Silvia Clara Giliani

List of Publications by Year
in descending order

136 papers	9,098 citations	47006 47 h-index	42399 92 g-index
138 all docs	138 docs citations	138 times ranked	8885 citing authors

#	Article	IF	Citations
1	When a Nontuberculous Mycobacterial Infection Reveals an Error of Immunity. Pediatric Infectious Disease Journal, 2022, Publish Ahead of Print, .	2.0	0
2	Eye model for floatersâ€™ studies: production of 3D printed scaffolds. Progress in Additive Manufacturing, 2022, 7, 1127-1140.	4.8	1
3	DNA damage contributes to neurotoxic inflammation in Aicardi-GoutiÃ¨res syndrome astrocytes. Journal of Experimental Medicine, 2022, 219, .	8.5	35
4	Differences Between Plasma and Cerebrospinal Fluid p-tau181 and p-tau231 in Early Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2022, 87, 991-997.	2.6	10
5	Biocompatibility evaluation of encapsulated silver-based printed circuits for in-vitro long-term sensing devices. Procedia CIRP, 2022, 110, 99-104.	1.9	2
6	Case Report: Hypomorphic Function and Somatic Reversion in DOCK8 Deficiency in One Patient With Two Novel Variants and Sclerosing Cholangitis. Frontiers in Immunology, 2021, 12, 673487.	4.8	5
7	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. Frontiers in Immunology, 2021, 12, 669943.	4.8	8
8	IFN-Î± levels in ruxolitinib-treatead Aicardi-GoutiÃ¨res patient during SARS-CoV-2 infection: A case report. Clinical Immunology, 2021, 227, 108743.	3.2	1
9	Establishment of three Joubert syndrome-derived induced pluripotent stem cell (iPSC) lines harbouring compound heterozygous mutations in CC2D2A gene. Stem Cell Research, 2021, 54, 102430.	0.7	2
10	Aerosol JetÂ® Printing of Poly(3,4-Ethylenedioxythiophene): Poly(Styrenesulfonate) onto Micropatterned Substrates for Neural Cells In Vitro Stimulation. International Journal of Bioprinting, 2021, 8, 504.	3.4	11
11	Incontinentia Pigmenti Associated with Aplasia Cutis Congenita in a Newborn Male with Klinefelter Syndrome: Is the Severity of Neurological Involvement Linked to Skin Manifestations?. Dermatology and Therapy, 2020, 10, 213-220.	3.0	3
12	Generation of 3 clones of induced pluripotent stem cells (iPSCs) from a patient affected by Autosomal Recessive Osteopetrosis due to mutations in TCIRG1 gene.. Stem Cell Research, 2020, 42, 101660.	0.7	6
13	Activated Phosphoinositide 3-Kinase Delta Syndrome 1: Clinical and Immunological Data from an Italian Cohort of Patients. Journal of Clinical Medicine, 2020, 9, 3335.	2.4	23
14	Paediatric MAS/HLH caused by a novel monoallelic activating mutation in p110Î´. Clinical Immunology, 2020, 219, 108543.	3.2	8
15	Selective Laser Melting and Electron Beam Melting of Ti6Al4V for Orthopedic Applications: A Comparative Study on the Applied Building Direction. Materials, 2020, 13, 5584.	2.9	38
16	Generation of induced pluripotent stem cell (iPSC) lines from a Joubert syndrome patient with compound heterozygous mutations in C5orf42 gene. Stem Cell Research, 2020, 49, 102007.	0.7	3
17	Partial T cell defects and expanded CD56bright NK cells in an SCID patient carrying hypomorphic mutation in the IL2RG gene. Journal of Leukocyte Biology, 2020, 108, 739-748.	3.3	3
18	Long-term follow-up of 168 patients with X-linked agammaglobulinemia reveals increased morbidity and mortality. Journal of Allergy and Clinical Immunology, 2020, 146, 429-437.	2.9	59

