

Anna Villa

List of Publications by Year in descending order

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179
papers

12,303
citations

28274

55
h-index

28297

105
g-index

182
all docs

182
docs citations

182
times ranked

10904
citing authors

#	ARTICLE	IF	CITATIONS
1	Editing T cell repertoire by thymic epithelial cell-directed gene transfer abrogates risk of type 1 diabetes development. <i>Molecular Therapy - Methods and Clinical Development</i> , 2022, 25, 508-519.	4.1	1
2	Efficacy and safety of anti-CD45 saporin as conditioning agent for RAG deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 309-320.e6.	2.9	27
3	Gut Microbiota-Host Interactions in Inborn Errors of Immunity. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1416.	4.1	18
4	Modeling, optimization, and comparable efficacy of T cell and hematopoietic stem cell gene editing for treating hyper IgM syndrome. <i>EMBO Molecular Medicine</i> , 2021, 13, e13545.	6.9	36
5	Autosomal recessive osteopetrosis: mechanisms and treatments. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	2.4	26
6	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
7	Ablation of collagen VI leads to the release of platelets with altered function. <i>Blood Advances</i> , 2021, 5, 5150-5163.	5.2	5
8	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
9	Innovative Cell-Based Therapies and Conditioning to Cure RAG Deficiency. <i>Frontiers in Immunology</i> , 2020, 11, 607926.	4.8	11
10	Expanded circulating hematopoietic stem/progenitor cells as novel cell source for the treatment of TCIRG1 osteopetrosis. <i>Haematologica</i> , 2020, 106, 74-86.	3.5	20
11	Artificial thymic organoids represent a reliable tool to study T-cell differentiation in patients with severe T-cell lymphopenia. <i>Blood Advances</i> , 2020, 4, 2611-2616.	5.2	65
12	Severe combined immune deficiency. , 2020, , 153-205.		7
13	Chromosome Transplantation: A Possible Approach to Treat Human X-linked Disorders. <i>Molecular Therapy - Methods and Clinical Development</i> , 2020, 17, 369-377.	4.1	10
14	Successful Preclinical Development of Gene Therapy for Recombinase-Activating Gene-1-Deficient SCID. <i>Molecular Therapy - Methods and Clinical Development</i> , 2020, 17, 666-682.	4.1	37
15	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
16	Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. <i>Blood</i> , 2020, 135, 610-619.	1.4	37
17	Pathobiologic Mechanisms of Neurodegeneration in Osteopetrosis Derived From Structural and Functional Analysis of 14 CLC-7 Mutants. <i>Journal of Bone and Mineral Research</i> , 2020, 36, 531-545.	2.8	16
18	Generation of an immunodeficient mouse model of tcirg1-deficient autosomal recessive osteopetrosis. <i>Bone Reports</i> , 2020, 12, 100242.	0.4	4

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19	Absence of Dipeptidyl Peptidase 3 Increases Oxidative Stress and Causes Bone Loss. <i>Journal of Bone and Mineral Research</i> , 2019, 34, 2133-2148.	2.8	32
20	The microbiome and immunodeficiencies: Lessons from rare diseases. <i>Journal of Autoimmunity</i> , 2019, 98, 132-148.	6.5	35
21	Lentiviral gene therapy corrects platelet phenotype and function in patients with Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 825-838.	2.9	50
22	Gene Modification and Three-Dimensional Scaffolds as Novel Tools to Allow the Use of Postnatal Thymic Epithelial Cells for Thymus Regeneration Approaches. <i>Stem Cells Translational Medicine</i> , 2019, 8, 1107-1122.	3.3	31
23	Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 333-336.	2.9	31
24	The RANKL-RANK Axis: A Bone to Thymus Round Trip. <i>Frontiers in Immunology</i> , 2019, 10, 629.	4.8	50
25	Chromosome Transplantation: Correction of the Chronic Granulomatous Disease Defect in Mouse Induced Pluripotent Stem Cells. <i>Stem Cells</i> , 2019, 37, 876-887.	3.2	3
26	Lentiviral haemopoietic stem/progenitor cell gene therapy for treatment of Wiskott-Aldrich syndrome: interim results of a non-randomised, open-label, phase 1/2 clinical study. <i>Lancet Haematology</i> , 2019, 6, e239-e253.	4.6	166
27	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the Clinical Diagnosis of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1763-1770.	3.8	381
28	One Disease, Many Genes: Implications for the Treatment of Osteopetroses. <i>Frontiers in Endocrinology</i> , 2019, 10, 85.	3.5	59
29	Mesenchymal Stromal Cell-Seeded Biomimetic Scaffolds as a Factory of Soluble RANKL in Rankl-Deficient Osteopetrosis. <i>Stem Cells Translational Medicine</i> , 2019, 8, 22-34.	3.3	34
30	<i>RAG</i> gene defects at the verge of immunodeficiency and immune dysregulation. <i>Immunological Reviews</i> , 2019, 287, 73-90.	6.0	44
31	Platelets in Wiskott-Aldrich syndrome: Victims or executioners?. <i>Journal of Leukocyte Biology</i> , 2018, 103, 577-590.	3.3	14
32	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
33	Neutrophils drive type I interferon production and autoantibodies in patients with Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1605-1617.e4.	2.9	21
34	Genetics of Osteopetrosis. <i>Current Osteoporosis Reports</i> , 2018, 16, 13-25.	3.6	84
35	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 928-941.e8.	2.9	28
36	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. <i>Blood</i> , 2018, 132, 2362-2374.	1.4	99

