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	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
2	Clinical, Immunological, and Molecular Analysis in a Large Cohort of Patients with X-Linked Agammaglobulinemia: An Italian Multicenter Study. Clinical Immunology, 2002, 104, 221-230.	3.2	299
3	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
4	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
5	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
6	Exposing the human nude phenotype. Nature, 1999, 398, 473-474.	27.8	247
7	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 424-432.e8.	2.9	247
8	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
9	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
10	Recommendations regarding splenectomy in hereditary hemolytic anemias. Haematologica, 2017, 102, 1304-1313.	3.5	138
11	Hereditary Deficiency of gp91 ^{phox} Is Associated With Enhanced Arterial Dilatation. Circulation, 2009, 120, 1616-1622.	1.6	123
12	Congenital alopecia and nail dystrophy associated with severe functional T-cell immunodeficiency in two sibs. American Journal of Medical Genetics Part A, 1996, 65, 167-170.	2.4	121
13	Clinical Features and Follow-Up in Patients with 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2014, 164, 1475-1480.e2.	1.8	119
14	First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases. Blood, 2011, 117, 688-696.	1.4	109
15	The European internet-based patient and research database for primary immunodeficiencies: update 2011. Clinical and Experimental Immunology, 2012, 167, 479-491.	2.6	91
16	New strategies for the treatment of lysosomal storage diseases (Review). International Journal of Molecular Medicine, 2013, 31, 11-20.	4.0	87
17	Severe combined immunodeficiencyâ€”an update. Annals of the New York Academy of Sciences, 2015, 1356, 90-106.	3.8	87
18	Jejunal bacterial overgrowth and intestinal permeability in children with immunodeficiency syndromes.. Gut, 1990, 31, 879-882.	12.1	85

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19	X-linked agammaglobulinemia (XLA): Phenotype, diagnosis, and therapeutic challenges around the world. World Allergy Organization Journal, 2019, 12, 100018.	3.5	83
20	DiGeorge anomaly associated with 10p deletion. American Journal of Medical Genetics Part A, 1991, 39, 215-216.	2.4	80
21	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
22	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
23	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
24	FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. Frontiers in Immunology, 2013, 4, 187.	4.8	72
25	Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. European Journal of Neurology, 2011, 18, 564-570.	3.3	62
26	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
27	Human equivalent of the mouse Nude/SCID phenotype: long-term evaluation of immunologic reconstitution after bone marrow transplantation. Blood, 2001, 97, 880-885.	1.4	59
28	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
29	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
30	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
31	The mutagenic hazards of environmental PM2.5 in Turin. Environmental Research, 2007, 103, 168-175.	7.5	57
32	Clinical and molecular analysis of patients with defects in Î¼ heavy chain gene. Journal of Clinical Investigation, 2002, 110, 1029-1035.	8.2	57
33	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
34	Rapid disease progression in HIV-1 perinatally infected children born to mothers receiving zidovudine monotherapy during pregnancy. Aids, 1999, 13, 927-933.	2.2	55
35	Abnormal GH Receptor Signaling in Children with Idiopathic Short Stature. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3882-3888.	3.6	55
36	Atypical features of familial hemophagocytic lymphohistiocytosis. Blood, 2004, 103, 4610-4612.	1.4	55

