haematology, 2018, 180, 454-456.	2.5	5
40	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.	1.2	10
41	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.	2.2	3
42	Neutralizing Anti-Cytokine Autoantibodies Against Interferon-Î± in Immunodysregulation Polyendocrinopathy Enteropathy X-Linked. Frontiers in Immunology, 2018, 9, 544.	4.8	46
43	Biweekly HizentraÂ® in Primary Immunodeficiency: a Multicenter, Observational Cohort Study (IBIS). Journal of Clinical Immunology, 2018, 38, 602-609.	3.8	11
44	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.	2.9	58
45	DiGeorgeâ€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.	1.2	8
46	Recommendations regarding splenectomy in hereditary hemolytic anemias. Haematologica, 2017, 102, 1304-1313.	3.5	138
47	Abnormal cell-clearance and accumulation of autophagic vesicles in lymphocytes from patients affected with Ataxia-Teleangiectasia. Clinical Immunology, 2017, 175, 16-25.	3.2	19
48	FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. Journal of Clinical Immunology, 2017, 37, 751-758.	3.8	36
49	Brain abscesses in children: an Italian multicentre study. Epidemiology and Infection, 2017, 145, 2848-2855.	2.1	21
50	Two Brothers with Atypical UNC13D-Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. Frontiers in Immunology, 2017, 8, 1892.	4.8	8
51	NADPH Oxidase Deficiency: A Multisystem Approach. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-23.	4.0	29
52	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
53	Novel Findings into AIRE Genetics and Functioning: Clinical Implications. Frontiers in Pediatrics, 2016, 4, 86.	1.9	25
54	Unbalanced Immune System: Immunodeficiencies and Autoimmunity. Frontiers in Pediatrics, 2016, 4, 107.	1.9	26

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55	Novel <scp>STAT</scp>1 gainâ€ofâ€function mutation and suppurative infections. Pediatric Allergy and Immunology, 2016, 27, 220-223.	2.6	14
56	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.	2.9	12
57	Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease. Data in Brief, 2016, 7, 311-315.	1.0	10
58	Clinical heterogeneity of dominant chronic mucocutaneous candidiasis disease: presenting as treatment-resistant candidiasis and chronic lung disease. Clinical Immunology, 2016, 164, 1-9.	3.2	27
59	Long-term effects of growth hormone (GH) replacement therapy on hematopoiesis in a large cohort of children with GH deficiency. Endocrine, 2016, 53, 192-198.	2.3	15
60	A Bronchovascular Anomaly in a Patient With 22q11.2 Deletion Syndrome. Journal of Investigational Allergology and Clinical Immunology, 2016, 26, 390-392.	1.3	5
61	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.	1.4	2
62	A mutation in caspase-9 decreases the expression of BAFFR and ICOS in patients with immunodeficiency and lymphoproliferation. Genes and Immunity, 2015, 16, 151-161.	4.1	8
63	Thymic Stromal Alterations and Genetic Disorders of Immune System. Frontiers in Immunology, 2015, 6, 81.	4.8	11
64	Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. International Reviews of Immunology, 2015, 35, 1-14.	3.3	7
65	Severe combined immunodeficiencyâ€"an update. Annals of the New York Academy of Sciences, 2015, 1356, 90-106.	3.8	87
66	Nutritional Status in Agammaglobulinemia: An Italian Multicenter Study. Journal of Clinical Immunology, 2015, 35, 595-597.	3.8	1
67	B cells from nuclear factor κB essential modulator deficient patients fail to differentiate to antibody secreting cells in response to TLR9 ligand. Clinical Immunology, 2015, 161, 131-135.	3.2	5
68	FOXN1 in Organ Development and Human Diseases. International Reviews of Immunology, 2014, 33, 83-93.	3.3	40
69	In This Issue: FOX Genes and the Immune Response. International Reviews of Immunology, 2014, 33, 81-82.	3.3	1
70	Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. Scandinavian Journal of Gastroenterology, 2014, 49, 274-279.	1.5	31
71	Insight intoIKBKG/NEMOLocus: Report of New Mutations and Complex Genomic Rearrangements Leading to Incontinentia Pigmenti Disease. Human Mutation, 2014, 35, 165-177.	2.5	74
72	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.	3.7	31