#	Article	IF	Citations
19	Biomarkers and Precision Therapy for Primary Immunodeficiencies: An In Vitro Study Based on Induced Pluripotent Stem Cells From Patients. Clinical Pharmacology and Therapeutics, 2020, 108, 358-367.	4.7	8
20	Transient Decrease of Circulating and Tissular Dendritic Cells in Patients With Mycobacterial Disease and With Partial Dominant IFNÎ³R1 Deficiency. Frontiers in Immunology, 2020, 11, 1161.	4.8	5
21	PAX1 is essential for development and function of the human thymus. Science Immunology, 2020, 5, .	11.9	55
22	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. Journal of Allergy and Clinical Immunology, 2020, 146, 1165-1179.e11.	2.9	13
23	Immunodeficiency with Multiple Intestinal Atresias (TTC7A). , 2020, , 379-382.		0
24	Establishment of three iPSC lines from fibroblasts of a patient with Aicardi GoutiÃ¨res syndrome mutated in RNaseH2B. Stem Cell Research, 2019, 41, 101620.	0.7	6
25	Generation of three isogenic induced Pluripotent Stem Cell lines (iPSCs) from fibroblasts of a patient with Aicardi GoutiÃ¨res Syndrome carrying a c.2471G>A dominant mutation in IFIH1 gene. Stem Cell Research, 2019, 41, 101623.	0.7	4
26	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
27	Generation of 3 clones of induced pluripotent stem cells (iPSCs) from a patient affected by Crohn's disease. Stem Cell Research, 2019, 40, 101548.	0.7	1
28	Generation of three iPSC lines from fibroblasts of a patient with Aicardi GoutiÃ¨res Syndrome mutated in TREX1. Stem Cell Research, 2019, 41, 101580.	0.7	8
29	Generation of induced pluripotent stem cells (iPSCs) from patient with Cri du Chat Syndrome. Stem Cell Research, 2019, 35, 101393.	0.7	3
30	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 316.	4.8	42
31	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2317-2321.e12.	2.9	21
32	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
33	Persistent Infection with Rotavirus Vaccine Strain in Severe Combined Immunodeficiency (SCID) Child: Is Rotavirus Vaccination in SCID Children a Janus Face?. Vaccines, 2019, 7, 185.	4.4	4
34	Comparison of Common Monogenic Defects in a Large Predominantly Antibody Deficiency Cohort. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 864-878.e9.	3.8	37
35	Next Generation Sequencing Analysis in Early Onset Dementia Patients. Journal of Alzheimer's Disease, 2019, 67, 243-256.	2.6	29
36	Wiskottâ€“Aldrich syndrome protein (WASP) is a tumor suppressor in T cell lymphoma. Nature Medicine, 2019, 25, 130-140.	30.7	57

#	Article	IF	Citations
37	Immunodeficiency with Multiple Intestinal Atresias (TTC7A). , 2019, , 1-4.		0
38	Pseudomonas aeruginosa severe skin infection in a toddler with X-linked agammaglobulinemia due to a novel BTK mutation. Infezioni in Medicina, 2019, 27, 73-76.	1.1	4
39	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. Journal of Allergy and Clinical Immunology, 2018, 142, 1272-1284.	2.9	28
40	Long term outcome of eight patients with type 1 Leukocyte Adhesion Deficiency (LAD-1): Not only infections, but high risk of autoimmune complications. Clinical Immunology, 2018, 191, 75-80.	3.2	33
41	Sine causa tetraparesis. Medicine (United States), 2018, 97, e13893.	1.0	9
42	Heterozygous Mutation in Adenosine Deaminase Gene in a Patient With Severe Lymphopenia Following Corticosteroid Treatment of Autoimmune Hemolytic Anemia. Frontiers in Pediatrics, 2018, 6, 272.	1.9	2
43	Clinical and molecular features of X-linked hyper IgM syndrome â€“ An experience from North India. Clinical Immunology, 2018, 195, 59-66.	3.2	16
44	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	8.5	146
45	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
46	Natural Killer Cells from Patients with Recombinase-Activating Gene and Non-Homologous End Joining Gene Defects Comprise a Higher Frequency of CD56bright NKG2A+++ Cells, and Yet Display Increased Degranulation and Higher Perforin Content. Frontiers in Immunology, 2017, 8, 798.	4.8	41
47	Characterization of T and B cell repertoire diversity in patients with RAG deficiency. Science Immunology, 2016, 1, .	11.9	88
48	Modeling altered T-cell development with induced pluripotent stem cells from patients with RAG1-dependent immune deficiencies. Blood, 2016, 128, 783-793.	1.4	45
49	A novel mitochondrial tRNAAla gene variant causes chronic progressive external ophthalmoplegia in a patient with Huntington disease. Molecular Genetics and Metabolism Reports, 2016, 6, 70-73.	1.1	4
50	Cohort of Iranian Patients with Congenital Agammaglobulinemia: Mutation Analysis and Novel Gene Defects. Expert Review of Clinical Immunology, 2016, 12, 479-486.	3.0	22
51	A novel mutation in the POLE2 geneÂcausing combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 635-638.e1.	2.9	49
52	Patients' Induced Pluripotent Stem Cells to Model Drug Induced Adverse Events: A Role in Predicting Thiopurine Induced Pancreatitis?. Current Drug Metabolism, 2015, 17, 91-98.	1.2	7
53	Altered germinal center reaction and abnormal B cell peripheral maturation in PI3KR1-mutated patients presenting with HIGM-like phenotype. Clinical Immunology, 2015, 159, 33-36.	3.2	51
54	Functional analysis of naturally occurring DCLRE1C mutations and correlation with the clinical phenotype of ARTEMIS deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 140-150.e7.	2.9	63