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37	Steroid-induced improvement of neurological signs in ataxia-telangiectasia patients. European Journal of Neurology, 2008, 15, 223-228.	3.3	55
38	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
39	Severe, Protracted Intestinal Cryptosporidiosis Associated with Interferon Â Deficiency: Pediatric Case Report. Clinical Infectious Diseases, 1996, 22, 848-850.	5.8	54
40	A91V is a polymorphism in the perforin gene not causative of an FHLH phenotype. Blood, 2004, 104, 1909-1910.	1.4	53
41	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
42	Reduced Atherosclerotic Burden in Subjects With Genetically Determined Low Oxidative Stress. Arteriosclerosis, Thrombosis, and Vascular Biology, 2013, 33, 406-412.	2.4	52
43	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
44	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
45	Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. Clinical Immunology, 2011, 139, 6-11.	3.2	49
46	Intergenerational and intrafamilial phenotypic variability in 22q11.2 Deletion syndrome subjects. BMC Medical Genetics, 2014, 15, 1.	2.1	48
47	Neutralizing Anti-Cytokine Autoantibodies Against Interferon-Î± in Immunodysregulation Polyendocrinopathy Enteropathy X-Linked. Frontiers in Immunology, 2018, 9, 544.	4.8	46
48	Updating about Reductions of Air and Blood Lead Concentrations in Turin, Italy, Following Reductions in the Lead Content of Gasoline. Environmental Research, 1995, 70, 30-34.	7.5	45
49	Multisystem Involvement in Congenital Insensitivity to Pain with Anhidrosis (CIPA), a Nerve Growth Factor Receptor(Trk A)-Related Disorder. Neuropediatrics, 2000, 31, 39-41.	0.6	45
50	Defective Function of Fas in Patients With Type 1 Diabetes Associated With Other Autoimmune Diseases. Diabetes, 2001, 50, 483-488.	0.6	45
51	Fc gamma RIIIA-mediated signaling involves src-family lck in human natural killer cells. Journal of Immunology, 1993, 151, 6794-800.	0.8	45
52	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, 311-318.	2.2	44
53	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
54	Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy: Insights into Genotype-Phenotype Correlation. International Journal of Endocrinology, 2012, 2012, 1-9.	1.5	42

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55	Severe Combined Immunodeficiences: New and Old Scenarios. International Reviews of Immunology, 2012, 31, 43-65.	3.3	42
56	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 316.	4.8	42
57	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
58	FOXN1 in Organ Development and Human Diseases. International Reviews of Immunology, 2014, 33, 83-93.	3.3	40
59	Occupancy of dipeptidyl peptidase IV activates an associated tyrosine kinase and triggers an apoptotic signal in human hepatocarcinoma cells. Hepatology, 1998, 27, 934-942.	7.3	39
60	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
61	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
62	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
63	FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. Journal of Clinical Immunology, 2017, 37, 751-758.	3.8	36
64	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
65	The Quality of Life of Children and Adolescents with X-Linked Agammaglobulinemia. Journal of Clinical Immunology, 2009, 29, 501-507.	3.8	34
66	Phosphorylation of src Family lck Tyrosine Kinase Following Interleukin-12 Activation of Human Natural Killer Cells. Cellular Immunology, 1995, 165, 211-216.	3.0	33
67	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
68	APECED: A Paradigm of Complex Interactions between Genetic Background and Susceptibility Factors. Frontiers in Immunology, 2013, 4, 331.	4.8	32
69	FOXN1 mutation abrogates prenatal T-cell development in humans. Journal of Medical Genetics, 2011, 48, 413-416.	3.2	31
70	Therapeutic options in pediatric non alcoholic fatty liver disease: current status and future directions. Italian Journal of Pediatrics, 2012, 38, 55.	2.6	31
71	Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. Scandinavian Journal of Gastroenterology, 2014, 49, 274-279.	1.5	31
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	The Cellular Amount of the Common Î³-Chain Influences Spontaneous or Induced Cell Proliferation. Journal of Immunology, 2009, 182, 3304-3309.	0.8	29
74	NADPH Oxidase Deficiency: A Multisystem Approach. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-23.	4.0	29
75	Apoptosis as a Mechanism of Peripheral Blood Mononuclear Cell Death after Measles and Varicella-Zoster Virus Infections in Children. Pediatric Research, 1998, 43, 77-83.	2.3	29
76	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
77	Airborne particulate matter: Ionic species role in different Italian sites. Environmental Research, 2007, 103, 1-8.	7.5	28
78	A91V perforin variation in healthy subjects and FHLH patients. International Journal of Immunogenetics, 2006, 33, 123-125.	1.8	27
79	Bone health in children with longâ€“term idiopathic subclinical hypothyroidism. Italian Journal of Pediatrics, 2012, 38, 56.	2.6	27
80	Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.	5.4	27
81	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
82	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. Clinical Genetics, 2019, 96, 169-175.	2.0	27
83	Defective surface expression of attractin on T cells in patients with common variable immunodeficiency (CVID). Clinical and Experimental Immunology, 2001, 123, 99-104.	2.6	26
84	Unbalanced Immune System: Immunodeficiencies and Autoimmunity. Frontiers in Pediatrics, 2016, 4, 107.	1.9	26
85	Novel Findings into AIRE Genetics and Functioning: Clinical Implications. Frontiers in Pediatrics, 2016, 4, 86.	1.9	25
86	Abnormal GH Receptor Signaling in Children with Idiopathic Short Stature. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3882-3888.	3.6	25
87	Search for poliovirus long-term excretors among patients affected by agammaglobulinemia. Clinical Immunology, 2004, 111, 98-102.	3.2	24
88	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
89	Clinical and molecular analysis of patients with defects in Î¼ heavy chain gene. Journal of Clinical Investigation, 2002, 110, 1029-1035.	8.2	23
90	DNA TYPING OF DQ AND DR ALLELES IN IgA-DEFICIENT SUBJECTS. International Journal of Immunogenetics, 1995, 22, 403-411.	1.2	22

