## Giuliana Giardino

List of Publications by Year in descending order

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| #  | Article                                                                                                                                                                                                                                                                       | IF  | CITATIONS |
|----|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 1  | Inherited defects in the complement system. Pediatric Allergy and Immunology, 2022, 33, 73-76.                                                                                                                                                                                | 2.6 | 7         |
| 2  | Activated phosphoinositide 3â€dinase delta syndrome (APDS): An update. Pediatric Allergy and<br>Immunology, 2022, 33, 69-72.                                                                                                                                                  | 2.6 | 5         |
| 3  | Primary atopic disorders and chronic skin disease. Pediatric Allergy and Immunology, 2022, 33, 65-68.                                                                                                                                                                         | 2.6 | 4         |
| 4  | Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.                                                                                                                                                                            | 2.4 | 8         |
| 5  | Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination.<br>Frontiers in Immunology, 2022, 13, 845496.                                                                                                                                    | 4.8 | 13        |
| 6  | Progressive Depletion of B and T Lymphocytes in Patients with Ataxia Telangiectasia: Results of the<br>Italian Primary Immunodeficiency Network. Journal of Clinical Immunology, 2022, 42, 783-797.                                                                           | 3.8 | 5         |
| 7  | Mechanisms of immune tolerance breakdown in inborn errors of immunity. , 2022, , 73-95.                                                                                                                                                                                       |     | 0         |
| 8  | 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        |
| 9  | 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         |
| 10 | In Ataxia-Telangiectasia, Oral Betamethasone Administration Ameliorates Lymphocytes Functionality<br>through Modulation of the IL-7/IL-7Rα Axis Paralleling the Neurological Behavior: A Comparative Report<br>of Two Cases. Immunological Investigations, 2021, 50, 295-303. | 2.0 | 3         |
| 11 | Complement system network in cell physiology and in human diseases. International Reviews of<br>Immunology, 2021, 40, 159-170.                                                                                                                                                | 3.3 | 10        |
| 12 | Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult<br>Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.                                                                                                                     | 0.8 | 3         |
| 13 | Inborn errors of immunity with atopic phenotypes: A practical guide for allergists. World Allergy<br>Organization Journal, 2021, 14, 100513.                                                                                                                                  | 3.5 | 25        |
| 14 | 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        |
| 15 | SARS-CoV-2 Infection in the Immunodeficient Host: Necessary and Dispensable Immune Pathways.<br>Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 3237-3248.                                                                                                  | 3.8 | 4         |
| 16 | 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        |
| 17 | Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164.                                                                                                                                                                               | 2.1 | 15        |
| 18 | Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult<br>Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.                                                                                                                     | 0.8 | 1         |