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73	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.	1.7	12
74	High-content cytometry and transcriptomic biomarker profiling of human B-cell activation. Journal of Allergy and Clinical Immunology, 2014, 133, 172-180.e10.	2.9	15
75	Intergenerational and intrafamilial phenotypic variability in 22q11.2 Deletion syndrome subjects. BMC Medical Genetics, 2014, 15, 1.	2.1	48
76	Clinical Features and Follow-Up in Patients with 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2014, 164, 1475-1480.e2.	1.8	119
77	The PedPAD study: boys predominate in the hypogammaglobulinaemia registry of the ESID online database. Clinical and Experimental Immunology, 2014, 176, 387-393.	2.6	21
78	The R156H variation in IL-12RÎ²1 is not a mutation. Italian Journal of Pediatrics, 2013, 39, 12.	2.6	1
79	Hypertransaminasemia and fatal lung disease: a case report. Italian Journal of Pediatrics, 2013, 39, 9.	2.6	1
80	Non-autoimmune subclinical hypothyroidism due to a mutation in TSH receptor: report on two brothers. Italian Journal of Pediatrics, 2013, 39, 5.	2.6	8
81	Treatment of children with chronic viral hepatitis: what is available and what is in store. World Journal of Pediatrics, 2013, 9, 212-220.	1.8	5
82	Steroid treatment in Ataxia-Telangiectasia induces alterations of functional magnetic resonance imaging during prono-supination task. European Journal of Paediatric Neurology, 2013, 17, 135-140.	1.6	23
83	Human skin-derived keratinocytes and fibroblasts co-cultured on 3D poly Îµ-caprolactone scaffold support in vitro HSC differentiation into T-lineage committed cells. International Immunology, 2013, 25, 703-714.	4.0	15
84	Non Invasive Assessment of Lung Disease in Ataxia Telangiectasia by High-Field Magnetic Resonance Imaging. Journal of Clinical Immunology, 2013, 33, 1185-1191.	3.8	21
85	Betamethasone therapy in <scp>a</scp>taxia <scp>t</scp>elangiectasia: unraveling the rationale of this serendipitous observation on the basis of the pathogenesis. European Journal of Neurology, 2013, 20, 740-747.	3.3	19
86	Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.	5.4	27
87	Reduced Atherosclerotic Burden in Subjects With Genetically Determined Low Oxidative Stress. Arteriosclerosis, Thrombosis, and Vascular Biology, 2013, 33, 406-412.	2.4	52
88	New strategies for the treatment of lysosomal storage diseases (Review). International Journal of Molecular Medicine, 2013, 31, 11-20.	4.0	87
89	FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. Frontiers in Immunology, 2013, 4, 187.	4.8	72
90	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.	3.0	12

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91	Phenotypic characterization and outcome of paediatric patients affected with haemophagocytic syndrome of unknown genetic cause. British Journal of Haematology, 2013, 162, 713-717.	2.5	3
92	APECED: A Paradigm of Complex Interactions between Genetic Background and Susceptibility Factors. Frontiers in Immunology, 2013, 4, 331.	4.8	32
93	Molecular Evidence for a Thymus-Independent Partial T Cell Development in a FOXN1âˆ'/âˆ' Athymic Human Fetus. PLoS ONE, 2013, 8, e81786.	2.5	5
94	Networking Between Î³c and GH-R Signaling in the Control of Cell Growth. Current Signal Transduction Therapy, 2013, 8, 67-73.	0.5	0
95	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy from the pediatric perspective. Journal of Endocrinological Investigation, 2013, 36, 903-12.	3.3	20
96	Anti-infective prophylaxis for primary immunodeficiencies: what is done in Italian Primary Immunodeficiency Network centers (IPINet) and review of the literature. Journal of Biological Regulators and Homeostatic Agents, 2013, 27, 935-46.	0.7	5
97	Networking Between Î³c and GH-R Signaling in the Control of Cell Growth. Current Signal Transduction Therapy, 2013, 8, 67-73.	0.5	0
98	Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy: Insights into Genotype-Phenotype Correlation. International Journal of Endocrinology, 2012, 2012, 1-9.	1.5	42
99	From Murine to Human Nude/SCID: The Thymus, T-Cell Development and the Missing Link. Clinical and Developmental Immunology, 2012, 2012, 1-12.	3.3	39
100	Role of the common Â chain in cell cycle progression of human malignant cell lines. International Immunology, 2012, 24, 159-167.	4.0	8
101	Clinical Heterogeneity in two patients with Noonan-like Syndrome associated with the same SHOC2 mutation. Italian Journal of Pediatrics, 2012, 38, 48.	2.6	21
102	Current Role of Leukotriene Receptor Antagonists in Preschool Asthma. Current Respiratory Medicine Reviews, 2012, 8, 391-395.	0.2	0
103	Severe Combined Immunodeficiences: New and Old Scenarios. International Reviews of Immunology, 2012, 31, 43-65.	3.3	42
104	De novo 13q12.3â€“q14.11 deletion involving BRCA2 gene in a patient with developmental delay, elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an Aâ€T like phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2571-2576.	1.2	6
105	Altered regulatory mechanisms governing cell survival in children affected with clustering of autoimmune disorders. Italian Journal of Pediatrics, 2012, 38, 42.	2.6	0
106	Hyper IgM syndrome presenting as chronic suppurative lung disease. Italian Journal of Pediatrics, 2012, 38, 45.	2.6	5
107	Interleukin 12 receptor deficiency in a child with recurrent bronchopneumonia and very high IgE levels. Italian Journal of Pediatrics, 2012, 38, 46.	2.6	6
108	A case of galactosemia misdiagnosed as cowâ€™s milk intolerance. Italian Journal of Pediatrics, 2012, 38, 47.	2.6	2