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37	Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. <i>Bone</i> , 2018, 114, 125-136.	2.9	24
38	Hematopoietic stem cell transplantation corrects osteopetrosis in a child carrying a novel homozygous mutation in the FERMT3 gene. <i>Bone</i> , 2017, 97, 126-129.	2.9	17
39	Murine <i>Rankl</i> ^{-/-} Mesenchymal Stromal Cells Display an Osteogenic Differentiation Defect Improved by a RANKL-Expressing Lentiviral Vector. <i>Stem Cells</i> , 2017, 35, 1365-1377.	3.2	18
40	Preclinical modeling highlights the therapeutic potential of hematopoietic stem cell gene editing for correction of SCID-X1. <i>Science Translational Medicine</i> , 2017, 9, .	12.4	176
41	Synonymous Mutations Add a Layer of Complexity in the Diagnosis of Human Osteopetrosis. <i>Journal of Bone and Mineral Research</i> , 2017, 32, 99-105.	2.8	11
42	Soluble Factors on Stage to Direct Mesenchymal Stem Cells Fate. <i>Frontiers in Bioengineering and Biotechnology</i> , 2017, 5, 32.	4.1	53
43	In Vivo Chronic Stimulation Unveils Autoreactive Potential of Wiskott-Aldrich Syndrome Protein-Deficient B Cells. <i>Frontiers in Immunology</i> , 2017, 8, 490.	4.8	10
44	Combined T- and B-Cell Immunodeficiencies. , 2017, , 83-182.		3
45	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED). , 2016, , 436-443.		0
46	RAGs and BUGS: An alliance for autoimmunity. <i>Gut Microbes</i> , 2016, 7, 503-511.	9.8	11
47	Intestinal microbiota sustains inflammation and autoimmunity induced by hypomorphic <i>RAG</i> defects. <i>Journal of Experimental Medicine</i> , 2016, 213, 355-375.	8.5	61
48	Buried in the Middle but Guilty: Intronic Mutations in the <i>TCIRG1</i> Gene Cause Human Autosomal Recessive Osteopetrosis. <i>Journal of Bone and Mineral Research</i> , 2015, 30, 1814-1821.	2.8	39
49	A pre-screening FISH-based method to detect CRISPR/Cas9 off-targets in mouse embryonic stem cells. <i>Scientific Reports</i> , 2015, 5, 12327.	3.3	20
50	IL-10 Critically Modulates B Cell Responsiveness in <i>Rankl</i> ^{-/-} Mice. <i>Journal of Immunology</i> , 2015, 194, 4144-4153.	0.8	8
51	B-cell reconstitution after lentiviral vector-mediated gene therapy in patients with Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 692-702.e2.	2.9	41
52	Targeted Gene Correction in Osteopetrotic-Induced Pluripotent Stem Cells for the Generation of Functional Osteoclasts. <i>Stem Cell Reports</i> , 2015, 5, 558-568.	4.8	21
53	Lentiviral-mediated gene therapy restores B cell tolerance in Wiskott-Aldrich syndrome patients. <i>Journal of Clinical Investigation</i> , 2015, 125, 3941-3951.	8.2	43
54	Chromosome transplantation as a novel approach for correcting complex genomic disorders. <i>Oncotarget</i> , 2015, 6, 35218-35230.	1.8	10

