# Claudio Pignata

## List of Publications by Citations



| | | | |
|---|---|---|---|
| 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 | 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 |
| 208 | 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 |
| 207 | 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 |
| 206 | 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 |
| 205 | Exposing the human nude phenotype. *Nature*, **1999**, 398, 473-4 | 50.4 | 218 |
| 204 | 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 |
| 203 | Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. *Journal of Allergy and Clinical Immunology*, **2010**, 125, 424-432.e8 | 11.5 | 200 |
| 202 | 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 |
| 201 | Hereditary deficiency of gp91(phox) is associated with enhanced arterial dilatation: results of a multicenter study. *Circulation*, **2009**, 120, 1616-22 | 16.7 | 113 |
| 200 | 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 |
| 199 | Congenital Alopecia and nail dystrophy associated with severe functional T-cell immunodeficiency in two sibs. *American Journal of Medical Genetics Part A*, **1996**, 65, 167-70 | | 101 |
| 198 | First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases. *Blood*, **2011**, 117, 688-96 | 2.2 | 88 |
| 197 | Clinical features and follow-up in patients with 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2014**, 164, 1475-80.e2 | 3.6 | 83 |
| 196 | The European internet-based patient and research database for primary immunodeficiencies: update 2011. *Clinical and Experimental Immunology*, **2012**, 167, 479-91 | 6.2 | 79 |
| 195 | Recommendations regarding splenectomy in hereditary hemolytic anemias. *Haematologica*, **2017**, 102, 1304-1313 | 6.6 | 77 |
| 194 | DiGeorge anomaly associated with 10p deletion. *American Journal of Medical Genetics Part A*, **1991**, 39, 215-6 | | 75 |
| 193 | Jejunal bacterial overgrowth and intestinal permeability in children with immunodeficiency syndromes. *Gut*, **1990**, 31, 879-82 | 19.2 | 73 |

| | | | |
|---|---|---|---|
| 192 | New strategies for the treatment of lysosomal storage diseases (review). *International Journal of Molecular Medicine*, **2013**, 31, 11-20 | 4.4 | 71 |
| 191 | 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 |
| 190 | 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 |
| 189 | Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. *European Journal of Neurology*, **2011**, 18, 564-70 | 6 | 55 |
| 188 | Severe combined immunodeficiency--an update. *Annals of the New York Academy of Sciences*, **2015**, 1356, 90-106 | 6.5 | 54 |
| 187 | X-linked agammaglobulinemia (XLA):Phenotype, diagnosis, and therapeutic challenges around the world. *World Allergy Organization Journal*, **2019**, 12, 100018 | 5.2 | 53 |
| 186 | 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 |
| 185 | Atypical features of familial hemophagocytic lymphohistiocytosis. *Blood*, **2004**, 103, 4610-2 | 2.2 | 53 |
| 184 | The mutagenic hazards of environmental PM2.5 in Turin. *Environmental Research*, **2007**, 103, 168-75 | 7.9 | 52 |
| 183 | A91V is a polymorphism in the perforin gene not causative of an FHLH phenotype. *Blood*, **2004**, 104, 1909; author reply 1910 | 2.2 | 52 |
| 182 | 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 |
| 181 | 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 |
| 180 | FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. *Frontiers in Immunology*, **2013**, 4, 187 | 8.4 | 51 |
| 179 | Abnormal GH receptor signaling in children with idiopathic short stature. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 3882-8 | 5.6 | 51 |
| 178 | Clinical and molecular analysis of patients with defects in heavy chain gene. *Journal of Clinical Investigation*, **2002**, 110, 1029-1035 | 15.9 | 51 |
| 177 | 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 |
| 176 | Steroid-induced improvement of neurological signs in ataxia-telangiectasia patients. *European Journal of Neurology*, **2008**, 15, 223-8 | 6 | 49 |
| 175 | Reduced atherosclerotic burden in subjects with genetically determined low oxidative stress. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2013**, 33, 406-12 | 9.4 | 46 |

