

Jean-Pierre de Villartay

List of Publications by Year in descending order

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139
papers

10,616
citations

31949

53
h-index

33869

99
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144
all docs

144
docs citations

144
times ranked

9811
citing authors

#	ARTICLE	IF	CITATIONS
1	Sustained Correction of X-Linked Severe Combined Immunodeficiency by ex Vivo Gene Therapy. <i>New England Journal of Medicine</i> , 2002, 346, 1185-1193.	13.9	1,075
2	Artemis, a Novel DNA Double-Strand Break Repair/V(D)J Recombination Protein, Is Mutated in Human Severe Combined Immune Deficiency. <i>Cell</i> , 2001, 105, 177-186.	13.5	817
3	Cernunnos, a Novel Nonhomologous End-Joining Factor, Is Mutated in Human Immunodeficiency with Microcephaly. <i>Cell</i> , 2006, 124, 287-299.	13.5	640
4	Impairment of immunity to <i>Candida</i> and <i>Mycobacterium</i> in humans with bi-allelic RORC mutations. <i>Science</i> , 2015, 349, 606-613.	6.0	366
5	Metallo-beta-lactamase fold within nucleic acids processing enzymes: the beta-CASP family. <i>Nucleic Acids Research</i> , 2002, 30, 3592-3601.	6.5	288
6	Long-term outcome after hematopoietic stem cell transplantation of a single-center cohort of 90 patients with severe combined immunodeficiency. <i>Blood</i> , 2009, 113, 4114-4124.	0.6	220
7	Severe combined immunodeficiency. A model disease for molecular immunology and therapy. <i>Immunological Reviews</i> , 2005, 203, 98-109.	2.8	212
8	Identical mutations in RAG1 or RAG2 genes leading to defective V(D)J recombinase activity can cause either T-B δ severe combined immune deficiency or Omenn syndrome. <i>Blood</i> , 2001, 97, 2772-2776.	0.6	190
9	Partial T and B lymphocyte immunodeficiency and predisposition to lymphoma in patients with hypomorphic mutations in Artemis. <i>Journal of Clinical Investigation</i> , 2003, 111, 381-387.	3.9	186
10	Severe combined immunodeficiency and microcephaly in siblings with hypomorphic mutations in DNA ligase IV. <i>European Journal of Immunology</i> , 2006, 36, 224-235.	1.6	182
11	A novel immunodeficiency associated with hypomorphic RAG1 mutations and CMV infection. <i>Journal of Clinical Investigation</i> , 2005, 115, 3291-3299.	3.9	177
12	Deletion of the human T-cell receptor β -gene by a site-specific recombination. <i>Nature</i> , 1988, 335, 170-174.	13.7	176
13	A Founder Mutation in Artemis, an SNM1-Like Protein, Causes SCID in Athabaskan-Speaking Native Americans. <i>Journal of Immunology</i> , 2002, 168, 6323-6329.	0.4	162
14	JAK2 stimulates homologous recombination and genetic instability: potential implication in the heterogeneity of myeloproliferative disorders. <i>Blood</i> , 2008, 112, 1402-1412.	0.6	159
15	Restricted heterogeneity of T lymphocytes in combined immunodeficiency with hypereosinophilia (Omenn's syndrome).. <i>Journal of Clinical Investigation</i> , 1991, 87, 1352-1359.	3.9	158
16	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</i> , The, 2004, 363, 2051-2054.	6.3	153
17	Human RTEL1 deficiency causes Hoyeraal-Hreidarsson syndrome with short telomeres and genome instability. <i>Human Molecular Genetics</i> , 2013, 22, 3239-3249.	1.4	150
18	SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID. <i>Blood</i> , 2014, 123, 281-289.	0.6	150

