Anna Villa

List of Publications by Year
in descending order

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179 papers	12,303 citations	28242 55 h-index	28275 105 g-index
182 all docs	182 docs citations	182 times ranked	10904 citing authors

#	Article	IF	Citations
1	Lentiviral Hematopoietic Stem Cell Gene Therapy in Patients with Wiskott-Aldrich Syndrome. Science, 2013, 341, 1233151.	6.0	900
2	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). Nature, 1995, 377, 65-68.	13.7	864
3	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.	9.4	629
4	Osteopetrosis: genetics, treatment and new insights into osteoclast function. Nature Reviews Endocrinology, 2013, 9, 522-536.	4.3	457
5	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. Cell, 1998, 93, 885-896.	13.5	429
6	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the ClinicalÂDiagnosis of Inborn Errors of Immunity. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1763-1770.	2.0	381
7	Osteoclast-poor human osteopetrosis due to mutations in the gene encoding RANKL. Nature Genetics, 2007, 39, 960-962.	9.4	346
8	Xâ€“linked thrombocytopenia and Wiskottâ€“Aldrich syndrome are allelic diseases with mutations in the WASP gene. Nature Genetics, 1995, 9, 414-417.	9.4	274
9	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	2.6	270
10	Grey-lethal mutation induces severe malignant autosomal recessive osteopetrosis in mouse and human. Nature Medicine, 2003, 9, 399-406.	15.2	245
11	Omenn syndrome: Inflammation in leaky severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2008, 122, 1082-1086.	1.5	213
12	Recent advances in understanding the pathophysiology of Wiskott-Aldrich syndrome. Blood, 2009, 113, 6288-6295.	0.6	207
13	Involvement of PLEKHM1 in osteoclastic vesicular transport and osteopetrosis in incisors absent rats and humans. Journal of Clinical Investigation, 2007, 117, 919-930.	3.9	204
14	Chloride Channel ClCN7 Mutations Are Responsible for Severe Recessive, Dominant, and Intermediate Osteopetrosis. Journal of Bone and Mineral Research, 2003, 18, 1740-1747.	3.1	202
15	Preclinical modeling highlights the therapeutic potential of hematopoietic stem cell gene editing for correction of SCID-X1. Science Translational Medicine, 2017, 9, .	5.8	176
16	Impaired gastric acidification negatively affects calcium homeostasis and bone mass. Nature Medicine, 2009, 15, 674-681.	15.2	172
17	WASP regulates suppressor activity of human and murine CD4+CD25+FOXP3+ natural regulatory T cells. Journal of Experimental Medicine, 2007, 204, 369-380.	4.2	167
18	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. Lancet Haematology,the, 2019, 6, e239-e253.	2.2	166

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19	AIRE deficiency in thymus of 2 patients with Omenn syndrome. Journal of Clinical Investigation, 2005, 115, 728-732.	3.9	146
20	Infantile Malignant, Autosomal Recessive Osteopetrosis: The Rich and The Poor. Calcified Tissue International, 2009, 84, 1-12.	1.5	142
21	Structural and Functional Basis for JAK3-Deficient Severe Combined Immunodeficiency. Blood, 1997, 90, 3996-4003.	0.6	138
22	Early defects in human T-cell development severely affect distribution and maturation of thymic stromal cells: possible implications for the pathophysiology of Omenn syndrome. Blood, 2009, 114, 105-108.	0.6	135
23	RAG-dependent primary immunodeficiencies. Human Mutation, 2006, 27, 1174-1184.	1.1	122
24	Early and multifocal tumors in breast, salivary, Harderian and epididymal tissues developed in MMTY-Neu transgenic mice. Cancer Letters, 1992, 64, 203-209.	3.2	111