| | | | |
|---|---|---|---|
| 174 | Severe, protracted intestinal cryptosporidiosis associated with interferon gamma deficiency: pediatric case report. *Clinical Infectious Diseases*, **1996**, 22, 848-50 | 11.6 | 46 |
| 173 | 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 |
| 172 | Rapid disease progression in HIV-1 perinatally infected children born to mothers receiving zidovudine monotherapy during pregnancy. *Aids*, **1999**, 13, 927-933 | 3.5 | 44 |
| 171 | Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. *Frontiers in Immunology*, **2016**, 7, 466 | 8.4 | 44 |
| 170 | Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. *Clinical Immunology*, **2011**, 139, 6-11 | 9 | 43 |
| 169 | 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 |
| 168 | Fc gamma RIIIA-mediated signaling involves src-family lck in human natural killer cells. *Journal of Immunology*, **1993**, 151, 6794-800 | 5.3 | 42 |
| 167 | Defective function of Fas in patients with type 1 diabetes associated with other autoimmune diseases. *Diabetes*, **2001**, 50, 483-8 | 0.9 | 41 |
| 166 | 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 |
| 165 | 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 |
| 164 | Updating about reductions of air and blood lead concentrations in Turin, Italy, following reductions in the lead content of gasoline. *Environmental Research*, **1995**, 70, 30-4 | 7.9 | 38 |
| 163 | Severe combined immunodeficiences: new and old scenarios. *International Reviews of Immunology*, **2012**, 31, 43-65 | 4.6 | 37 |
| 162 | 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 |
| 161 | FOXN1 in organ development and human diseases. *International Reviews of Immunology*, **2014**, 33, 83-93 | 4.6 | 35 |
| 160 | Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy: insights into genotype-phenotype correlation. *International Journal of Endocrinology*, **2012**, 2012, 353250 | 2.7 | 35 |
| 159 | Occupancy of dipeptidyl peptidase IV activates an associated tyrosine kinase and triggers an apoptotic signal in human hepatocarcinoma cells. *Hepatology*, **1998**, 27, 934-42 | 11.2 | 35 |
| 158 | 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 |
| 157 | From murine to human nude/SCID: the thymus, T-cell development and the missing link. *Clinical and Developmental Immunology*, **2012**, 2012, 467101 | | 32 |

| | | | |
|---|---|---|---|
| 156 | 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 |
| 155 | The quality of life of children and adolescents with X-linked agammaglobulinemia. *Journal of Clinical Immunology*, **2009**, 29, 501-7 | 5.7 | 30 |
| 154 | Phosphorylation of src family lck tyrosine kinase following interleukin-12 activation of human natural killer cells. *Cellular Immunology*, **1995**, 165, 211-6 | 4.4 | 30 |
| 153 | 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 |
| 152 | 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 |
| 151 | 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 |
| 150 | Intergenerational and intrafamilial phenotypic variability in 22q11.2 deletion syndrome subjects. *BMC Medical Genetics*, **2014**, 15, 1 | 2.1 | 28 |
| 149 | Therapeutic options in pediatric non alcoholic fatty liver disease: current status and future directions. *Italian Journal of Pediatrics*, **2012**, 38, 55 | 3.2 | 28 |
| 148 | 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 |
| 147 | The cellular amount of the common gamma-chain influences spontaneous or induced cell proliferation. *Journal of Immunology*, **2009**, 182, 3304-9 | 5.3 | 28 |
| 146 | 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 |
| 145 | Nail dystrophy associated with a heterozygous mutation of the nude/SCID human FOXN1 (WHN) gene. *Archives of Dermatology*, **2005**, 141, 647-8 | | 27 |
| 144 | FOXN1 mutation abrogates prenatal T-cell development in humans. *Journal of Medical Genetics*, **2011**, 48, 413-6 | 5.8 | 26 |
| 143 | A91V perforin variation in healthy subjects and FHLH patients. *International Journal of Immunogenetics*, **2006**, 33, 123-5 | 2.3 | 26 |
| 142 | Does NADPH oxidase deficiency cause artery dilatation in humans?. *Antioxidants and Redox Signaling*, **2013**, 18, 1491-6 | 8.4 | 25 |
| 141 | Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. *Scandinavian Journal of Gastroenterology*, **2014**, 49, 274-9 | 2.4 | 25 |
| 140 | APECED: A Paradigm of Complex Interactions between Genetic Background and Susceptibility Factors. *Frontiers in Immunology*, **2013**, 4, 331 | 8.4 | 25 |
| 139 | Airborne particulate matter: Ionic species role in different Italian sites. *Environmental Research*, **2007**, 103, 1-8 | 7.9 | 25 |