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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, 54-59.	1.7	22
92	Follow-up and outcome of symptomatic partial or absolute IgA deficiency in children. European Journal of Pediatrics, 2019, 178, 51-60.	2.7	22
93	Human ClinicalPhenotype Associated with FOXN1 Mutations. Advances in Experimental Medicine and Biology, 2009, , 195-206.	1.6	21
94	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
95	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
96	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
97	Brain abscesses in children: an Italian multicentre study. Epidemiology and Infection, 2017, 145, 2848-2855.	2.1	21
98	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
99	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
100	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
101	T Cell Activation Deficiency Associated with an Aberrant Pattern of Protein Tyrosine Phosphorylation after CD3 Perturbation in Down's Syndrome. Pediatric Research, 1998, 44, 252-258.	2.3	20
102	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy from the pediatric perspective. Journal of Endocrinological Investigation, 2013, 36, 903-12.	3.3	20
103	Italian guidelines for antiretroviral therapy in children with human immunodeficiency virus-type 1 infection. Acta Paediatrica, International Journal of Paediatrics, 1999, 88, 228-232.	1.5	19
104	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
105	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
106	Increased CD5+CD19+ B lymphocytes at the onset of type 1 diabetes in children. Acta Diabetologica, 1997, 34, 271-274.	2.5	18
107	Atypical X-linked SCID phenotype associated with growth hormone hyporesponsiveness. Clinical and Experimental Immunology, 2002, 129, 502-509.	2.6	18
108	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

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109	Precocious puberty in Turner Syndrome: report of a case and review of the literature. Italian Journal of Pediatrics, 2012, 38, 54.	2.6	18
110	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
111	Impaired suppressor activity in children affected by coeliac disease.. Gut, 1985, 26, 285-290.	12.1	17
112	Functional Interaction of Common Î³-Chain and Growth Hormone Receptor Signaling Apparatus. Journal of Immunology, 2006, 177, 6889-6895.	0.8	17
113	Defective activation of mitogen-activated protein kinase after allogeneic bone marrow transplantation. Blood, 1996, 88, 2334-2341.	1.4	16
114	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
115	High intrafamilial variability in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy: a case study. Journal of Endocrinological Investigation, 2012, 35, 77-81.	3.3	16
116	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
117	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
118	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
119	The Italian Registry for Primary Immunodeficiencies (Italian Primary Immunodeficiency Network;) Tj ETQq1 1 0.784314 rgBT /Overlock	3.8	15
120	Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164.	2.1	15
121	Cutaneous Manifestations as Presenting Sign of Autoimmune Lymphoproliferative Syndrome in Childhood. Dermatology, 2005, 210, 336-340.	2.1	14
122	Novel <scp>STAT</scp>1 gainâ€ofâ€function mutation and suppurative infections. Pediatric Allergy and Immunology, 2016, 27, 220-223.	2.6	14
123	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
124	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination. Frontiers in Immunology, 2022, 13, 845496.	4.8	13
125	Progressive Deficiencies in Blood T Cells Associated with a 10p12-13 Interstitial Deletion. Clinical Immunology and Immunopathology, 1996, 80, 9-15.	2.0	12
126	Brain alteration in a Nude/SCID fetus carrying FOXN1 homozygous mutation. Journal of the Neurological Sciences, 2010, 298, 121-123.	0.6	12