25	Autoimmunity in Wiskottâ€“Aldrich Syndrome: An Unsolved Enigma. Frontiers in Immunology, 2012, 3, 209.	2.2	110
26	Alterations in the adenosine metabolism and CD39/CD73 adenosinergic machinery cause loss of Treg cell function and autoimmunity in ADA-deficient SCID. Blood, 2012, 119, 1428-1439.	0.6	107
27	Human Peripheral Lymphoid Tissues Contain Autoimmune Regulator-Expressing Dendritic Cells. American Journal of Pathology, 2010, 176, 1104-1112.	1.9	101
28	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood, 2018, 132, 2362-2374.	0.6	99
29	Genotype-Phenotype Relationship in Human ATP6i-Dependent Autosomal Recessive Osteopetrosis. American Journal of Pathology, 2003, 162, 57-68.	1.9	97
30	Mutations in OSTM1 (Grey Lethal) Define a Particularly Severe Form of Autosomal Recessive Osteopetrosis With Neural Involvement. Journal of Bone and Mineral Research, 2006, 21, 1098-1105.	3.1	97
31	A hypomorphic R229Q Rag2 mouse mutant recapitulates human Omenn syndrome. Journal of Clinical Investigation, 2007, 117, 1260-1269.	3.9	97
32	TCIRG1-dependent recessive osteopetrosis: Mutation analysis, functional identification of the splicing defects, andin vitro rescue by U1 snRNA. Human Mutation, 2004, 24, 225-235.	1.1	90
33	CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. Trends in Immunology, 1996, 17, 511-516.	7.5	88
34	Genetics of Osteopetrosis. Current Osteoporosis Reports, 2018, 16, 13-25.	1.5	84
35	A singleâ€center experience in 20 patients with infantile malignant osteopetrosis. American Journal of Hematology, 2009, 84, 473-479.	2.0	83
36	Defect of regulatory T cells in patients with Omenn syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 209-216.	1.5	83

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37	ADA-deficient SCID is associated with a specific microenvironment and bone phenotype characterized by RANKL/OPG imbalance and osteoblast insufficiency. Blood, 2009, 114, 3216-3226.	0.6	82
38	Intrathymic Restriction and Peripheral Expansion of the T-Cell Repertoire in Omenn Syndrome. Blood, 1999, 94, 3468-3478.	0.6	79
39	Evidence for Long-term Efficacy and Safety of Gene Therapy for Wiskottâ€“Aldrich Syndrome in Preclinical Models. Molecular Therapy, 2009, 17, 1073-1082.	3.7	77
40	Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Human Mutation, 2010, 31, E1071-E1080.	1.1	77
41	Omenn syndrome: a disorder of Rag1 and Rag2 genes. Journal of Clinical Immunology, 1999, 19, 87-97.	2.0	73
42	The RAG1/RAG2 Complex Constitutes a 3â€² Flap Endonuclease. Molecular Cell, 1999, 4, 935-947.	4.5	73
43	Preclinical Safety and Efficacy of Human CD34+ Cells Transduced With Lentiviral Vector for the Treatment of Wiskott-Aldrich Syndrome. Molecular Therapy, 2013, 21, 175-184.	3.7	72
44	Wiskottâ€“Aldrich Syndrome protein deficiency perturbs the homeostasis of B-cell compartment in humans. Journal of Autoimmunity, 2014, 50, 42-50.	3.0	72
45	4 Primary immunodeficiency mutation databases. Advances in Genetics, 2001, 43, 103-188.	0.8	70
46	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.	1.5	70
47	AIRE deficiency in thymus of 2 patients with Omenn syndrome. Journal of Clinical Investigation, 2005, 115, 728-732.	3.9	69
48	Homeostatic expansion of autoreactive immunoglobulin-secreting cells in the Rag2 mouse model of Omenn syndrome. Journal of Experimental Medicine, 2010, 207, 1525-1540.	4.2	66
49	RANK-dependent autosomal recessive osteopetrosis: Characterization of five new cases with novel mutations. Journal of Bone and Mineral Research, 2012, 27, 342-351.	3.1	66