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55	Development of Central Nervous System Autoimmunity Is Impaired in the Absence of Wiskott-Aldrich Syndrome Protein. PLoS ONE, 2014, 9, e86942.	2.5	2
56	Severe Combined Immunodeficiencies. , 2014, , 87-141.		1
57	As Little as Needed: The Extraordinary Case of a Mild Recessive Osteopetrosis Owing to a Novel Splicing Hypomorphic Mutation in the <i>TCIRG1</i> Gene. Journal of Bone and Mineral Research, 2014, 29, 1646-1650.	2.8	22
58	Osteopetrosis mimicking juvenile myelomonocytic leukemia. Pediatrics International, 2014, 56, 779-782.	0.5	10
59	Rag Defects and Thymic Stroma: Lessons from Animal Models. Frontiers in Immunology, 2014, 5, 259.	4.8	21
60	<i>Wiskott-Aldrich</i> syndrome protein deficiency in natural killer and dendritic cells affects antitumor immunity. European Journal of Immunology, 2014, 44, 1039-1045.	2.9	29
61	Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 2014, 59, 122-126.	2.9	26
62	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
63	Recombination-activating gene 1 (<i>Rag1</i>)-deficient mice with severe combined immunodeficiency treated with lentiviral gene therapy demonstrate autoimmune Omenn-like syndrome. Journal of Allergy and Clinical Immunology, 2014, 133, 1116-1123.	2.9	56
64	Reply. Journal of Allergy and Clinical Immunology, 2014, 134, 243-244.	2.9	3
65	Sphingosine-1-phosphate receptors control B-cell migration through signaling components associated with primary immunodeficiencies, chronic lymphocytic leukemia, and multiple sclerosis. Journal of Allergy and Clinical Immunology, 2014, 134, 420-428.e15.	2.9	70
66	Wiskott-Aldrich Syndrome protein deficiency perturbs the homeostasis of B-cell compartment in humans. Journal of Autoimmunity, 2014, 50, 42-50.	6.5	72
67	Osteopetrosis: genetics, treatment and new insights into osteoclast function. Nature Reviews Endocrinology, 2013, 9, 522-536.	9.6	457
68	Lentiviral Hematopoietic Stem Cell Gene Therapy in Patients with Wiskott-Aldrich Syndrome. Science, 2013, 341, 1233-1235.	12.6	900
69	Lentiviral gene transfer of <i>TCIRG1</i> into peripheral blood CD34+ cells restores osteoclast function in infantile malignant osteopetrosis. Bone, 2013, 57, 1-9.	2.9	20
70	Homozygous stop mutation in the <i>SNX10</i> gene in a consanguineous Iraqi boy with osteopetrosis and corpus callosum hypoplasia. European Journal of Medical Genetics, 2013, 56, 32-35.	1.3	30
71	Hypomorphic mutation in the <i>RAG2</i> gene affects dendritic cell distribution and migration. Journal of Leukocyte Biology, 2013, 94, 1221-1230.	3.3	8
72	<i>SNX10</i> mutations define a subgroup of human autosomal recessive osteopetrosis with variable clinical severity. Journal of Bone and Mineral Research, 2013, 28, 1041-1049.	2.8	59