| | | | |
|---|---|---|---|
| 138 | 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 |
| 137 | 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 |
| 136 | Search for poliovirus long-term excretors among patients affected by agammaglobulinemia. *Clinical Immunology*, **2004**, 111, 98-102 | 9 | 23 |
| 135 | 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 | 3.2 | 23 |
| 134 | Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. *Frontiers in Immunology*, **2019**, 10, 316 | 8.4 | 22 |
| 133 | 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 |
| 132 | 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 |
| 131 | NADPH Oxidase Deficiency: A Multisystem Approach. *Oxidative Medicine and Cellular Longevity*, **2017**, 2017, 4590127 | 6.7 | 21 |
| 130 | Novel Findings into AIRE Genetics and Functioning: Clinical Implications. *Frontiers in Pediatrics*, **2016** , 4, 86 | 3.4 | 21 |
| 129 | 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 |
| 128 | DNA typing of DQ and DR alleles in IgA-deficient subjects. *International Journal of Immunogenetics*, **1995**, 22, 403-11 | | 20 |
| 127 | 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 |
| 126 | Neutralizing Anti-Cytokine Autoantibodies Against Interferon-□n Immunodysregulation Polyendocrinopathy Enteropathy X-Linked. *Frontiers in Immunology*, **2018**, 9, 544 | 8.4 | 19 |
| 125 | Italian guidelines for antiretroviral therapy in children with human immunodeficiency virus-type 1 infection. *Acta Paediatrica, International Journal of Paediatrics*, **1999**, 88, 228-232 | 3.1 | 19 |
| 124 | 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 |
| 123 | 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 |
| 122 | Bone health in children with long-term idiopathic subclinical hypothyroidism. *Italian Journal of Pediatrics*, **2012**, 38, 56 | 3.2 | 18 |
| 121 | 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 |

| | | | |
|---|---|---|---|
| 120 | FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. *Journal of Clinical Immunology*, **2017**, 37, 751-758 | 5.7 | 17 |
| 119 | Precocious puberty in Turner Syndrome: report of a case and review of the literature. *Italian Journal of Pediatrics*, **2012**, 38, 54 | 3.2 | 17 |
| 118 | Human ClinicalPhenotype Associated with FOXN1 Mutations. *Advances in Experimental Medicine and Biology*, **2009**, 195-206 | 3.6 | 17 |
| 117 | Increased CD5+CD19+ B lymphocytes at the onset of type 1 diabetes in children. *Acta Diabetologica* , **1997**, 34, 271-4 | 3.9 | 17 |
| 116 | Atypical X-linked SCID phenotype associated with growth hormone hyporesponsiveness. *Clinical and Experimental Immunology*, **2002**, 129, 502-9 | 6.2 | 17 |
| 115 | Clinical and molecular analysis of patients with defects in micro heavy chain gene. *Journal of Clinical Investigation*, **2002**, 110, 1029-35 | 15.9 | 17 |
| 114 | T cell activation deficiency associated with an aberrant pattern of protein tyrosine phosphorylation after CD3 perturbation in Down syndrome. *Pediatric Research*, **1998**, 44, 252-8 | 3.2 | 17 |
| 113 | Abnormal cell-clearance and accumulation of autophagic vesicles in lymphocytes from patients affected with Ataxia-Teleangiectasia. *Clinical Immunology*, **2017**, 175, 16-25 | 9 | 16 |
| 112 | Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. *Clinical Genetics*, **2019**, 96, 169-175 | 4 | 16 |
| 111 | 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 |
| 110 | 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 |
| 109 | Functional interaction of common gamma-chain and growth hormone receptor signaling apparatus. *Journal of Immunology*, **2006**, 177, 6889-95 | 5.3 | 16 |
| 108 | Impaired suppressor activity in children affected by coeliac disease. *Gut*, **1985**, 26, 285-90 | 19.2 | 16 |
| 107 | Unbalanced Immune System: Immunodeficiencies and Autoimmunity. *Frontiers in Pediatrics*, **2016**, 4, 107 | 3.4 | 16 |
| 106 | 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 |
| 105 | Defective activation of mitogen-activated protein kinase after allogeneic bone marrow transplantation. *Blood*, **1996**, 88, 2334-2341 | 2.2 | 15 |
| 104 | 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 |
| 103 | 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 |