50	Artificial thymic organoids represent a reliable tool to study T-cell differentiation in patients with severe T-cell lymphopenia. Blood Advances, 2020, 4, 2611-2616.	2.5	65
51	Gene therapy for primary immunodeficiencies: Part 2. Current Opinion in Immunology, 2012, 24, 585-591.	2.4	61
52	Intestinal microbiota sustains inflammation and autoimmunity induced by hypomorphic RAG defects. Journal of Experimental Medicine, 2016, 213, 355-375.	4.2	61
53	Vacuolar H+-ATPase d2 Subunit: Molecular Characterization, Developmental Regulation, and Localization to Specialized Proton Pumps in Kidney and Bone. Journal of the American Society of Nephrology: JASN, 2005, 16, 1245-1256.	3.0	59
54	Impaired Osteoblastogenesis in a Murine Model of Dominant Osteogenesis Imperfecta: A New Target for Osteogenesis Imperfecta Pharmacological Therapy. Stem Cells, 2012, 30, 1465-1476.	1.4	59

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55	SNX10 mutations define a subgroup of human autosomal recessive osteopetrosis with variable clinical severity. Journal of Bone and Mineral Research, 2013, 28, 1041-1049.	3.1	59
56	One Disease, Many Genes: Implications for the Treatment of Osteopetroses. Frontiers in Endocrinology, 2019, 10, 85.	1.5	59
57	RAG and RAG defects. Current Opinion in Immunology, 1999, 11, 435-442.	2.4	58
58	Mutations in Conserved Regions of the Predicted RAG2 Kelch Repeats Block Initiation of V(D)J Recombination and Result in Primary Immunodeficiencies. Molecular and Cellular Biology, 2000, 20, 5653-5664.	1.1	58
59	Rescue of ATPa3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation in utero. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 14629-14634.	3.3	58
60	Correction of Murine Rag2 Severe Combined Immunodeficiency by Lentiviral Gene Therapy Using a Codon-optimized RAG2 Therapeutic Transgene. Molecular Therapy, 2012, 20, 1968-1980.	3.7	57
61	The genetic and biochemical basis of Omenn syndrome. Immunological Reviews, 2000, 178, 64-74.	2.8	56
62	Recombination-activating gene 1 (Rag1)â€“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.	1.5	56
63	The Wiskott-Aldrich syndrome protein is required for iNKT cell maturation and function. Journal of Experimental Medicine, 2009, 206, 735-742.	4.2	53
64	Soluble Factors on Stage to Direct Mesenchymal Stem Cells Fate. Frontiers in Bioengineering and Biotechnology, 2017, 5, 32.	2.0	53
65	Artemis C-terminal region facilitates V(D)J recombination through its interactions with DNA Ligase IV and DNA-PKcs. Journal of Experimental Medicine, 2012, 209, 955-963.	4.2	51
66	Lentiviral gene therapy corrects platelet phenotype and function in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2019, 144, 825-838.	1.5	50
67	The RANKL-RANK Axis: A Bone to Thymus Round Trip. Frontiers in Immunology, 2019, 10, 629.	2.2	50
68	Lymphoid abnormalities in CD40 ligand transgenic mice suggest the need for tight regulation in gene therapy approaches to hyper immunoglobulin M (IgM) syndrome. Cancer Gene Therapy, 2000, 7, 1299-1306.	2.2	49
69	Wiskott-Aldrich syndrome proteinâ€“mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. Journal of Experimental Medicine, 2013, 210, 355-374.	4.2	49
70	Inhibition of BUB1 results in genomic instability and anchorage-independent growth of normal human fibroblasts. Cancer Research, 2003, 63, 2855-63.	0.4	47
71	Ataxia-telangiectasia-mutated dependent phosphorylation of Artemis in response to DNA damage. Cancer Science, 2005, 96, 134-141.	1.7	45
72	Omenn syndrome does not live by V(D)J recombination alone. Current Opinion in Allergy and Clinical Immunology, 2011, 11, 525-531.	1.1	44