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19	Defect in Rearrangement of the Most 5â€² TCRâ€² Jâ€² Following Targeted Deletion of T Early 1â€² (TEA): Implications for TCR 1â€² Locus Accessibility. <i>Immunity</i> , 1996, 5, 331-342.	6.6	149
20	A Human Severe Combined Immunodeficiency (SCID) Condition with Increased Sensitivity to Ionizing Radiations and Impaired V(D)J Rearrangements Defines a New DNA Recombination/Repair Deficiency. <i>Journal of Experimental Medicine</i> , 1998, 188, 627-634.	4.2	143
21	Mucosal-associated invariant T cellâ€²-rich congenic mouse strain allows functional evaluation. <i>Journal of Clinical Investigation</i> , 2015, 125, 4171-4185.	3.9	143
22	V(D)J and immunoglobulin class switch recombinations: a paradigm to study the regulation of DNA end-joining. <i>Oncogene</i> , 2007, 26, 7780-7791.	2.6	136
23	Role for DNA repair factor XRCC4 in immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2007, 204, 1717-1727.	4.2	132
24	Whole-exome sequencing identifies Coronin-1A deficiency in 3 siblings with immunodeficiency and EBV-associated B-cell lymphoproliferation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1594-1603.e9.	1.5	127
25	Structural characterization of filaments formed by human Xrcc4â€²Cernunnos/XLF complex involved in nonhomologous DNA end-joining. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 12663-12668.	3.3	126
26	Independent mutations of the human CD3â€²Î¼ gene resulting in a T cell receptor/CD3 complex immunodeficiency. <i>Nature Genetics</i> , 1993, 3, 77-81.	9.4	122
27	Inherited CD70 deficiency in humans reveals a critical role for the CD70â€²CD27 pathway in immunity to Epstein-Barr virus infection. <i>Journal of Experimental Medicine</i> , 2017, 214, 73-89.	4.2	122
28	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 1991-2006.	3.9	115
29	Cernunnos Interacts with the XRCC4â€²DNA-ligase IV Complex and Is Homologous to the Yeast Nonhomologous End-joining Factor Nej1*. <i>Journal of Biological Chemistry</i> , 2006, 281, 13857-13860.	1.6	112
30	Composition and dosage of a multipartite enhancer cluster control developmental expression of <i>Ihh</i> (Indian hedgehog). <i>Nature Genetics</i> , 2017, 49, 1539-1545.	9.4	107
31	T Cell Receptor Î³ Gene Rearrangement and T Early 1â€² (TEA) Expression in Immature 1â€²2 Lineage Thymocytes: Implications for 1â€²2/1â€²1 Lineage Commitment. <i>Immunity</i> , 1996, 4, 37-45.	6.6	88
32	A new gene involved in DNA double-strand break repair and V(D)J recombination is located on human chromosome 10p. <i>Human Molecular Genetics</i> , 2000, 9, 583-588.	1.4	85
33	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulatorâ€²dependent autoimmunity. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1578-1588.e5.	1.5	84
34	Polymerase Î¼1 mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature (â€²FELS syndromeâ€²). <i>Journal of Experimental Medicine</i> , 2012, 209, 2323-2330.	4.2	83
35	Increased radiosensitivity of granulocyte macrophage colony-forming units and skin fibroblasts in human autosomal recessive severe combined immunodeficiency.. <i>Journal of Clinical Investigation</i> , 1993, 91, 1214-1218.	3.9	82
36	The Metallo-Î²-Lactamase/Î²-CASP Domain of Artemis Constitutes the Catalytic Core for V(D)J Recombination. <i>Journal of Experimental Medicine</i> , 2004, 199, 315-321.	4.2	79