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127	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
128	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
129	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
130	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
131	Thymic Stromal Alterations and Genetic Disorders of Immune System. Frontiers in Immunology, 2015, 6, 81.	4.8	11
132	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
133	Biweekly HizentraÂ® in Primary Immunodeficiency: a Multicenter, Observational Cohort Study (IBIS). Journal of Clinical Immunology, 2018, 38, 602-609.	3.8	11
134	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
135	Chronic Unexplained Liver Disease in Children With Primary Immunodeficiency Syndromes. Journal of Clinical Gastroenterology, 1998, 26, 187-192.	2.2	11
136	Prolonged Q-T Interval Syndrome Presenting as Idiopathic Epilepsy. Neuropediatrics, 1983, 14, 235-236.	0.6	10
137	Clustering of distinct autoimmune diseases associated with functional abnormalities of T cell survival in children. Clinical and Experimental Immunology, 2000, 121, 53-58.	2.6	10
138	Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease. Data in Brief, 2016, 7, 311-315.	1.0	10
139	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
140	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
141	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
142	Complement system network in cell physiology and in human diseases. International Reviews of Immunology, 2021, 40, 159-170.	3.3	10
143	Simple Measurement of IgA Predicts Immunity and Mortality in Ataxia-Telangiectasia. Journal of Clinical Immunology, 2021, 41, 1878-1892.	3.8	9
144	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

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145	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
146	Role of the common Â chain in cell cycle progression of human malignant cell lines. International Immunology, 2012, 24, 159-167.	4.0	8
147	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
148	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
149	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
150	Two Brothers with Atypical UNC13D-Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. Frontiers in Immunology, 2017, 8, 1892.	4.8	8
151	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	2.4	8
152	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
153	Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. International Reviews of Immunology, 2015, 35, 1-14.	3.3	7
154	Immunoregulatory T Subsets in Chronic Active Viral Hepatitis. Journal of Pediatric Gastroenterology and Nutrition, 1983, 2, 229-233.	1.8	6
155	Pertussis immunization in HIV-1-infected infants: a model to assess the effects of repeated T cell-dependent antigen administrations on HIV-1 progression.. Vaccine, 2000, 18, 1203-1209.	3.8	6
156	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
157	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
158	Clinical and molecular analysis of patients with defects in Î¼ heavy chain gene. Journal of Clinical Investigation, 2002, 110, 1029-1035.	8.2	6
159	Human clinical phenotype associated with FOXN1 mutations. Advances in Experimental Medicine and Biology, 2009, 665, 195-206.	1.6	6
160	Simultaneous peripubertal onset of multireactive autoimmune diseases with an unusual long-lasting remission of type 1 diabetes mellitus. Clinical Endocrinology, 2000, 53, 649-653.	2.4	5
161	Hyper IgM syndrome presenting as chronic suppurative lung disease. Italian Journal of Pediatrics, 2012, 38, 45.	2.6	5
162	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

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163	Molecular Evidence for a Thymus-Independent Partial T Cell Development in a FOXN1â^'/â^' Athymic Human Fetus. PLoS ONE, 2013, 8, e81786.	2.5	5
164	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, 131-135.	3.2	5
165	Impaired platelet activation in patients with hereditary deficiency of p47^{phox}. British Journal of Haematology, 2018, 180, 454-456.	2.5	5
166	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
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