#	Article	IF	Citations
55	Broad spectrum of autoantibodies in patients with Wiskott-Aldrich syndrome and X-linked thrombocytopenia. Journal of Allergy and Clinical Immunology, 2015, 136, 1401-1404.e3.	2.9	25
56	A systematic analysis of recombination activity andÂgenotype-phenotype correlation in human recombination-activating gene 1 deficiency. Journal of Allergy and Clinical Immunology, 2014, 133, 1099-1108.e12.	2.9	132
57	Severe Combined Immunodeficiency in Serbia and Montenegro Between Years 1986 and 2010: A Single-Center Experience. Journal of Clinical Immunology, 2014, 34, 304-308.	3.8	14
58	Small RNAs derived from lncRNA RNase MRP have gene-silencing activity relevant to human cartilageâ€“hair hypoplasia. Human Molecular Genetics, 2014, 23, 368-382.	2.9	83
59	Genetic variation in schlafen genes in a patient with a recapitulation of the murine Elektra phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1462-1465.e5.	2.9	10
60	Differential role of nonhomologous end joining factors in the generation, DNA damage response, and myeloid differentiation of human induced pluripotent stem cells. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 8889-8894.	7.1	34
61	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. Journal of Allergy and Clinical Immunology, 2013, 132, 656-664.e17.	2.9	140
62	Intronic SH2D1A mutation with impaired SAP expression and agammaglobulinemia. Clinical Immunology, 2013, 146, 84-89.	3.2	6
63	Adult-onset manifestation of idiopathic T-cell lymphopenia due to a heterozygous RAG1 mutation. Journal of Allergy and Clinical Immunology, 2013, 131, 1421-1423.	2.9	37
64	Hypomorphic Janus kinase 3 mutations result in a spectrum of immune defects, including partial maternal T-cell engraftment. Journal of Allergy and Clinical Immunology, 2013, 131, 1136-1145.	2.9	27
65	Expansion of somatically reverted memory CD8+ T cells in patients with X-linked lymphoproliferative disease caused by selective pressure from Epstein-Barr virus. Journal of Experimental Medicine, 2012, 209, 913-924.	8.5	59
66	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. Journal of Experimental Medicine, 2012, 209, 29-34.	8.5	158
67	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	14.5	410
68	Toll-like receptor 3 gene polymorphisms and severity of pandemic A/H1N1/2009 influenza in otherwise healthy children. Virology Journal, 2012, 9, 270.	3.4	65
69	A novel homozygous mutation in recombination activating gene 2 in 2 relatives with different clinical phenotypes: Omenn syndrome and hyper-IgM syndrome. Journal of Allergy and Clinical Immunology, 2012, 130, 1414-1416.	2.9	43
70	The role of induced pluripotent stem cells in research and therapy of primary immunodeficiencies. Current Opinion in Immunology, 2012, 24, 617-624.	5.5	12
71	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. Journal of Cell Biology, 2012, 196, i1-i1.	5.2	0
72	Induced pluripotent stem cells: AÂnovel frontier in the study of human primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2011, 127, 1400-1407.e4.	2.9	37

