

Silvia Clara Giliani

List of Publications by Year in descending order

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136
papers

9,098
citations

47006

47
h-index

42399

92
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138
all docs

138
docs citations

138
times ranked

8885
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). <i>Nature</i> , 1995, 377, 65-68.	27.8	864
2	Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. <i>Nature Genetics</i> , 2000, 25, 343-346.	21.4	629
3	X-Linked Lymphoproliferative Disease. <i>Journal of Experimental Medicine</i> , 2000, 192, 337-346.	8.5	438
4	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. <i>Cell</i> , 1998, 93, 885-896.	28.9	429
5	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. <i>Nature Immunology</i> , 2012, 13, 1178-1186.	14.5	410
6	Mutations of CD40 gene cause an autosomal recessive form of immunodeficiency with hyper IgM. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2001, 98, 12614-12619.	7.1	347
7	Mutations of the Wiskott-Aldrich Syndrome Protein (WASP): hotspots, effect on transcription, and translation and phenotype/genotype correlation. <i>Blood</i> , 2004, 104, 4010-4019.	1.4	308
8	Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. <i>Blood</i> , 2011, 118, 1675-1684.	1.4	296
9	In-utero transplantation of parental CD34 haematopoietic progenitor cells in a patient with X-linked severe combined immunodeficiency (SCIDX1). <i>Lancet, The</i> , 1996, 348, 1484-1487.	13.7	244
10	Interleukin-7 receptor alpha (IL-7Ralpha) deficiency: cellular and molecular bases. Analysis of clinical, immunological, and molecular features in 16 novel patients. <i>Immunological Reviews</i> , 2005, 203, 110-126.	6.0	162
11	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. <i>Journal of Experimental Medicine</i> , 2012, 209, 29-34.	8.5	158
12	Severe cutaneous papillomavirus disease after haemopoietic stem-cell transplantation in patients with severe combined immune deficiency caused by common γ c cytokine receptor subunit or JAK-3 deficiency. <i>Lancet, The</i> , 2004, 363, 2051-2054.	13.7	153
13	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2567-2585.	8.5	146
14	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 656-664.e17.	2.9	140
15	Structural and Functional Basis for JAK3-Deficient Severe Combined Immunodeficiency. <i>Blood</i> , 1997, 90, 3996-4003.	1.4	138
16	IL-21 is the primary common γ chain-binding cytokine required for human B-cell differentiation in vivo. <i>Blood</i> , 2011, 118, 6824-6835.	1.4	132
17	A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1099-1108.e12.	2.9	132
18	Impaired natural and CD16-mediated NK cell cytotoxicity in patients with WAS and XLT: ability of IL-2 to correct NK cell functional defect. <i>Blood</i> , 2004, 104, 436-443.	1.4	130

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19	Defective Expression of CD40 Ligand on T Cells Causes X-Linked Immunodeficiency with Hyper-IgM (HIGM1) Immunological Reviews, 1994, 138, 39-59.	6.0	122
20	Genetic heterogeneity of the excision repair defect associated with trichothiodystrophy. Carcinogenesis, 1993, 14, 1101-1105.	2.8	110
21	Omenn syndrome in an infant with IL7RA gene mutation. Journal of Pediatrics, 2006, 148, 272-274.	1.8	102
22	Ineffective expression of CD40 ligand on cord blood T cells may contribute to poor immunoglobulin production in the newborn. European Journal of Immunology, 1994, 24, 1919-1924.	2.9	99
23	Of genes and phenotypes: the immunological and molecular spectrum of combined immune deficiency. Defects of the gc-JAK3 signaling pathway as a model. Immunological Reviews, 2000, 178, 39-48.	6.0	97
24	Missense mutations of the WASP gene cause intermittent X-linked thrombocytopenia. Blood, 2002, 99, 2268-2269.	1.4	93
25	Expansion of immunoglobulin-secreting cells and defects in B cell tolerance in Rag<i>-dependent immunodeficiency. Journal of Experimental Medicine, 2010, 207, 1541-1554.	8.5	90
26	CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. Trends in Immunology, 1996, 17, 511-516.	7.5	88
27	Characterization of T and B cell repertoire diversity in patients with RAG deficiency. Science Immunology, 2016, 1, .	11.9	88
28	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	2.9	87
29	Severe impairment of IFN- γ and IFN- α responses in cells of a patient with a novel STAT1 splicing mutation. Blood, 2011, 118, 1806-1817.	1.4	84
30	Defect of regulatory T cells in patients with Omenn syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 209-216.	2.9	83
31	Small RNAs derived from lncRNA RNase MRP have gene-silencing activity relevant to human cartilage hair hypoplasia. Human Molecular Genetics, 2014, 23, 368-382.	2.9	83
32	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
33	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Syndrome: Time to Review Diagnostic Criteria?. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3146-3148.	3.6	75
34	Stem cell transplantation for the Wiskott-Aldrich syndrome: a single-center experience confirms efficacy of matched unrelated donor transplantation. Bone Marrow Transplantation, 2006, 38, 671-679.	2.4	74
35	4 Primary immunodeficiency mutation databases. Advances in Genetics, 2001, 43, 103-188.	1.8	70
36	Organization of the human CD40L gene: implications for molecular defects in X chromosome-linked hyper-IgM syndrome and prenatal diagnosis.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 2110-2114.	7.1	68