| | | | |
|---|---|---|---|
| 102 | High intrafamilial variability in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy: a case study. *Journal of Endocrinological Investigation*, **2012**, 35, 77-81 | 5.2 | 14 |
| 101 | Brain abscesses in children: an Italian multicentre study. *Epidemiology and Infection*, **2017**, 145, 2848-2855 | 4.3 | 13 |
| 100 | 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 |
| 99 | Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy from the pediatric perspective. *Journal of Endocrinological Investigation*, **2013**, 36, 903-12 | 5.2 | 13 |
| 98 | 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 |
| 97 | 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 |
| 96 | 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 |
| 95 | Brain alteration in a Nude/SCID fetus carrying FOXN1 homozygous mutation. *Journal of the Neurological Sciences*, **2010**, 298, 121-3 | 3.2 | 11 |
| 94 | Progressive deficiencies in blood T cells associated with a 10p12-13 interstitial deletion. *Clinical Immunology and Immunopathology*, **1996**, 80, 9-15 | | 11 |
| 93 | Novel STAT1 gain-of-function mutation and suppurative infections. *Pediatric Allergy and Immunology*, **2016**, 27, 220-3 | 4.2 | 11 |
| 92 | Follow-up and outcome of symptomatic partial or absolute IgA deficiency in children. *European Journal of Pediatrics*, **2019**, 178, 51-60 | 4.1 | 10 |
| 91 | 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 |
| 90 | Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease. *Data in Brief*, **2016**, 7, 311-5 | 1.2 | 9 |
| 89 | 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 |
| 88 | Cutaneous manifestations as presenting sign of autoimmune lymphoproliferative syndrome in childhood. *Dermatology*, **2005**, 210, 336-40 | 4.4 | 9 |
| 87 | Chronic unexplained liver disease in children with primary immunodeficiency syndromes. *Journal of Clinical Gastroenterology*, **1998**, 26, 187-92 | 3 | 9 |
| 86 | 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 |
| 85 | 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 |

| | | | |
|---|---|---|---|
| 84 | Role of the common □chain in cell cycle progression of human malignant cell lines. *International Immunology*, **2012**, 24, 159-67 | 4.9 | 8 |
| 83 | 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 |
| 82 | Prolonged Q-T interval syndrome presenting as idiopathic epilepsy. *Neuropediatrics*, **1983**, 14, 235-6 | 1.6 | 8 |
| 81 | Biweekly Hizentra□ in Primary Immunodeficiency: a Multicenter, Observational Cohort Study (IBIS). *Journal of Clinical Immunology*, **2018**, 38, 602-609 | 5.7 | 7 |
| 80 | 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 |
| 79 | 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 |
| 78 | 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 |
| 77 | 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 |
| 76 | Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. *International Reviews of Immunology*, **2016**, 35, 25-38 | 4.6 | 6 |
| 75 | Two Brothers with Atypical Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. *Frontiers in Immunology*, **2017**, 8, 1892 | 8.4 | 6 |
| 74 | 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 |
| 73 | 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 |
| 72 | Pertussis immunization in HIV-1-infected infants: a model to assess the effects of repeated T cell-dependent antigen administrations on HIV-1 progression. Italian Register for HIV infection in children. *Vaccine*, **2000**, 18, 1203-9 | 4.1 | 6 |
| 71 | 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 |
| 70 | 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 |
| 69 | Human clinical phenotype associated with FOXN1 mutations. *Advances in Experimental Medicine and Biology*, **2009**, 665, 195-206 | 3.6 | 6 |
| 68 | Impaired platelet activation in patients with hereditary deficiency of p47. *British Journal of Haematology*, **2018**, 180, 454-456 | 4.5 | 5 |
| 67 | 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 |