#	Article	IF	Citations
73	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. Journal of Allergy and Clinical Immunology, 2011, 127, 998-1005.e2.	2.9	25
74	Reduced thymic output, cell cycle abnormalities, and increased apoptosis of T lymphocytes in patients with cartilage-hair hypoplasia. Journal of Allergy and Clinical Immunology, 2011, 128, 139-146.	2.9	36
75	Abnormalities of Thymic Stroma may Contribute to Immune Dysregulation in Murine Models of Leaky Severe Combined Immunodeficiency. Frontiers in Immunology, 2011, 2, .	4.8	34
76	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. Blood, 2011, 118, 1675-1684.	1.4	296
77	Severe impairment of IFN-Î³ and IFN-Î± responses in cells of a patient with a novel STAT1 splicing mutation. Blood, 2011, 118, 1806-1817.	1.4	84
78	Severe Combined Immunodeficiency in Greek Children over a 20-Year Period. Journal of Clinical Immunology, 2011, 31, 778-783.	3.8	19
79	IL-21 is the primary common Î³ chain-binding cytokine required for human B-cell differentiation in vivo. Blood, 2011, 118, 6824-6835.	1.4	132
80	Impaired NK-cell migration in WAS/XLT patients: role of Cdc42/WASp pathway in the control of chemokine-induced Î²2 integrin high-affinity state. Blood, 2010, 115, 2818-2826.	1.4	50
81	Different molecular behavior of CD40 mutants causing hyper-IgM syndrome. Blood, 2010, 116, 5867-5874.	1.4	29
82	A custom 148 gene-based resequencing chip and the SNP explorer software: new tools to study antibody deficiency. Human Mutation, 2010, 31, 1080-1088.	2.5	11
83	Expansion of immunoglobulin-secreting cells and defects in B cell tolerance in Rag-dependent immunodeficiency. Journal of Experimental Medicine, 2010, 207, 1541-1554.	8.5	90
84	Homozygous DNA ligase IV R278H mutation in mice leads to leaky SCID and represents a model for human LIG4 syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 3024-3029.	7.1	39
85	Defect of regulatory T cells in patients with Omenn syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 209-216.	2.9	83
86	Reversible severe combined immunodeficiency phenotype secondary to a mutation of the proton-coupled folate transporter. Clinical Immunology, 2009, 133, 287-294.	3.2	61
87	Single-center analysis of long-term outcome after hematopoietic cell transplantation in children with congenital severe T cell immunodeficiency. Immunologic Research, 2009, 44, 4-17.	2.9	13
88	Characterization of a Novel Alu-Alu Recombination-Mediated Genomic Deletion in the TCIRG1 Gene in Five Osteopetrotic Patients. Journal of Bone and Mineral Research, 2009, 24, 162-167.	2.8	11
89	Novel presentation of Omenn syndrome in association with aniridia. Journal of Allergy and Clinical Immunology, 2009, 123, 966-969.	2.9	10
90	Variability of clinical and laboratory features among patients with ribonuclease mitochondrial RNA processing endoribonuclease gene mutations. Journal of Allergy and Clinical Immunology, 2008, 122, 1178-1184.	2.9	58

#	Article	IF	Citations
91	Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. Blood, 2008, 111, 271-274.	1.4	28
92	Cartilage-hair hypoplasia: molecular basis and heterogeneity of the immunological phenotype. Current Opinion in Allergy and Clinical Immunology, 2008, 8, 534-539.	2.3	50
93	The Wiskottâ€“Aldrich syndrome: from genotypeâ€“phenotype correlation to treatment. Expert Review of Clinical Immunology, 2007, 3, 813-824.	3.0	16
94	First report of successful stem cell transplantation in a child with CD40 deficiency. Bone Marrow Transplantation, 2007, 40, 279-281.	2.4	34
95	Immunodeficiencies due to defects of class-switch recombination. Immunologic Research, 2007, 38, 68-77.	2.9	20
96	Omenn syndrome in an infant with IL7RA gene mutation. Journal of Pediatrics, 2006, 148, 272-274.	1.8	102
97	Stem cell transplantation for the Wiskottâ€“Aldrich syndrome: a single-center experience confirms efficacy of matched unrelated donor transplantation. Bone Marrow Transplantation, 2006, 38, 671-679.	2.4	74
98	Cytokine-mediated signalling and early defects in lymphoid development. Current Opinion in Allergy and Clinical Immunology, 2005, 5, 519-524.	2.3	8
99	Hematopoietic stem cell transplantation in Omenn syndrome: a single-center experience. Bone Marrow Transplantation, 2005, 36, 107-114.	2.4	42
100	Interleukin-7 receptor alpha (IL-7Ralpha) deficiency: cellular and molecular bases. Analysis of clinical, immunological, and molecular features in 16 novel patients. Immunological Reviews, 2005, 203, 110-126.	6.0	162
101	Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256.	2.9	30
102	A novel activation-induced cytidine deaminase gene mutation in a Tunisian family with hyper IgM syndrome. European Journal of Pediatrics, 2004, 163, 704-708.	2.7	8
103	Mechanisms of primary immunodeficiencies: from bed-side to bench and back. Drug Discovery Today Disease Mechanisms, 2004, 1, 383-390.	0.8	1
104	Severe cutaneous papillomavirus disease after haemopoietic stem-cell transplantation in patients with severe combined immune deficiency caused by common Î³c cytokine receptor subunit or JAK-3 deficiency. Lancet, The, 2004, 363, 2051-2054.	13.7	153
105	Mutations of the Wiskott-Aldrich Syndrome Protein (WASP): hotspots, effect on transcription, and translation and phenotype/genotype correlation. Blood, 2004, 104, 4010-4019.	1.4	308
106	Impaired natural and CD16-mediated NK cell cytotoxicity in patients with WAS and XLT: ability of IL-2 to correct NK cell functional defect. Blood, 2004, 104, 436-443.	1.4	130
107	Reconstitution of T-cell compartment after in utero stem cell transplantation: analysis of T-cell repertoire and thymic output. Haematologica, 2004, 89, 450-61.	3.5	24
108	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Syndrome: Time to Review Diagnostic Criteria?. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3146-3148.	3.6	75