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37	Toll-like receptor 3 gene polymorphisms and severity of pandemic A/H1N1/2009 influenza in otherwise healthy children. <i>Virology Journal</i> , 2012, 9, 270.	3.4	65
38	Functional analysis of naturally occurring DCLRE1C mutations and correlation with the clinical phenotype of ARTEMIS deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 140-150.e7.	2.9	63
39	Reversible severe combined immunodeficiency phenotype secondary to a mutation of the proton-coupled folate transporter. <i>Clinical Immunology</i> , 2009, 133, 287-294.	3.2	61
40	Expansion of somatically reverted memory CD8+ T cells in patients with X-linked lymphoproliferative disease caused by selective pressure from Epstein-Barr virus. <i>Journal of Experimental Medicine</i> , 2012, 209, 913-924.	8.5	59
41	Long-term follow-up of 168 patients with X-linked agammaglobulinemia reveals increased morbidity and mortality. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 429-437.	2.9	59
42	Variability of clinical and laboratory features among patients with ribonuclease mitochondrial RNA processing endoribonuclease gene mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2008, 122, 1178-1184.	2.9	58
43	Wiskott-Aldrich syndrome protein (WASP) is a tumor suppressor in T cell lymphoma. <i>Nature Medicine</i> , 2019, 25, 130-140.	30.7	57
44	PAX1 is essential for development and function of the human thymus. <i>Science Immunology</i> , 2020, 5, .	11.9	55
45	Defective actin polymerization in EBV-transformed B-cell lines from patients with the Wiskott-Aldrich syndrome. , 1998, 185, 99-107.		51
46	Altered germinal center reaction and abnormal B cell peripheral maturation in PI3KR1-mutated patients presenting with HIGM-like phenotype. <i>Clinical Immunology</i> , 2015, 159, 33-36.	3.2	51
47	Cartilage-hair hypoplasia: molecular basis and heterogeneity of the immunological phenotype. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2008, 8, 534-539.	2.3	50
48	Impaired NK-cell migration in WAS/XLT patients: role of Cdc42/WASp pathway in the control of chemokine-induced β 2 integrin high-affinity state. <i>Blood</i> , 2010, 115, 2818-2826.	1.4	50
49	A novel mutation in the POLE2 gene causing combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 635-638.e1.	2.9	49
50	Modeling altered T-cell development with induced pluripotent stem cells from patients with RAG1-dependent immune deficiencies. <i>Blood</i> , 2016, 128, 783-793.	1.4	45
51	A novel homozygous mutation in recombination activating gene 2 in 2 relatives with different clinical phenotypes: Omenn syndrome and hyper-IgM syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 1414-1416.	2.9	43
52	Development of Autologous T Lymphocytes in Two Males with X-Linked Severe Combined Immune Deficiency: Molecular and Cellular Characterization. <i>Clinical Immunology</i> , 2000, 95, 39-50.	3.2	42
53	Hematopoietic stem cell transplantation in Omenn syndrome: a single-center experience. <i>Bone Marrow Transplantation</i> , 2005, 36, 107-114.	2.4	42
54	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. <i>Frontiers in Immunology</i> , 2019, 10, 316.	4.8	42