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109	Acute adrenal failure as the presenting feature of primary antiphospholipid syndrome in a child. Italian Journal of Pediatrics, 2012, 38, 49.	2.6	3
110	Precocious puberty in Turner Syndrome: report of a case and review of the literature. Italian Journal of Pediatrics, 2012, 38, 54.	2.6	18
111	Therapeutic options in pediatric non alcoholic fatty liver disease: current status and future directions. Italian Journal of Pediatrics, 2012, 38, 55.	2.6	31
112	Bone health in children with longâ€“term idiopathic subclinical hypothyroidism. Italian Journal of Pediatrics, 2012, 38, 56.	2.6	27
113	Genetic Basis of Altered Central Tolerance and Autoimmune Diseases: A Lesson from AIRE Mutations. International Reviews of Immunology, 2012, 31, 344-362.	3.3	18
114	Human FOXN1-Deficiency Is Associated with Î±Î² Double-Negative and FoxP3+ T-Cell Expansions That Are Distinctly Modulated upon Thymic Transplantation. PLoS ONE, 2012, 7, e37042.	2.5	35
115	Noonanâ€like syndrome with loose anagen hair associated with growth hormone insensitivity and atypical neurological manifestations. American Journal of Medical Genetics, Part A, 2012, 158A, 856-860.	1.2	16
116	Magnetic resonance imaging is an accurate and reliable method to evaluate nonâ€cystic fibrosis paediatric lung disease. Respirology, 2012, 17, 87-91.	2.3	58
117	The European internet-based patient and research database for primary immunodeficiencies: update 2011. Clinical and Experimental Immunology, 2012, 167, 479-491.	2.6	91
118	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.	3.3	21
119	High intrafamilial variability in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy: a case study. Journal of Endocrinological Investigation, 2012, 35, 77-81.	3.3	16
120	SCID-like phenotype associated with an inhibitory autoreactive immunoglobulin. Journal of Investigational Allergology and Clinical Immunology, 2012, 22, 67-70.	1.3	2
121	Chronic granulomatous disease with gastrointestinal presentation: diagnostic pitfalls and novel ultrastructural findings. Journal of Investigational Allergology and Clinical Immunology, 2012, 22, 527-9.	1.3	1
122	First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases. Blood, 2011, 117, 688-696.	1.4	109
123	Intergenerational anticipation of disease onset in people with multiple autoimmune syndrome. Diabetes Research and Clinical Practice, 2011, 94, e37-e39.	2.8	4
124	Involvement of nitro-compounds in the mutagenicity of urban Pm2.5 and Pm10 in Turin. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2011, 726, 54-59.	1.7	22
125	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.	1.4	268
126	Immunodeficiency diagnosis: a Mondrian or Pollock scenario?. Blood, 2011, 118, 5714-5716.	1.4	1