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73	Osteopetrosis rescue upon RANKL administration to Ranklâ^'â^' mice: A new therapy for human RANKL-dependent ARO. Journal of Bone and Mineral Research, 2012, 27, 2501-2510.	3.1	44
74	<scp>RAG</scp> gene defects at the verge of immunodeficiency and immune dysregulation. Immunological Reviews, 2019, 287, 73-90.	2.8	44
75	Lentiviral-mediated gene therapy restores B cell tolerance in Wiskott-Aldrich syndrome patients. Journal of Clinical Investigation, 2015, 125, 3941-3951.	3.9	43
76	B-cell reconstitution after lentiviral vectorâ€"mediated gene therapy in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2015, 136, 692-702.e2.	1.5	41
77	Buried in the Middle but Guilty: Intronic Mutations in the TCIRG1 Gene Cause Human Autosomal Recessive Osteopetrosis. Journal of Bone and Mineral Research, 2015, 30, 1814-1821.	3.1	39
78	Development of Autologous, Oligoclonal, Poorly Functioning T Lymphocytes in a Patient With Autosomal Recessive Severe Combined Immunodeficiency Caused by Defects of the Jak3 Tyrosine Kinase. Blood, 1998, 91, 949-955.	0.6	37
79	Successful Preclinical Development of Gene Therapy for Recombinase-Activating Gene-1-Deficient SCID. Molecular Therapy - Methods and Clinical Development, 2020, 17, 666-682.	1.8	37
80	Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. Blood, 2020, 135, 610-619.	0.6	37
81	Modeling, optimization, and comparable efficacy of T cell and hematopoietic stem cell gene editing for treating hyperâ€IgM syndrome. EMBO Molecular Medicine, 2021, 13, e13545.	3.3	36
82	The microbiome and immunodeficiencies: Lessons from rare diseases. Journal of Autoimmunity, 2019, 98, 132-148.	3.0	35
83	Lentiviral-mediated gene therapy leads to improvement of B-cell functionality in a murine model of Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2011, 127, 1376-1384.e5.	1.5	34
84	Mesenchymal Stromal Cell-Seeded Biomimetic Scaffolds as a Factory of Soluble RANKL in Rankl-Deficient Osteopetrosis. Stem Cells Translational Medicine, 2019, 8, 22-34.	1.6	34
85	Absence of Dipeptidyl Peptidase 3 Increases Oxidative Stress and Causes Bone Loss. Journal of Bone and Mineral Research, 2019, 34, 2133-2148.	3.1	32
86	In Vitro Differentiation of CD14 Cells From Osteopetrotic Subjects: Contrasting Phenotypes With TCIRG1, CLCN7, and Attachment Defects. Journal of Bone and Mineral Research, 2004, 19, 1329-1338.	3.1	31
87	Polymorphisms of the CLCN7 Gene Are Associated With BMD in Women. Journal of Bone and Mineral Research, 2005, 20, 1960-1967.	3.1	31
88	Revertant T lymphocytes in a patient with Wiskott-Aldrich syndrome: Analysis of function and distribution in lymphoid organs. Journal of Allergy and Clinical Immunology, 2010, 125, 439-448.e8.	1.5	31
89	Gene Modification and Three-Dimensional Scaffolds as Novel Tools to Allow the Use of Postnatal Thymic Epithelial Cells for Thymus Regeneration Approaches. Stem Cells Translational Medicine, 2019, 8, 1107-1122.	1.6	31
90	Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. Journal of Allergy and Clinical Immunology, 2019, 144, 333-336.	1.5	31

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91	Analysis of mutations from SCID and Omenn syndrome patients reveals the central role of the Rag2 PHD domain in regulating V(D)J recombination. Journal of Clinical Investigation, 2010, 120, 1337-1344.	3.9	31
92	Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256.	1.6	30
93	Homozygous stop mutation in the SNX10 gene in a consanguineous Iraqi boy with osteopetrosis and corpus callosum hypoplasia. European Journal of Medical Genetics, 2013, 56, 32-35.	0.7	30