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55	Natural Killer Cells from Patients with Recombinase-Activating Gene and Non-Homologous End Joining Gene Defects Comprise a Higher Frequency of CD56 ^{bright} NKG2A ⁺⁺⁺ Cells, and Yet Display Increased Degranulation and Higher Perforin Content. <i>Frontiers in Immunology</i> , 2017, 8, 798.	4.8	41
56	Clinical, Immunological, and Molecular Features of Typical and Atypical Severe Combined Immunodeficiency: Report of the Italian Primary Immunodeficiency Network. <i>Frontiers in Immunology</i> , 2019, 10, 1908.	4.8	41
57	Homozygous DNA ligase IV R278H mutation in mice leads to leaky SCID and represents a model for human LIG4 syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 3024-3029.	7.1	39
58	Selective Laser Melting and Electron Beam Melting of Ti6Al4V for Orthopedic Applications: A Comparative Study on the Applied Building Direction. <i>Materials</i> , 2020, 13, 5584.	2.9	38
59	Induced pluripotent stem cells: A novel frontier in the study of human primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 1400-1407.e4.	2.9	37
60	Adult-onset manifestation of idiopathic T-cell lymphopenia due to a heterozygous RAG1 mutation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1421-1423.	2.9	37
61	Comparison of Common Monogenic Defects in a Large Predominantly Antibody Deficiency Cohort. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 864-878.e9.	3.8	37
62	Reduced thymic output, cell cycle abnormalities, and increased apoptosis of T lymphocytes in patients with cartilage-hair hypoplasia. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 128, 139-146.	2.9	36
63	DNA damage contributes to neurotoxic inflammation in Aicardi-Goutières syndrome astrocytes. <i>Journal of Experimental Medicine</i> , 2022, 219, .	8.5	35
64	Mutations of the X-linked lymphoproliferative disease gene SH2D1A mimicking common variable immunodeficiency. <i>European Journal of Pediatrics</i> , 2002, 161, 656-659.	2.7	34
65	First report of successful stem cell transplantation in a child with CD40 deficiency. <i>Bone Marrow Transplantation</i> , 2007, 40, 279-281.	2.4	34
66	Abnormalities of Thymic Stroma may Contribute to Immune Dysregulation in Murine Models of Leaky Severe Combined Immunodeficiency. <i>Frontiers in Immunology</i> , 2011, 2, .	4.8	34
67	Differential role of nonhomologous end joining factors in the generation, DNA damage response, and myeloid differentiation of human induced pluripotent stem cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 8889-8894.	7.1	34
68	Long term outcome of eight patients with type 1 Leukocyte Adhesion Deficiency (LAD-1): Not only infections, but high risk of autoimmune complications. <i>Clinical Immunology</i> , 2018, 191, 75-80.	3.2	33
69	Damaging-agent sensitivity of Artemis-deficient cell lines. <i>European Journal of Immunology</i> , 2005, 35, 1250-1256.	2.9	30
70	Structural Basis for SH2D1A Mutations in X-Linked Lymphoproliferative Disease. <i>Biochemical and Biophysical Research Communications</i> , 2000, 269, 124-130.	2.1	29
71	Different molecular behavior of CD40 mutants causing hyper-IgM syndrome. <i>Blood</i> , 2010, 116, 5867-5874.	1.4	29
72	Next Generation Sequencing Analysis in Early Onset Dementia Patients. <i>Journal of Alzheimer's Disease</i> , 2019, 67, 243-256.	2.6	29