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73	Preclinical Safety and Efficacy of Human CD34+ Cells Transduced With Lentiviral Vector for the Treatment of Wiskott-Aldrich Syndrome. <i>Molecular Therapy</i> , 2013, 21, 175-184.	8.2	72
74	Wiskott-Aldrich syndrome protein-mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. <i>Journal of Experimental Medicine</i> , 2013, 210, 355-374.	8.5	49
75	RANKL Cytokine: From Pioneer of the Osteoimmunology Era to Cure for a Rare Disease. <i>Clinical and Developmental Immunology</i> , 2013, 2013, 1-9.	3.3	30
76	Wiskott-Aldrich syndrome protein-mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. <i>Journal of Cell Biology</i> , 2013, 200, i6-i6.	5.2	0
77	Autoimmunity in Wiskott-Aldrich Syndrome: An Unsolved Enigma. <i>Frontiers in Immunology</i> , 2012, 3, 209.	4.8	110
78	Artemis C-terminal region facilitates V(D)J recombination through its interactions with DNA Ligase IV and DNA-PKcs. <i>Journal of Experimental Medicine</i> , 2012, 209, 955-963.	8.5	51
79	Correction of Murine Rag2 Severe Combined Immunodeficiency by Lentiviral Gene Therapy Using a Codon-optimized RAG2 Therapeutic Transgene. <i>Molecular Therapy</i> , 2012, 20, 1968-1980.	8.2	57
80	Alterations in the adenosine metabolism and CD39/CD73 adenosinergic machinery cause loss of Treg cell function and autoimmunity in ADA-deficient SCID. <i>Blood</i> , 2012, 119, 1428-1439.	1.4	107
81	Anti-CD3 μ mAb improves thymic architecture and prevents autoimmune manifestations in a mouse model of Omenn syndrome: therapeutic implications. <i>Blood</i> , 2012, 120, 1005-1014.	1.4	22
82	Osteopetrosis rescue upon RANKL administration to <i>Rankl</i> ^{-/-} mice: A new therapy for human RANKL-dependent ARO. <i>Journal of Bone and Mineral Research</i> , 2012, 27, 2501-2510.	2.8	44
83	Gene therapy for primary immunodeficiencies: Part 2. <i>Current Opinion in Immunology</i> , 2012, 24, 585-591.	5.5	61
84	A Homozygous Contiguous Gene Deletion in Chromosome 16p13.3 Leads to Autosomal Recessive Osteopetrosis in a Jordanian Patient. <i>Calcified Tissue International</i> , 2012, 91, 250-254.	3.1	7
85	Impaired Osteoblastogenesis in a Murine Model of Dominant Osteogenesis Imperfecta: A New Target for Osteogenesis Imperfecta Pharmacological Therapy. <i>Stem Cells</i> , 2012, 30, 1465-1476.	3.2	59
86	RANK-dependent autosomal recessive osteopetrosis: Characterization of five new cases with novel mutations. <i>Journal of Bone and Mineral Research</i> , 2012, 27, 342-351.	2.8	66
87	Identification of the first deletion in the LRP5 gene in a patient with Autosomal Dominant Osteopetrosis type I. <i>Bone</i> , 2011, 49, 568-571.	2.9	27
88	Lentiviral-mediated gene therapy leads to improvement of B-cell functionality in a murine model of Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 1376-1384.e5.	2.9	34
89	Omenn syndrome does not live by V(D)J recombination alone. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2011, 11, 525-531.	2.3	44
90	Severe Combined Immunodeficiency in Greek Children over a 20-Year Period. <i>Journal of Clinical Immunology</i> , 2011, 31, 778-783.	3.8	19

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91	Omenn Syndrome: inflammation and autoimmunity. <i>Journal of Translational Medicine</i> , 2011, 9, .	4.4	3
92	A new familial sclerosing bone dysplasia. <i>Journal of Bone and Mineral Research</i> , 2010, 25, 676-680.	2.8	6
93	Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. <i>Human Mutation</i> , 2010, 31, E1071-E1080.	2.5	77
94	Homeostatic expansion of autoreactive immunoglobulin-secreting cells in the Rag2 mouse model of Omenn syndrome. <i>Journal of Experimental Medicine</i> , 2010, 207, 1525-1540.	8.5	66
95	Defect of regulatory T cells in patients with Omenn syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2010, 125, 209-216.	2.9	83
96	Revertant T lymphocytes in a patient with Wiskott-Aldrich syndrome: Analysis of function and distribution in lymphoid organs. <i>Journal of Allergy and Clinical Immunology</i> , 2010, 125, 439-448.e8.	2.9	31
97	Human Peripheral Lymphoid Tissues Contain Autoimmune Regulator-Expressing Dendritic Cells. <i>American Journal of Pathology</i> , 2010, 176, 1104-1112.	3.8	101
98	Analysis of mutations from SCID and Omenn syndrome patients reveals the central role of the Rag2 PHD domain in regulating V(D)J recombination. <i>Journal of Clinical Investigation</i> , 2010, 120, 1337-1344.	8.2	31
99	Evidence for Long-term Efficacy and Safety of Gene Therapy for Wiskott-Aldrich Syndrome in Preclinical Models. <i>Molecular Therapy</i> , 2009, 17, 1073-1082.	8.2	77
100	The Wiskott-Aldrich syndrome protein is required for iNKT cell maturation and function. <i>Journal of Experimental Medicine</i> , 2009, 206, 735-742.	8.5	53
101	A single-center experience in 20 patients with infantile malignant osteopetrosis. <i>American Journal of Hematology</i> , 2009, 84, 473-479.	4.1	83
102	Infantile Malignant, Autosomal Recessive Osteopetrosis: The Rich and The Poor. <i>Calcified Tissue International</i> , 2009, 84, 1-12.	3.1	142
103	Impaired gastric acidification negatively affects calcium homeostasis and bone mass. <i>Nature Medicine</i> , 2009, 15, 674-681.	30.7	172
104	Characterization of a Novel Alu-Alu Recombination-Mediated Genomic Deletion in the TCIRG1 Gene in Five Osteopetrotic Patients. <i>Journal of Bone and Mineral Research</i> , 2009, 24, 162-167.	2.8	11
105	Recent advances in understanding the pathophysiology of Wiskott-Aldrich syndrome. <i>Blood</i> , 2009, 113, 6288-6295.	1.4	207
106	ADA-deficient SCID is associated with a specific microenvironment and bone phenotype characterized by RANKL/OPG imbalance and osteoblast insufficiency. <i>Blood</i> , 2009, 114, 3216-3226.	1.4	82
107	Early defects in human T-cell development severely affect distribution and maturation of thymic stromal cells: possible implications for the pathophysiology of Omenn syndrome. <i>Blood</i> , 2009, 114, 105-108.	1.4	135
108	The Wiskott-Aldrich syndrome protein is required for iNKT cell maturation and function. <i>Journal of Cell Biology</i> , 2009, 185, i1-i1.	5.2	0