| #  | Article                                                                                                                                                                                                                                                                        | IF  | CITATIONS |
|----|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 19 | CD4+ T Cell Defects in a Mulibrey Patient With Specific TRIM37 Mutations. Frontiers in Immunology, 2020, 11, 1742.                                                                                                                                                             | 4.8 | 5         |
| 20 | Immunological basis of virusâ€host interaction in COVIDâ€19. Pediatric Allergy and Immunology, 2020, 31,<br>75-78.                                                                                                                                                             | 2.6 | 9         |
| 21 | 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        |
| 22 | 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        |
| 23 | Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels.<br>Journal of Clinical Medicine, 2020, 9, 818.                                                                                                                             | 2.4 | 4         |
| 24 | Clinical Phenotype, Immunological Abnormalities, and Genomic Findings in Patients with DiGeorge<br>Spectrum Phenotype without 22q11.2 Deletion. Journal of Allergy and Clinical Immunology: in Practice,<br>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<br>and/or neonatal rash: Evidence from the Italian cohort of 61 patients with elevated IgE. Journal of<br>Allergy and Clinical Immunology: in Practice, 2019, 7, 2072-2075.e4. | 3.8 | 10        |
| 27 | Clinical, Immunological, and Molecular Features of Typical and Atypical Severe Combined<br>Immunodeficiency: Report of the Italian Primary Immunodeficiency Network. Frontiers in Immunology,<br>2019, 10, 1908.                                                               | 4.8 | 41        |
| 28 | Heterozygous FOXN1 Variants Cause Low TRECs and Severe T Cell Lymphopenia, Revealing a Crucial Role<br>of FOXN1 in Supporting Early Thymopoiesis. American Journal of Human Genetics, 2019, 105, 549-561.                                                                      | 6.2 | 52        |
| 29 | Clinical and immunological features in a cohort of patients with partial DiGeorge syndrome followed at a single center. Blood, 2019, 133, 2586-2596.                                                                                                                           | 1.4 | 39        |
| 30 | Asthma: An Undermined State of Immunodeficiency. International Reviews of Immunology, 2019, 38, 70-78.                                                                                                                                                                         | 3.3 | 14        |
| 31 | 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         |
| 32 | Oral Thrush and Onychomycosis. , 2019, , 371-376.                                                                                                                                                                                                                              |     | 0         |
| 33 | Recurrent Cold Suppurative Granulomatous Lymphadenitis. , 2019, , 347-352.                                                                                                                                                                                                     |     | Ο         |
| 34 | Impaired natural killer cell functions in patients with signal transducer and activator of<br>transcription 1 (STAT1) gain-of-function mutations. Journal of Allergy and Clinical Immunology, 2017,<br>140, 553-564.e4.                                                        | 2.9 | 58        |
| 35 | 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         |
| 36 | FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. Journal of Clinical Immunology, 2017, 37, 751-758.                                                                                                                                                       | 3.8 | 36        |

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|----|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 37 | Risk factors predisposing to the development of hypogammaglobulinemia and infections post-Rituximab. International Reviews of Immunology, 2017, 36, 352-359.                                            | 3.3 | 84        |
| 38 | Two Brothers with Atypical UNC13D-Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. Frontiers in Immunology, 2017, 8, 1892.                               | 4.8 | 8         |
| 39 | NADPH Oxidase Deficiency: A Multisystem Approach. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-23.                                                                                          | 4.0 | 29        |
| 40 | Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in<br>Immunology, 2016, 7, 466.                                                                                 | 4.8 | 80        |
| 41 | Unbalanced Immune System: Immunodeficiencies and Autoimmunity. Frontiers in Pediatrics, 2016, 4, 107.                                                                                                   | 1.9 | 26        |
| 42 | Novel <scp>STAT</scp> 1 gainâ€ofâ€function mutation and suppurative infections. Pediatric Allergy and Immunology, 2016, 27, 220-223.                                                                    | 2.6 | 14        |
| 43 | 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        |
| 44 | Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype.<br>Blood, 2016, 127, 3154-3164.                                                                        | 1.4 | 465       |
| 45 | Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease.<br>Data in Brief, 2016, 7, 311-315.                                                                    | 1.0 | 10        |
| 46 | 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        |
| 47 | 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         |
| 48 | Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. International<br>Reviews of Immunology, 2015, 35, 1-14.                                                                | 3.3 | 7         |
| 49 | Severe combined immunodeficiency—an update. Annals of the New York Academy of Sciences, 2015, 1356, 90-106.                                                                                             | 3.8 | 87        |
| 50 | 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         |
| 51 | FOXN1 in Organ Development and Human Diseases. International Reviews of Immunology, 2014, 33, 83-93.                                                                                                    | 3.3 | 40        |
| 52 | Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. Scandinavian<br>Journal of Gastroenterology, 2014, 49, 274-279.                                                       | 1.5 | 31        |
| 53 | Insight into <i>IKBKG</i> / <i>NEMO</i> Locus: Report of New Mutations and Complex Genomic<br>Rearrangements Leading to Incontinentia Pigmenti Disease. Human Mutation, 2014, 35, 165-177.              | 2.5 | 74        |
| 54 | Intergenerational and intrafamilial phenotypic variability in 22q11.2 Deletion syndrome subjects. BMC<br>Medical Genetics, 2014, 15, 1.                                                                 | 2.1 | 48        |