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73	Mutation analysis by a non-radioactive single-strand conformation polymorphism assay in nine families with X-linked severe combined immunodeficiency (SCIDX1). <i>British Journal of Haematology</i> , 1998, 101, 582-587.	2.5	28
74	Combined Immunodeficiencies Due to Defects in Signal Transduction: Defects of the I^{β}c -JAK3 Signaling Pathway as a Model. <i>Immunobiology</i> , 2000, 202, 106-119.	1.9	28
75	Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. <i>Blood</i> , 2008, 111, 271-274.	1.4	28
76	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1272-1284.	2.9	28
77	Hypomorphic Janus kinase 3 mutations result in a spectrum of immune defects, including partial maternal T-cell engraftment. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1136-1145.	2.9	27
78	A peptide derived from the Wiskott-Aldrich syndrome (WAS) protein-interacting protein (WIP) restores WAS protein level and actin cytoskeleton reorganization in lymphocytes from patients with WAS mutations that disrupt WIP binding. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 998-1005.e2.	2.9	25
79	Broad spectrum of autoantibodies in patients with Wiskott-Aldrich syndrome and X-linked thrombocytopenia. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1401-1404.e3.	2.9	25
80	Reconstitution of T-cell compartment after in utero stem cell transplantation: analysis of T-cell repertoire and thymic output. <i>Haematologica</i> , 2004, 89, 450-61.	3.5	24
81	Monocytes from Wiskott-Aldrich patients differentiate in functional mature dendritic cells with a defect in CD83 expression. <i>European Journal of Immunology</i> , 2001, 31, 3413-3421.	2.9	23
82	Activated Phosphoinositide 3-Kinase Delta Syndrome 1: Clinical and Immunological Data from an Italian Cohort of Patients. <i>Journal of Clinical Medicine</i> , 2020, 9, 3335.	2.4	23
83	Cohort of Iranian Patients with Congenital Agammaglobulinemia: Mutation Analysis and Novel Gene Defects. <i>Expert Review of Clinical Immunology</i> , 2016, 12, 479-486.	3.0	22
84	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 2317-2321.e12.	2.9	21
85	Immunodeficiencies due to defects of class-switch recombination. <i>Immunologic Research</i> , 2007, 38, 68-77.	2.9	20
86	Severe Combined Immunodeficiency in Greek Children over a 20-Year Period. <i>Journal of Clinical Immunology</i> , 2011, 31, 778-783.	3.8	19
87	The Wiskott-Aldrich syndrome: from genotype to phenotype correlation to treatment. <i>Expert Review of Clinical Immunology</i> , 2007, 3, 813-824.	3.0	16
88	Clinical and molecular features of X-linked hyper IgM syndrome – An experience from North India. <i>Clinical Immunology</i> , 2018, 195, 59-66.	3.2	16
89	Severe Combined Immunodeficiency in Serbia and Montenegro Between Years 1986 and 2010: A Single-Center Experience. <i>Journal of Clinical Immunology</i> , 2014, 34, 304-308.	3.8	14
90	Single-center analysis of long-term outcome after hematopoietic cell transplantation in children with congenital severe T cell immunodeficiency. <i>Immunologic Research</i> , 2009, 44, 4-17.	2.9	13