| | | | |
|---|---|---|---|
| 66 | 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 |
| 65 | Molecular evidence for a thymus-independent partial T cell development in a FOXN1-/- athymic human fetus. *PLoS ONE*, **2013**, 8, e81786 | 3.7 | 5 |
| 64 | Immunoregulatory T Subsets in Chronic Active Viral Hepatitis. *Journal of Pediatric Gastroenterology and Nutrition*, **1983**, 2, 229-233 | 2.8 | 5 |
| 63 | 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 |
| 62 | Thymic stromal alterations and genetic disorders of immune system. *Frontiers in Immunology*, **2015**, 6, 81 | 8.4 | 4 |
| 61 | 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 |
| 60 | 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 |
| 59 | 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 |
| 58 | Hyper IgM syndrome presenting as chronic suppurative lung disease. *Italian Journal of Pediatrics*, **2012**, 38, 45 | 3.2 | 4 |
| 57 | 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 |
| 56 | Intergenerational anticipation of disease onset in people with multiple autoimmune syndrome. *Diabetes Research and Clinical Practice*, **2011**, 94, e37-9 | 7.4 | 4 |
| 55 | Chain transducing element: a shared pathway between endocrine and immune system. *Cellular Immunology*, **2011**, 269, 10-5 | 4.4 | 4 |
| 54 | 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 |
| 53 | A solid-phase radioimmunoassay for IgG gliadin antibodies using 125I-labelled staphylococcal protein A. *Journal of Immunological Methods*, **1983**, 63, 163-70 | 2.5 | 4 |
| 52 | Colostral T lymphocytes detected by intracytoplasmic and membrane markers. *Neonatology*, **1982**, 42, 217-21 | 4 | 4 |
| 51 | Clinical Manifestations of 22q11.2 Deletion Syndrome. *Heart Failure Clinics*, **2022**, 18, 155-164 | 3.3 | 4 |
| 50 | Complement system network in cell physiology and in human diseases. *International Reviews of Immunology*, **2021**, 40, 159-170 | 4.6 | 4 |
| 49 | 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 |

| | | | |
|---|---|---|---|
| 48 | Brain migration disorder and T-cell activation deficiency associated with abnormal signaling through TCR/CD3 complex and hyperactivity of Fyn tyrosine kinase. *Neuropediatrics*, **2000**, 31, 265-8 | 1.6 | 3 |
| 47 | Heterogeneity of IgA deficiency in childhood. *Pediatric Allergy and Immunology*, **1991**, 2, 38-40 | 4.2 | 3 |
| 46 | T cell immunodeficiency in a patient with 10p deletion syndrome. *Journal of Pediatrics*, **1989**, 115, 330 | 3.6 | 3 |
| 45 | Defective interleukin-2 production in children with chronic hepatitis B: role of adherent cells. *Journal of Pediatric Gastroenterology and Nutrition*, **1997**, 24, 312-6 | 2.8 | 3 |
| 44 | Defective activation of mitogen-activated protein kinase after allogeneic bone marrow transplantation. *Blood*, **1996**, 88, 2334-41 | 2.2 | 3 |
| 43 | 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 |
| 42 | A case of galactosemia misdiagnosed as cow milk intolerance. *Italian Journal of Pediatrics*, **2012**, 38, 47 | 3.2 | 2 |
| 41 | Effect of prednisone on DR-positive T cells in children with chronic active hepatitis B. *Journal of Pediatric Gastroenterology and Nutrition*, **1989**, 8, 288-91 | 2.8 | 2 |
| 40 | Immunoregulatory functional abnormalities in children affected by HBsAg-positive chronic active hepatitis: role of prostaglandins in T-mediated suppression. *Journal of Pediatric Gastroenterology and Nutrition*, **1986**, 5, 537-41 | 2.8 | 2 |
| 39 | Chronic diarrhea and failure to thrive in an infant with Campylobacter jejuni. *Journal of Pediatric Gastroenterology and Nutrition*, **1984**, 3, 812-4 | 2.8 | 2 |
| 38 | Immunoregulatory T subsets in chronic active viral hepatitis: characterization by monoclonal antibodies. *Journal of Pediatric Gastroenterology and Nutrition*, **1983**, 2, 229-33 | 2.8 | 2 |
| 37 | 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 |
| 36 | Immunoregulatory T Subsets in Chronic Active Viral Hepatitis. *Journal of Pediatric Gastroenterology and Nutrition*, **1983**, 2, 229-233 | 2.8 | 2 |
| 35 | 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 |
| 34 | 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 |
| 33 | Simple Measurement of IgA Predicts Immunity and Mortality in Ataxia-Telangiectasia. *Journal of Clinical Immunology*, **2021**, 41, 1878-1892 | 5.7 | 2 |
| 32 | SCID-like phenotype associated with an inhibitory autoreactive immunoglobulin. *Journal of Investigational Allergology and Clinical Immunology*, **2012**, 22, 67-70 | 2.3 | 2 |
| 31 | Nutritional Status in Agammaglobulinemia: An Italian Multicenter Study. *Journal of Clinical Immunology*, **2015**, 35, 595-7 | 5.7 | 1 |