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109	Prognostic potential of precise molecular diagnosis of Autosomal Recessive Osteopetrosis with respect to the outcome of bone marrow transplantation. <i>Cytotechnology</i> , 2008, 58, 57-62.	1.6	5
110	Genetically determined lymphopenia and autoimmune manifestations. <i>Current Opinion in Immunology</i> , 2008, 20, 318-324.	5.5	22
111	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. <i>American Journal of Human Genetics</i> , 2008, 83, 64-76.	6.2	270
112	Omenn syndrome: Inflammation in leaky severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2008, 122, 1082-1086.	2.9	213
113	Of Omenn and mice. <i>Trends in Immunology</i> , 2008, 29, 133-140.	6.8	16
114	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
115	WASP regulates suppressor activity of human and murine CD4+CD25+FOXP3+ natural regulatory T cells. <i>Journal of Experimental Medicine</i> , 2007, 204, 369-380.	8.5	167
116	The Dissection of Human Autosomal Recessive Osteopetrosis Identifies an Osteoclast-Poor Form Due to RANKL Deficiency. <i>Cell Cycle</i> , 2007, 6, 3027-3033.	2.6	11
117	Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis. <i>European Journal of Medical Genetics</i> , 2007, 50, 188-199.	1.3	28
118	Osteoclast-poor human osteopetrosis due to mutations in the gene encoding RANKL. <i>Nature Genetics</i> , 2007, 39, 960-962.	21.4	346
119	GvHD-associated cytokine polymorphisms do not associate with Omenn syndrome rather than T α ^h B α ^h SCID in patients with defects in RAG genes. <i>Clinical Immunology</i> , 2007, 124, 165-169.	3.2	9
120	Involvement of PLEKHM1 in osteoclastic vesicular transport and osteopetrosis in incisors absent rats and humans. <i>Journal of Clinical Investigation</i> , 2007, 117, 919-930.	8.2	204
121	A hypomorphic R229Q Rag2 mouse mutant recapitulates human Omenn syndrome. <i>Journal of Clinical Investigation</i> , 2007, 117, 1260-1269.	8.2	97
122	Tissue-specific sensitivity to AID expression in transgenic mouse models. <i>Gene</i> , 2006, 377, 150-158.	2.2	18
123	Osteopetroses and immunodeficiencies in humans. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2006, 6, 421-427.	2.3	13
124	Mutations in OSTM1 (Grey Lethal) Define a Particularly Severe Form of Autosomal Recessive Osteopetrosis With Neural Involvement. <i>Journal of Bone and Mineral Research</i> , 2006, 21, 1098-1105.	2.8	97
125	RAG-dependent primary immunodeficiencies. <i>Human Mutation</i> , 2006, 27, 1174-1184.	2.5	122
126	Polymorphisms of the CLCN7 Gene Are Associated With BMD in Women. <i>Journal of Bone and Mineral Research</i> , 2005, 20, 1960-1967.	2.8	31