| #  | Article                                                                                                                                                                                                                                                                           | IF  | CITATIONS |
|----|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 55 | The R156H variation in IL-12RÎ <sup>2</sup> 1 is not a mutation. Italian Journal of Pediatrics, 2013, 39, 12.                                                                                                                                                                     | 2.6 | 1         |
| 56 | Steroid treatment in Ataxia-Telangiectasia induces alterations of functional magnetic resonance<br>imaging during prono-supination task. European Journal of Paediatric Neurology, 2013, 17, 135-140.                                                                             | 1.6 | 23        |
| 57 | Non Invasive Assessment of Lung Disease in Ataxia Telangiectasia by High-Field Magnetic Resonance<br>Imaging. Journal of Clinical Immunology, 2013, 33, 1185-1191.                                                                                                                | 3.8 | 21        |
| 58 | 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        |
| 59 | Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.                                                                                                                                                          | 5.4 | 27        |
| 60 | FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. Frontiers in Immunology, 2013, 4, 187.                                                                                                                                                                   | 4.8 | 72        |
| 61 | Alterations of the autoimmune regulator transcription factor and failure of central tolerance:<br>APECED as a model. Expert Review of Clinical Immunology, 2013, 9, 43-51.                                                                                                        | 3.0 | 12        |
| 62 | 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         |
| 63 | Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy: Insights into Genotype-Phenotype<br>Correlation. International Journal of Endocrinology, 2012, 2012, 1-9.                                                                                                         | 1.5 | 42        |
| 64 | 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        |
| 65 | Severe Combined Immunodeficiences: New and Old Scenarios. International Reviews of Immunology, 2012, 31, 43-65.                                                                                                                                                                   | 3.3 | 42        |
| 66 | De novo 13q12.3–q14.11 deletion involving <i>BRCA2</i> gene in a patient with developmental delay,<br>elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an Aâ€T like phenotype.<br>American Journal of Medical Genetics, Part A, 2012, 158A, 2571-2576. | 1.2 | 6         |
| 67 | Altered regulatory mechanisms governing cell survival in children affected with clustering of autoimmune disorders. Italian Journal of Pediatrics, 2012, 38, 42.                                                                                                                  | 2.6 | Ο         |
| 68 | Hyper IgM syndrome presenting as chronic suppurative lung disease. Italian Journal of Pediatrics, 2012, 38, 45.                                                                                                                                                                   | 2.6 | 5         |
| 69 | Interleukin 12 receptor deficiency in a child with recurrent bronchopneumonia and very high IgE<br>levels. Italian Journal of Pediatrics, 2012, 38, 46.                                                                                                                           | 2.6 | 6         |
| 70 | Genetic Basis of Altered Central Tolerance and Autoimmune Diseases: A Lesson from AIRE Mutations.<br>International Reviews of Immunology, 2012, 31, 344-362.                                                                                                                      | 3.3 | 18        |
| 71 | SCID-like phenotype associated with an inhibitory autoreactive immunoglobulin. Journal of Investigational Allergology and Clinical Immunology, 2012, 22, 67-70.                                                                                                                   | 1.3 | 2         |
| 72 | 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         |

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|----|-------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 73 | Intergenerational anticipation of disease onset in people with multiple autoimmune syndrome.<br>Diabetes Research and Clinical Practice, 2011, 94, e37-e39. | 2.8 | 4         |
| 74 | Immunodeficiency diagnosis: a Mondrian or Pollock scenario?. Blood, 2011, 118, 5714-5716.                                                                   | 1.4 | 1         |
| 75 | Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. European<br>Journal of Neurology, 2011, 18, 564-570.             | 3.3 | 62        |
| 76 | Altered signaling through IL-12 receptor in children with very high serum IgE levels. Cellular<br>Immunology, 2010, 265, 74-79.                             | 3.0 | 1         |