Giuliana Giardino

List of Publications by Year
in descending order

76 papers	2,019 citations	257450 24 h-index	265206 42 g-index
80 all docs	80 docs citations	80 times ranked	3272 citing authors

#	Article	IF	Citations
1	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	1.4	465
2	Severe combined immunodeficiencyâ€"an update. Annals of the New York Academy of Sciences, 2015, 1356, 90-106.	3.8	87
3	Risk factors predisposing to the development of hypogammaglobulinemia and infections post-Rituximab. International Reviews of Immunology, 2017, 36, 352-359.	3.3	84
4	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
5	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
6	FOXN1: A Master Regulator Gene of Thymic Epithelial Development Program. Frontiers in Immunology, 2013, 4, 187.	4.8	72
7	Efficacy of very-low-dose betamethasone on neurological symptoms in ataxia-telangiectasia. European Journal of Neurology, 2011, 18, 564-570.	3.3	62
8	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
9	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
10	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
11	Intergenerational and intrafamilial phenotypic variability in 22q11.2 Deletion syndrome subjects. BMC Medical Genetics, 2014, 15, 1.	2.1	48
12	Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy: Insights into Genotype-Phenotype Correlation. International Journal of Endocrinology, 2012, 2012, 1-9.	1.5	42
13	Severe Combined Immunodeficiences: New and Old Scenarios. International Reviews of Immunology, 2012, 31, 43-65.	3.3	42
14	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
15	FOXN1 in Organ Development and Human Diseases. International Reviews of Immunology, 2014, 33, 83-93.	3.3	40
16	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
17	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
18	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

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19	FOXN1 Deficiency: from the Discovery to Novel Therapeutic Approaches. Journal of Clinical Immunology, 2017, 37, 751-758.	3.8	36
20	Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome. Scandinavian Journal of Gastroenterology, 2014, 49, 274-279.	1.5	31
21	NADPH Oxidase Deficiency: A Multisystem Approach. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-23.	4.0	29
22	Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.	5.4	27
23	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
24	Unbalanced Immune System: Immunodeficiencies and Autoimmunity. Frontiers in Pediatrics, 2016, 4, 107.	1.9	26
25	Inborn errors of immunity with atopic phenotypes: A practical guide for allergists. World Allergy Organization Journal, 2021, 14, 100513.	3.5	25
26	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
27	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
28	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
29	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
30	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
31	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
32	Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164.	2.1	15
33	Novel <scp>STAT</scp>1 gainâ€ofâ€function mutation and suppurative infections. Pediatric Allergy and Immunology, 2016, 27, 220-223.	2.6	14
34	Asthma: An Undermined State of Immunodeficiency. International Reviews of Immunology, 2019, 38, 70-78.	3.3	14
35	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
36	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination. Frontiers in Immunology, 2022, 13, 845496.	4.8	13

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37	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
38	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
39	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
40	Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease. Data in Brief, 2016, 7, 311-315.	1.0	10
41	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
42	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
43	Complement system network in cell physiology and in human diseases. International Reviews of Immunology, 2021, 40, 159-170.	3.3	10
44	Immunological basis of virusâ€host interaction in COVIDâ€19. Pediatric Allergy and Immunology, 2020, 31, 75-78.	2.6	9
45	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
46	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
47	Two Brothers with Atypical UNC13D-Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. Frontiers in Immunology, 2017, 8, 1892.	4.8	8
48	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	2.4	8
49	Unraveling the Link Between Ectodermal Disorders and Primary Immunodeficiencies. International Reviews of Immunology, 2015, 35, 1-14.	3.3	7
50	Inherited defects in the complement system. Pediatric Allergy and Immunology, 2022, 33, 73-76.	2.6	7
51	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
52	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
53	Hyper IgM syndrome presenting as chronic suppurative lung disease. Italian Journal of Pediatrics, 2012, 38, 45.	2.6	5
54	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

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55	CD4+ T Cell Defects in a Mulibrey Patient With Specific TRIM37 Mutations. Frontiers in Immunology, 2020, 11, 1742.	4.8	5
56	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
57	Activated phosphoinositide 3â€dinase delta syndrome (APDS): An update. Pediatric Allergy and Immunology, 2022, 33, 69-72.	2.6	5
58	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
59	Intergenerational anticipation of disease onset in people with multiple autoimmune syndrome. Diabetes Research and Clinical Practice, 2011, 94, e37-e39.	2.8	4
60	Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. Journal of Clinical Medicine, 2020, 9, 818.	2.4	4
61	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
62	Primary atopic disorders and chronic skin disease. Pediatric Allergy and Immunology, 2022, 33, 65-68.	2.6	4
63	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
64	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
65	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.	0.8	3
66	SCID-like phenotype associated with an inhibitory autoreactive immunoglobulin. Journal of Investigational Allergology and Clinical Immunology, 2012, 22, 67-70.	1.3	2
67	Altered signaling through IL-12 receptor in children with very high serum IgE levels. Cellular Immunology, 2010, 265, 74-79.	3.0	1
68	Immunodeficiency diagnosis: a Mondrian or Pollock scenario?. Blood, 2011, 118, 5714-5716.	1.4	1
69	The R156H variation in IL-12RÎ²1 is not a mutation. Italian Journal of Pediatrics, 2013, 39, 12.	2.6	1
70	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
71	Respiratory Manifestations in Primary Immunodeficiencies: Findings From a Pediatric and Adult Cohort. Archivos De Bronconeumologia, 2021, 57, 712-714.	0.8	1
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	Altered regulatory mechanisms governing cell survival in children affected with clustering of autoimmune disorders. Italian Journal of Pediatrics, 2012, 38, 42.	2.6	0
74	Oral Thrush and Onychomycosis. , 2019, , 371-376.		0
75	Recurrent Cold Suppurative Granulomatous Lymphadenitis. , 2019, , 347-352.		0
76	Mechanisms of immune tolerance breakdown in inborn errors of immunity. , 2022, , 73-95.		0