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127	Ataxia-telangiectasia-mutated dependent phosphorylation of Artemis in response to DNA damage. <i>Cancer Science</i> , 2005, 96, 134-141.	3.9	45
128	Damaging-agent sensitivity of Artemis-deficient cell lines. <i>European Journal of Immunology</i> , 2005, 35, 1250-1256.	2.9	30
129	Vacuolar H ⁺ -ATPase d2 Subunit: Molecular Characterization, Developmental Regulation, and Localization to Specialized Proton Pumps in Kidney and Bone. <i>Journal of the American Society of Nephrology: JASN</i> , 2005, 16, 1245-1256.	6.1	59
130	Rescue of ATP3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation <i>in utero</i>. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 14629-14634.	7.1	58
131	Omenn's syndrome occurring in patients without mutations in recombination activating genes. <i>Clinical Immunology</i> , 2005, 116, 246-256.	3.2	28
132	The genetics of dominant osteopetrosis. <i>Drug Discovery Today Disease Mechanisms</i> , 2005, 2, 503-509.	0.8	2
133	AIRE deficiency in thymus of 2 patients with Omenn syndrome. <i>Journal of Clinical Investigation</i> , 2005, 115, 728-732.	8.2	146
134	AIRE deficiency in thymus of 2 patients with Omenn syndrome. <i>Journal of Clinical Investigation</i> , 2005, 115, 728-732.	8.2	69
135	In Vitro Differentiation of CD14 Cells From Osteopetrotic Subjects: Contrasting Phenotypes With TCIRG1, CLCN7, and Attachment Defects. <i>Journal of Bone and Mineral Research</i> , 2004, 19, 1329-1338.	2.8	31
136	TCIRG1-dependent recessive osteopetrosis: Mutation analysis, functional identification of the splicing defects, and in vitro rescue by U1 snRNA. <i>Human Mutation</i> , 2004, 24, 225-235.	2.5	90
137	Chloride Channel <i>CLCN7</i> Mutations Are Responsible for Severe Recessive, Dominant, and Intermediate Osteopetrosis. <i>Journal of Bone and Mineral Research</i> , 2003, 18, 1740-1747.	2.8	202
138	Grey-lethal mutation induces severe malignant autosomal recessive osteopetrosis in mouse and human. <i>Nature Medicine</i> , 2003, 9, 399-406.	30.7	245
139	Genotype-Phenotype Relationship in Human ATP6i-Dependent Autosomal Recessive Osteopetrosis. <i>American Journal of Pathology</i> , 2003, 162, 57-68.	3.8	97
140	Inhibition of BUB1 results in genomic instability and anchorage-independent growth of normal human fibroblasts. <i>Cancer Research</i> , 2003, 63, 2855-63.	0.9	47
141	Omenn syndrome in the context of other B cell-negative severe combined immunodeficiencies. <i>Israel Medical Association Journal</i> , 2002, 4, 218-21.	0.1	7
142	Recombinase activating gene enzymes of lymphocytes. <i>Current Opinion in Hematology</i> , 2001, 8, 41-46.	2.5	12
143	4 Primary immunodeficiency mutation databases. <i>Advances in Genetics</i> , 2001, 43, 103-188.	1.8	70
144	Identification of anti-herpes simplex virus antibody-producing B cells in a patient with an atypical RAG1 immunodeficiency. <i>Blood</i> , 2001, 98, 1464-1468.	1.4	19

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145	Recombination activating gene and its defects. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2001, 1, 491-495.	2.3	10
146	Prenatal diagnosis of RAG-deficient Omenn syndrome. , 2000, 20, 56-59.		13
147	The genetic and biochemical basis of Omenn syndrome. <i>Immunological Reviews</i> , 2000, 178, 64-74.	6.0	56
148	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
149	Lymphoid abnormalities in CD40 ligand transgenic mice suggest the need for tight regulation in gene therapy approaches to hyper immunoglobulin M (IgM) syndrome. <i>Cancer Gene Therapy</i> , 2000, 7, 1299-1306.	4.6	49
150	Mutations in Conserved Regions of the Predicted RAG2 Kelch Repeats Block Initiation of V(D)J Recombination and Result in Primary Immunodeficiencies. <i>Molecular and Cellular Biology</i> , 2000, 20, 5653-5664.	2.3	58
151	Molecular Modeling of the Jak3 Kinase Domains and Structural Basis for Severe Combined Immunodeficiency. <i>Clinical Immunology</i> , 2000, 96, 108-118.	3.2	23
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