#	Article	IF	Citations
109	Missense mutations of the WASP gene cause intermittent X-linked thrombocytopenia. Blood, 2002, 99, 2268-2269.	1.4	93
110	Mutations of the X-linked lymphoproliferative disease gene SH2D1A mimicking common variable immunodeficiency. European Journal of Pediatrics, 2002, 161, 656-659.	2.7	34
111	4 Primary immunodeficiency mutation databases. Advances in Genetics, 2001, 43, 103-188.	1.8	70
112	Monocytes from Wiskott-Aldrich patients differentiate in functional mature dendritic cells with a defect in CD83 expression. European Journal of Immunology, 2001, 31, 3413-3421.	2.9	23
113	Mutations ofCD40gene cause an autosomal recessive form of immunodeficiency with hyper IgM. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 12614-12619.	7.1	347
114	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
115	Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genetics, 2000, 25, 343-346.	21.4	629
116	X-Linked Lymphoproliferative Disease. Journal of Experimental Medicine, 2000, 192, 337-346.	8.5	438
117	Structural Basis for SH2D1A Mutations in X-Linked Lymphoproliferative Disease. Biochemical and Biophysical Research Communications, 2000, 269, 124-130.	2.1	29
118	Development of Autologous T Lymphocytes in Two Males with X-Linked Severe Combined Immune Deficiency: Molecular and Cellular Characterization. Clinical Immunology, 2000, 95, 39-50.	3.2	42
119	Combined Immunodeficiencies Due to Defects in Signal Transduction: Defects of the Î³c-JAK3 Signaling Pathway as a Model. Immunobiology, 2000, 202, 106-119.	1.9	28
120	Defective actin polymerization in EBV-transformed B-cell lines from patients with the Wiskott-Aldrich syndrome. , 1998, 185, 99-107.		51
121	Mutation analysis by a nonâ€radioactive singleâ€strand conformation polymorphism assay in nine families with Xâ€linked severe combined immunodeficiency (SCIDX1). British Journal of Haematology, 1998, 101, 582-587.	2.5	28
122	Report of the ESID collaborative study on clinical features and molecular analysis in X-linked hyper-IgM syndrome. Molecular Immunology, 1998, 35, 665.	2.2	0
123	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. Cell, 1998, 93, 885-896.	28.9	429
124	Structural and Functional Basis for JAK3-Deficient Severe Combined Immunodeficiency. Blood, 1997, 90, 3996-4003.	1.4	138
125	In-utero transplantation of parental CD34 haematopoietic progenitor cells in a patient with X-linked severe combined immunodeficiency (SCIDX1). Lancet, The, 1996, 348, 1484-1487.	13.7	244
126	CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. Trends in Immunology, 1996, 17, 511-516.	7.5	88

#	Article	IF	Citations
127	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). Nature, 1995, 377, 65-68.	27.8	864
128	The Genomic Organization of the Human Transcription Factor 3 (TFE3) Gene. Genomics, 1995, 28, 491-494.	2.9	8
129	Mutation analysis in Wiskott Aldrich syndrome on chorionic villus DNA. Lancet, The, 1995, 346, 641-642.	13.7	8
130	C to T mutation causing premature termination of CD40 ligand at amino acid 221 in a patient affected by Hyper IgM syndrome. Human Mutation, 1994, 3, 73-75.	2.5	11
131	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
132	Defective Expression of CD40 Ligand on T Cells Causes â€œXâ€Łinked Immunodeficiency with Hyperâ€łgM (HIGM1)â€• Immunological Reviews, 1994, 138, 39-59.	6.0	122
133	Molecular analysis of the XP-D gene in Italian families with patients affected by trichothiodystrophy and xeroderma pigmentosum group D. Mutation Research DNA Repair, 1994, 314, 159-165.	3.7	7
134	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
135	Application of Molecular Analysis to Genetic Counseling in the Wiskottâ€“Aldrich Syndrome (WAS). DNA and Cell Biology, 1993, 12, 645-649.	1.9	4
136	Genetic heterogeneity of the excision repair defect associated with trichothiodystrophy. Carcinogenesis, 1993, 14, 1101-1105.	2.8	110