94	RANKL Cytokine: From Pioneer of the Osteoimmunology Era to Cure for a Rare Disease. Clinical and Developmental Immunology, 2013, 2013, 1-9.	3.3	30
95	<scp>W</scp>iskottâ€“<scp>A</scp>ldrich syndrome protein deficiency in natural killer and dendritic cells affects antitumor immunity. European Journal of Immunology, 2014, 44, 1039-1045.	1.6	29
96	Omenn's syndrome occurring in patients without mutations in recombination activating genes. Clinical Immunology, 2005, 116, 246-256.	1.4	28
97	Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis. European Journal of Medical Genetics, 2007, 50, 188-199.	0.7	28
98	Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. Blood, 2008, 111, 271-274.	0.6	28
99	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. Journal of Allergy and Clinical Immunology, 2018, 142, 1272-1284.	1.5	28
100	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. Journal of Allergy and Clinical Immunology, 2018, 142, 928-941.e8.	1.5	28
101	Identification of the first deletion in the LRP5 gene in a patient with Autosomal Dominant Osteopetrosis type I. Bone, 2011, 49, 568-571.	1.4	27
102	Efficacy and safety of anti-CD45â€“saporin as conditioning agent for RAG deficiency. Journal of Allergy and Clinical Immunology, 2021, 147, 309-320.e6.	1.5	27
103	Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 2014, 59, 122-126.	1.4	26
104	Autosomal recessive osteopetrosis: mechanisms and treatments. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	26
105	Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. Bone, 2018, 114, 125-136.	1.4	24
106	Molecular Modeling of the Jak3 Kinase Domains and Structural Basis for Severe Combined Immunodeficiency. Clinical Immunology, 2000, 96, 108-118.	1.4	23
107	Establishment and characterization of a new mammary adenocarcinoma cell line derived from MMTV neu transgenic mice. Breast Cancer Research and Treatment, 1998, 47, 171-180.	1.1	22
108	Genetically determined lymphopenia and autoimmune manifestations. Current Opinion in Immunology, 2008, 20, 318-324.	2.4	22

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109	Anti-CD3Îµ mAb improves thymic architecture and prevents autoimmune manifestations in a mouse model of Omenn syndrome: therapeutic implications. Blood, 2012, 120, 1005-1014.	0.6	22
110	As Little as Needed: The Extraordinary Case of a Mild Recessive Osteopetrosis Owing to a Novel Splicing Hypomorphic Mutation in the TCIRG1 Gene. Journal of Bone and Mineral Research, 2014, 29, 1646-1650.	3.1	22
111	Rag Defects and Thymic Stroma: Lessons from Animal Models. Frontiers in Immunology, 2014, 5, 259.	2.2	21
112	Targeted Gene Correction in Osteopetrotic-Induced Pluripotent Stem Cells for the Generation of Functional Osteoclasts. Stem Cell Reports, 2015, 5, 558-568.	2.3	21
113	Neutrophils drive type I interferon production and autoantibodies in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2018, 142, 1605-1617.e4.	1.5	21
114	Lentiviral gene transfer of TCIRG1 into peripheral blood CD34+ cells restores osteoclast function in infantile malignant osteopetrosis. Bone, 2013, 57, 1-9.	1.4	20
115	A pre-screening FISH-based method to detect CRISPR/Cas9 off-targets in mouse embryonic stem cells. Scientific Reports, 2015, 5, 12327.	1.6	20
116	Expanded circulating hematopoietic stem/progenitor cells as novel cell source for the treatment of TCIRG1 osteopetrosis. Haematologica, 2020, 106, 74-86.	1.7	20
117	ZNF75: Isolation of a cDNA Clone of the KRAB Zinc Finger Gene Subfamily Mapped in YACs 1 Mb Telomeric of HPRT. Genomics, 1993, 18, 223-229.	1.3	19