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127	Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. European Journal of Neurology, 2011, 18, 564-570.	3.3	62
128	Î³ Chain transducing element: A shared pathway between endocrine and immune system. Cellular Immunology, 2011, 269, 10-15.	3.0	4
129	Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. Clinical Immunology, 2011, 139, 6-11.	3.2	49
130	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, 315-322.	3.8	252
131	FOXN1 mutation abrogates prenatal T-cell development in humans. Journal of Medical Genetics, 2011, 48, 413-416.	3.2	31
132	Patient-centred screening for primary immunodeficiency, a multi-stage diagnostic protocol designed for non-immunologists: 2011 update. Clinical and Experimental Immunology, 2011, 167, 108-119.	2.6	143
133	Altered signaling through IL-12 receptor in children with very high serum IgE levels. Cellular Immunology, 2010, 265, 74-79.	3.0	1
134	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 424-432.e8.	2.9	247
135	Brain alteration in a Nude/SCID fetus carrying FOXN1 homozygous mutation. Journal of the Neurological Sciences, 2010, 298, 121-123.	0.6	12
136	Hereditary Deficiency of gp91 ^{phox} Is Associated With Enhanced Arterial Dilatation. Circulation, 2009, 120, 1616-1622.	1.6	123
137	The Cellular Amount of the Common Î³-Chain Influences Spontaneous or Induced Cell Proliferation. Journal of Immunology, 2009, 182, 3304-3309.	0.8	29
138	The Quality of Life of Children and Adolescents with X-Linked Agammaglobulinemia. Journal of Clinical Immunology, 2009, 29, 501-507.	3.8	34
139	The European internet-based patient and research database for primary immunodeficiencies: results 2006â€“2008. Clinical and Experimental Immunology, 2009, 157, 3-11.	2.6	203
140	In ataxiaâ€teleangiectasia betamethasone response is inversely correlated to cerebellar atrophy and directly to antioxidative capacity. European Journal of Neurology, 2009, 16, 755-759.	3.3	42
141	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, 1935-1941.	2.2	36
142	Human ClinicalPhenotype Associated with FOXN1 Mutations. Advances in Experimental Medicine and Biology, 2009, , 195-206.	1.6	21
143	Assessment of Chest High-Field Magnetic Resonance Imaging in Children and Young Adults With Noncystic Fibrosis Chronic Lung Disease. Investigative Radiology, 2009, 44, 532-538.	6.2	52
144	Human clinical phenotype associated with FOXN1 mutations. Advances in Experimental Medicine and Biology, 2009, 665, 195-206.	1.6	6

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145	Steroid-induced improvement of neurological signs in ataxia-telangiectasia patients. European Journal of Neurology, 2008, 15, 223-228.	3.3	55
146	FOXN1 homozygous mutation associated with anencephaly and severe neural tube defect in human athymic Nude/SCID fetus. Clinical Genetics, 2008, 73, 380-384.	2.0	55
147	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, 511-513.	2.4	18
148	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, 155-164.	3.2	293
149	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, 343-352.	2.1	61
150	Shared Signaling Pathways Between Endocrine and Immune System Receptors: The Model of Gamma Chain. Current Signal Transduction Therapy, 2008, 3, 206-214.	0.5	0
151	The mutagenic hazards of environmental PM2.5 in Turin. Environmental Research, 2007, 103, 168-175.	7.5	57
152	Airborne particulate matter: Ionic species role in different Italian sites. Environmental Research, 2007, 103, 1-8.	7.5	28
153	Chemical characteristics and mutagenic activity of PM10 in Torino, a Northern Italian City. Science of the Total Environment, 2007, 385, 97-107.	8.0	33
154	Long-Term Follow-Up and Outcome of a Large Cohort of Patients with Common Variable Immunodeficiency. Journal of Clinical Immunology, 2007, 27, 308-316.	3.8	465
155	A91V perforin variation in healthy subjects and FHLH patients. International Journal of Immunogenetics, 2006, 33, 123-125.	1.8	27
156	Functional Interaction of Common Î³-Chain and Growth Hormone Receptor Signaling Apparatus. Journal of Immunology, 2006, 177, 6889-6895.	0.8	17
157	Primary immunodeficiencies: the tip of an iceberg?. Blood, 2005, 105, 2626-2627.	1.4	1
158	Nail Dystrophy Associated With a Heterozygous Mutation of the Nude/SCID Human FOXN1 (WHN) Gene. Archives of Dermatology, 2005, 141, 647-8.	1.4	28
159	Cutaneous Manifestations as Presenting Sign of Autoimmune Lymphoproliferative Syndrome in Childhood. Dermatology, 2005, 210, 336-340.	2.1	14
160	Allogeneic bone marrow transplantation restores IGF-I production and linear growth in a Î³-SCID patient with abnormal growth hormone receptor signaling. Bone Marrow Transplantation, 2004, 33, 773-775.	2.4	7
161	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, 265-268.	0.8	75
162	Search for poliovirus long-term excretors among patients affected by agammaglobulinemia. Clinical Immunology, 2004, 111, 98-102.	3.2	24

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163	A91V is a polymorphism in the perforin gene not causative of an FHLH phenotype. Blood, 2004, 104, 1909-1910.	1.4	53
164	Atypical features of familial hemophagocytic lymphohistiocytosis. Blood, 2004, 103, 4610-4612.	1.4	55
165	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, 409-414.	1.8	8
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