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91	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 1165-1179.e11.	2.9	13
92	The role of induced pluripotent stem cells in research and therapy of primary immunodeficiencies. <i>Current Opinion in Immunology</i> , 2012, 24, 617-624.	5.5	12
93	C to T mutation causing premature termination of CD40 ligand at amino acid 221 in a patient affected by Hyper IgM syndrome. <i>Human Mutation</i> , 1994, 3, 73-75.	2.5	11
94	Characterization of a Novel Alu-Alu Recombination-Mediated Genomic Deletion in the <i>TCIRG1</i> Gene in Five Osteopetrotic Patients. <i>Journal of Bone and Mineral Research</i> , 2009, 24, 162-167.	2.8	11
95	A custom 148 gene-based resequencing chip and the SNP explorer software: new tools to study antibody deficiency. <i>Human Mutation</i> , 2010, 31, 1080-1088.	2.5	11
96	Aerosol Jet® Printing of Poly(3,4-Ethylenedioxythiophene): Poly(Styrenesulfonate) onto Micropatterned Substrates for Neural Cells In Vitro Stimulation. <i>International Journal of Bioprinting</i> , 2021, 8, 504.	3.4	11
97	Novel presentation of Omenn syndrome in association with aniridia. <i>Journal of Allergy and Clinical Immunology</i> , 2009, 123, 966-969.	2.9	10
98	Genetic variation in schlafen genes in a patient with a recapitulation of the murine Elektra phenotype. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1462-1465.e5.	2.9	10
99	Differences Between Plasma and Cerebrospinal Fluid p-tau181 and p-tau231 in Early Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2022, 87, 991-997.	2.6	10
100	Sine causa tetraparesis. <i>Medicine (United States)</i> , 2018, 97, e13893.	1.0	9
101	The Genomic Organization of the Human Transcription Factor 3 (TFE3) Gene. <i>Genomics</i> , 1995, 28, 491-494.	2.9	8
102	Mutation analysis in Wiskott Aldrich syndrome on chorionic villus DNA. <i>Lancet, The</i> , 1995, 346, 641-642.	13.7	8
103	A novel activation-induced cytidine deaminase gene mutation in a Tunisian family with hyper IgM syndrome. <i>European Journal of Pediatrics</i> , 2004, 163, 704-708.	2.7	8
104	Cytokine-mediated signalling and early defects in lymphoid development. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2005, 5, 519-524.	2.3	8
105	Generation of three iPSC lines from fibroblasts of a patient with Aicardi Goutières Syndrome mutated in TREX1. <i>Stem Cell Research</i> , 2019, 41, 101580.	0.7	8
106	Paediatric MAS/HLH caused by a novel monoallelic activating mutation in p110 β . <i>Clinical Immunology</i> , 2020, 219, 108543.	3.2	8
107	Biomarkers and Precision Therapy for Primary Immunodeficiencies: An In Vitro Study Based on Induced Pluripotent Stem Cells From Patients. <i>Clinical Pharmacology and Therapeutics</i> , 2020, 108, 358-367.	4.7	8
108	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. <i>Frontiers in Immunology</i> , 2021, 12, 669943.	4.8	8