118	Identification of antiâ€“herpes simplex virus antibodyâ€“producing B cells in a patient with an atypical RAG1 immunodeficiency. Blood, 2001, 98, 1464-1468.	0.6	19
119	Severe Combined Immunodeficiency in Greek Children over a 20-Year Period. Journal of Clinical Immunology, 2011, 31, 778-783.	2.0	19
120	Severe combined immune deficiencies due to defects of the common ? chain-JAK3 signaling pathway. Seminars in Immunopathology, 1998, 19, 401-415.	4.0	18
121	Tissue-specific sensitivity to AID expression in transgenic mouse models. Gene, 2006, 377, 150-158.	1.0	18
122	Murine Ranklâ^'/â^' Mesenchymal Stromal Cells Display an Osteogenic Differentiation Defect Improved by a RANKL-Expressing Lentiviral Vector. Stem Cells, 2017, 35, 1365-1377.	1.4	18
123	Gut Microbiotaâ€“Host Interactions in Inborn Errors of Immunity. International Journal of Molecular Sciences, 2021, 22, 1416.	1.8	18
124	Hematopoietic stem cell transplantation corrects osteopetrosis in a child carrying a novel homozygous mutation in the FERMT3 gene. Bone, 2017, 97, 126-129.	1.4	17
125	A new mutation (TTR Ala-47) in the transthyretin gene associated with hereditary amyloidosis. Human Mutation, 1994, 4, 61-64.	1.1	16
126	Of Omenn and mice. Trends in Immunology, 2008, 29, 133-140.	2.9	16

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127	Pathobiologic Mechanisms of Neurodegeneration in Osteopetrosis Derived From Structural and Functional Analysis of 14 ClC-7 Mutants. Journal of Bone and Mineral Research, 2020, 36, 531-545.	3.1	16
128	The Chromosome Localization and the HCF Repeats of the Human Host Cell Factor Gene (HCFC1) Are Conserved in the Mouse Homologue. Genomics, 1996, 32, 277-280.	1.3	15
129	Severe Combined Immunodeficiency in Serbia and Montenegro Between Years 1986 and 2010: A Single-Center Experience. Journal of Clinical Immunology, 2014, 34, 304-308.	2.0	14
130	Platelets in Wiskott-Aldrich syndrome: Victims or executioners?. Journal of Leukocyte Biology, 2018, 103, 577-590.	1.5	14
131	Prenatal diagnosis of RAG-deficient Omenn syndrome. , 2000, 20, 56-59.		13
132	Osteopetroses and immunodeficiencies in humans. Current Opinion in Allergy and Clinical Immunology, 2006, 6, 421-427.	1.1	13
133	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. Journal of Allergy and Clinical Immunology, 2020, 146, 1165-1179.e11.	1.5	13
134	Lymphoid abnormalities in CD40 ligand transgenic mice suggest the need for tight regulation in gene therapy approaches to hyper immunoglobulin M (IgM) syndrome. Cancer Gene Therapy, 2000, 7, 1299-1306.	2.2	13
135	Isolation of a zinc finger motif (ZNF75) mapping on chromosome Xq26. Genomics, 1992, 13, 1231-1236.	1.3	12
136	The ZNF75 Zinc Finger Gene Subfamily: Isolation and Mapping of the Four Members in Humans and Great Apes. Genomics, 1996, 35, 312-320.	1.3	12
137	Recombinase activating gene enzymes of lymphocytes. Current Opinion in Hematology, 2001, 8, 41-46.	1.2	12
138	The Exonâ€“Intron Structure of HumanLHX1 Gene. Biochemical and Biophysical Research Communications, 1996, 229, 494-497.	1.0	11
139	The Dissection of Human Autosomal Recessive Osteopetrosis Identifies an Osteoclast-Poor Form Due to RANKL Deficiency. Cell Cycle, 2007, 6, 3027-3033.	1.3	11
140	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.	3.1	11
141	RAGs and BUGS: An alliance for autoimmunity. Gut Microbes, 2016, 7, 503-511.	4.3	11
142	Synonymous Mutations Add a Layer of Complexity in the Diagnosis of Human Osteopetrosis. Journal of Bone and Mineral Research, 2017, 32, 99-105.	3.1	11
143	Innovative Cell-Based Therapies and Conditioning to Cure RAG Deficiency. Frontiers in Immunology, 2020, 11, 607926.	2.2	11