| | | | |
|---|---|---|---|
| 30 | Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. *Journal of Clinical Medicine*, **2020**, 9, | 5.1 | 1 |
| 29 | The R156H variation in IL-12Rβ1 is not a mutation. *Italian Journal of Pediatrics*, **2013**, 39, 12 | 3.2 | 1 |
| 28 | Acute adrenal failure as the presenting feature of primary antiphospholipid syndrome in a child. *Italian Journal of Pediatrics*, **2012**, 38, 49 | 3.2 | 1 |
| 27 | 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 |
| 26 | Immunodeficiency diagnosis: a Mondrian or Pollock scenario?. *Blood*, **2011**, 118, 5714-6 | 2.2 | 1 |
| 25 | Altered signaling through IL-12 receptor in children with very high serum IgE levels. *Cellular Immunology*, **2010**, 265, 74-9 | 4.4 | 1 |
| 24 | Primary immunodeficiencies: the tip of an iceberg?. *Blood*, **2005**, 105, 2626-2627 | 2.2 | 1 |
| 23 | 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 |
| 22 | 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 |
| 21 | Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. *Archivos De Bronconeumologia*, **2021**, 57, 712-712 | 0.7 | 1 |
| 20 | 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 |
| 19 | 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 |
| 18 | Congenital alopecia and nail dystrophy associated with severe functional T-cell immunodeficiency in two sibs **1996**, 65, 167 | | 1 |
| 17 | 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 |
| 16 | Hypertransaminasemia and fatal lung disease: a case report. *Italian Journal of Pediatrics*, **2013**, 39, 9 | 3.2 | 0 |
| 15 | 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 |
| 14 | Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination.. *Frontiers in Immunology*, **2022**, 13, 845496 | 8.4 | 0 |
| 13 | 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 |

# List of Publications

| | | |
|---|---|---|
| 12 | Networking Between β and GH-R Signaling in the Control of Cell Growth. *Current Signal Transduction Therapy*, **2013**, 8, 67-73 | 0.8 |
| 11 | Altered regulatory mechanisms governing cell survival in children affected with clustering of autoimmune disorders. *Italian Journal of Pediatrics*, **2012**, 38, 42 | 3.2 |
| 10 | Current Role of Leukotriene Receptor Antagonists in Preschool Asthma. *Current Respiratory Medicine Reviews*, **2012**, 8, 391-395 | 0.3 |
| 9 | Multisystemic disease with involvement of immune, endocrine, and neurologic systems. *Journal of Child Neurology*, **1997**, 12, 396-8 | 2.5 |
| 8 | Combined immunodeficiency phenotype associated with inappropriate spontaneous and activation-induced apoptosis. *Clinical and Experimental Immunology*, **1997**, 108, 484-9 | 6.2 |
| 7 | T Cell Immunodeficiency, Congenital Alopecia, and Nail Dystrophy **2022**, 1-6 | |
| 6 | DiGeorge Syndrome **2022**, 1-7 | |
| 5 | Clinical quiz. Crohn's disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2000**, 31, 90, 92 | 2.8 |
| 4 | Oral Thrush and Onychomycosis **2019**, 371-376 | |
| 3 | Recurrent Cold Suppurative Granulomatous Lymphadenitis **2019**, 347-352 | |
| 2 | Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. *Archivos De Bronconeumologia*, **2021**, 57, 712-714 | 0.7 |
| 1 | Mechanisms of immune tolerance breakdown in inborn errors of immunity **2022**, 73-95 | |