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109	Molecular analysis of the XP-D gene in Italian families with patients affected by trichothiodystrophy and xeroderma pigmentosum group D. <i>Mutation Research DNA Repair</i> , 1994, 314, 159-165.	3.7	7
110	Patients' Induced Pluripotent Stem Cells to Model Drug Induced Adverse Events: A Role in Predicting Thiopurine Induced Pancreatitis?. <i>Current Drug Metabolism</i> , 2015, 17, 91-98.	1.2	7
111	Intronic SH2D1A mutation with impaired SAP expression and agammaglobulinemia. <i>Clinical Immunology</i> , 2013, 146, 84-89.	3.2	6
112	Establishment of three iPSC lines from fibroblasts of a patient with Aicardi Goutières syndrome mutated in RNaseH2B. <i>Stem Cell Research</i> , 2019, 41, 101620.	0.7	6
113	Generation of 3 clones of induced pluripotent stem cells (iPSCs) from a patient affected by Autosomal Recessive Osteopetrosis due to mutations in TCIRG1 gene.. <i>Stem Cell Research</i> , 2020, 42, 101660.	0.7	6
114	Transient Decrease of Circulating and Tissue Dendritic Cells in Patients With Mycobacterial Disease and With Partial Dominant IFN γ R1 Deficiency. <i>Frontiers in Immunology</i> , 2020, 11, 1161.	4.8	5
115	Case Report: Hypomorphic Function and Somatic Reversion in DOCK8 Deficiency in One Patient With Two Novel Variants and Sclerosing Cholangitis. <i>Frontiers in Immunology</i> , 2021, 12, 673487.	4.8	5
116	Application of Molecular Analysis to Genetic Counseling in the Wiskott-Alldrich Syndrome (WAS). <i>DNA and Cell Biology</i> , 1993, 12, 645-649.	1.9	4
117	A novel mitochondrial tRNA ^{Ala} gene variant causes chronic progressive external ophthalmoplegia in a patient with Huntington disease. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 6, 70-73.	1.1	4
118	Generation of three isogenic induced Pluripotent Stem Cell lines (iPSCs) from fibroblasts of a patient with Aicardi Goutières Syndrome carrying a c.2471G>A dominant mutation in IFIH1 gene. <i>Stem Cell Research</i> , 2019, 41, 101623.	0.7	4
119	Persistent Infection with Rotavirus Vaccine Strain in Severe Combined Immunodeficiency (SCID) Child: Is Rotavirus Vaccination in SCID Children a Janus Face?. <i>Vaccines</i> , 2019, 7, 185.	4.4	4
120	<i>Pseudomonas aeruginosa</i> severe skin infection in a toddler with X-linked agammaglobulinemia due to a novel BTK mutation. <i>Infezioni in Medicina</i> , 2019, 27, 73-76.	1.1	4
121	Generation of induced pluripotent stem cells (iPSCs) from patient with Cri du Chat Syndrome. <i>Stem Cell Research</i> , 2019, 35, 101393.	0.7	3
122	Incontinentia Pigmenti Associated with Aplasia Cutis Congenita in a Newborn Male with Klinefelter Syndrome: Is the Severity of Neurological Involvement Linked to Skin Manifestations?. <i>Dermatology and Therapy</i> , 2020, 10, 213-220.	3.0	3
123	Generation of induced pluripotent stem cell (iPSC) lines from a Joubert syndrome patient with compound heterozygous mutations in C5orf42 gene. <i>Stem Cell Research</i> , 2020, 49, 102007.	0.7	3
124	Partial T cell defects and expanded CD56 ^{bright} NK cells in an SCID patient carrying hypomorphic mutation in the <i>IL2RG</i> gene. <i>Journal of Leukocyte Biology</i> , 2020, 108, 739-748.	3.3	3
125	Heterozygous Mutation in Adenosine Deaminase Gene in a Patient With Severe Lymphopenia Following Corticosteroid Treatment of Autoimmune Hemolytic Anemia. <i>Frontiers in Pediatrics</i> , 2018, 6, 272.	1.9	2
126	Establishment of three Joubert syndrome-derived induced pluripotent stem cell (iPSC) lines harbouring compound heterozygous mutations in CC2D2A gene. <i>Stem Cell Research</i> , 2021, 54, 102430.	0.7	2

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127	Biocompatibility evaluation of encapsulated silver-based printed circuits for in-vitro long-term sensing devices. <i>Procedia CIRP</i> , 2022, 110, 99-104.	1.9	2
128	Mechanisms of primary immunodeficiencies: from bed-side to bench and back. <i>Drug Discovery Today Disease Mechanisms</i> , 2004, 1, 383-390.	0.8	1
129	Generation of 3 clones of induced pluripotent stem cells (iPSCs) from a patient affected by Crohn's disease. <i>Stem Cell Research</i> , 2019, 40, 101548.	0.7	1
130	IFN- λ levels in ruxolitinib-treated Aicardi-Goutières patient during SARS-CoV-2 infection: A case report. <i>Clinical Immunology</i> , 2021, 227, 108743.	3.2	1
131	Eye model for floaters™ studies: production of 3D printed scaffolds. <i>Progress in Additive Manufacturing</i> , 2022, 7, 1127-1140.	4.8	1
132	Report of the ESID collaborative study on clinical features and molecular analysis in X-linked hyper-IgM syndrome. <i>Molecular Immunology</i> , 1998, 35, 665.	2.2	0
133	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. <i>Journal of Cell Biology</i> , 2012, 196, i1-i1.	5.2	0
134	Immunodeficiency with Multiple Intestinal Atresias (TTC7A)., 2019, , 1-4.		0
135	Immunodeficiency with Multiple Intestinal Atresias (TTC7A)., 2020, , 379-382.		0
136	When a Nontuberculous Mycobacterial Infection Reveals an Error of Immunity. <i>Pediatric Infectious Disease Journal</i> , 2022, Publish Ahead of Print, .	2.0	0