144	Recombination activating gene and its defects. Current Opinion in Allergy and Clinical Immunology, 2001, 1, 491-495.	1.1	10

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145	Osteopetrosis mimicking juvenile myelomonocytic leukemia. Pediatrics International, 2014, 56, 779-782.	0.2	10
146	In Vivo Chronic Stimulation Unveils Autoreactive Potential of Wiskottâ€“Aldrich Syndrome Protein-Deficient B Cells. Frontiers in Immunology, 2017, 8, 490.	2.2	10
147	Chromosome Transplantation: A Possible Approach to Treat Human X-linked Disorders. Molecular Therapy - Methods and Clinical Development, 2020, 17, 369-377.	1.8	10
148	Chromosome transplantation as a novel approach for correcting complex genomic disorders. Oncotarget, 2015, 6, 35218-35230.	0.8	10
149	GvHD-associated cytokine polymorphisms do not associate with Omenn syndrome rather than Tâ˜Bâ˜ SCID in patients with defects in RAG genes. Clinical Immunology, 2007, 124, 165-169.	1.4	9
150	The Genomic Organization of the Human Transcription Factor 3 (TFE3) Gene. Genomics, 1995, 28, 491-494.	1.3	8
151	Hypomorphic mutation in the RAG2 gene affects dendritic cell distribution and migration. Journal of Leukocyte Biology, 2013, 94, 1221-1230.	1.5	8
152	IL-10 Critically Modulates B Cell Responsiveness in Ranklâ˜/â˜ Mice. Journal of Immunology, 2015, 194, 4144-4153.	0.4	8
153	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. Frontiers in Immunology, 2021, 12, 669943.	2.2	8
154	Human CD40L Gene Maps between DXS144E and DXS300 in Xq26. Genomics, 1994, 22, 249-251.	1.3	7
155	A Homozygous Contiguous Gene Deletion in Chromosome 16p13.3 Leads to Autosomal Recessive Osteopetrosis in a Jordanian Patient. Calcified Tissue International, 2012, 91, 250-254.	1.5	7
156	Severe combined immune deficiency. , 2020, , 153-205.		7
157	Omenn syndrome in the context of other B cell-negative severe combined immunodeficiencies. Israel Medical Association Journal, 2002, 4, 218-21.	0.1	7
158	Preimplantation embryo sexing by polymerase chain reaction amplification of the sry gene on single mouse blastomeres. Genetic Analysis, Techniques and Applications, 1993, 10, 147-149.	1.5	6
159	A new familial sclerosing bone dysplasia. Journal of Bone and Mineral Research, 2010, 25, 676-680.	3.1	6
160	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.	0.3	6
161	Prognostic potential of precise molecular diagnosis of Autosomal Recessive Osteopetrosis with respect to the outcome of bone marrow transplantation. Cytotechnology, 2008, 58, 57-62.	0.7	5
162	Ablation of collagen VI leads to the release of platelets with altered function. Blood Advances, 2021, 5, 5150-5163.	2.5	5

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163	Generation of an immunodeficient mouse model of tcirg1-deficient autosomal recessive osteopetrosis. Bone Reports, 2020, 12, 100242.	0.2	4
164	Omenn Syndrome: inflammation and autoimmunity. Journal of Translational Medicine, 2011, 9, .	1.8	3
165	Reply. Journal of Allergy and Clinical Immunology, 2014, 134, 243-244.	1.5	3
166	Chromosome Transplantation: Correction of the Chronic Granulomatous Disease Defect in Mouse Induced Pluripotent Stem Cells. Stem Cells, 2019, 37, 876-887.	1.4	3
167	Combined T- and B-Cell Immunodeficiencies. , 2017, , 83-182.		3
168	RAG MUTATIONS IN SEVERE COMBINED IMMUNODEFICIENCY AND OMENN'S SYNDROME. Immunology and Allergy Clinics of North America, 2000, 20, 129-142.	0.7	2
169	The genetics of dominant osteopetrosis. Drug Discovery Today Disease Mechanisms, 2005, 2, 503-509.	0.8	2
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