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37	Highly restricted human T cell repertoire in peripheral blood and tissue-infiltrating lymphocytes in Omenn's syndrome.. <i>Journal of Clinical Investigation</i> , 1998, 102, 312-321.	3.9	79
38	Diagnosis of Fanconi anemia in patients with bone marrow failure. <i>Haematologica</i> , 2009, 94, 487-495.	1.7	77
39	Gene therapy of RAG-2 ^Δ /Δ ⁺ mice: sustained correction of the immunodeficiency. <i>Blood</i> , 2002, 100, 3942-3949.	0.6	76
40	Interplay between Ku, Artemis, and the DNA-dependent Protein Kinase Catalytic Subunit at DNA Ends. <i>Journal of Biological Chemistry</i> , 2006, 281, 27784-27793.	1.6	76
41	Function of Apollo (SNM1B) at telomere highlighted by a splice variant identified in a patient with Hoyeraal-Hreidarsson syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 10097-10102.	3.3	76
42	ROR γ^3 , a thymus-specific isoform of the orphan nuclear receptor ROR γ , is up-regulated by signaling through the pre-T cell receptor and binds to the TEA promoter. <i>European Journal of Immunology</i> , 1999, 29, 4072-4080.	1.6	73
43	Long-term immune reconstitution in RAG-1-deficient mice treated by retroviral gene therapy: a balance between efficiency and toxicity. <i>Blood</i> , 2006, 107, 63-72.	0.6	64
44	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1970-1985.e4.	2.0	64
45	Cernunnos Deficiency Reduces Thymocyte Life Span and Alters the T Cell Repertoire in Mice and Humans. <i>Molecular and Cellular Biology</i> , 2013, 33, 701-711.	1.1	63
46	An <i>in vivo</i> genetic reversion highlights the crucial role of Myb-Like, SWIRM, and MPN domains 1 (MYSM1) in human hematopoiesis and lymphocyte differentiation. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1619-1626.e5.	1.5	63
47	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.	1.5	63
48	Coronin 1 Regulates Cognition and Behavior through Modulation of cAMP/Protein Kinase A Signaling. <i>PLoS Biology</i> , 2014, 12, e1001820.	2.6	62
49	The mechanisms of immune diversification and their disorders. <i>Nature Reviews Immunology</i> , 2003, 3, 962-972.	10.6	59
50	The Repair of DNA Damages/Modifications During the Maturation of the Immune System: Lessons from Human Primary Immunodeficiency Disorders and Animal Models. <i>Advances in Immunology</i> , 2005, 87, 237-295.	1.1	58
51	Artemis Phosphorylated by DNA-dependent Protein Kinase Associates Preferentially with Discrete Regions of Chromatin. <i>Journal of Molecular Biology</i> , 2006, 358, 1200-1211.	2.0	58
52	Stable and Functional Lymphoid Reconstitution in Artemis-deficient Mice Following Lentiviral Artemis Gene Transfer Into Hematopoietic Stem Cells. <i>Molecular Therapy</i> , 2008, 16, 1490-1499.	3.7	58
53	V(D)J Recombination Deficiencies. <i>Advances in Experimental Medicine and Biology</i> , 2009, 650, 46-58.	0.8	58
54	RAG2 and XLF/Cernunnos interplay reveals a novel role for the RAG complex in DNA repair. <i>Nature Communications</i> , 2016, 7, 10529.	5.8	57

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55	Correction of Fas (CD95) deficiency by haploidentical bone marrow transplantation. <i>European Journal of Immunology</i> , 1997, 27, 2043-2047.	1.6	51
56	Phosphorylation of Artemis following irradiation-induced DNA damage. <i>European Journal of Immunology</i> , 2004, 34, 3146-3155.	1.6	51
57	Lack of detectable defect in DNA double-strand break repair and DNA-dependent protein kinase activity in radiosensitive human severe combined immunodeficiency fibroblasts. <i>European Journal of Immunology</i> , 1996, 26, 1118-1122.	1.6	49
58	A primary immunodeficiency characterized by defective immunoglobulin class switch recombination and impaired DNA repair. <i>Journal of Experimental Medicine</i> , 2007, 204, 1207-1216.	4.2	47
59	Three-dimensional Clustering of Human RAG2 Gene Mutations in Severe Combined Immune Deficiency. <i>Journal of Biological Chemistry</i> , 2000, 275, 12672-12675.	1.6	45
60	Delineation of the Xrcc4-interacting Region in the Globular Head Domain of Cernunnos/XLF. <i>Journal of Biological Chemistry</i> , 2010, 285, 26475-26483.	1.6	44
61	Cernunnos influences human immunoglobulin class switch recombination and may be associated with B cell lymphomagenesis. <i>Journal of Experimental Medicine</i> , 2012, 209, 291-305.	4.2	44
62	Somatic genetic rescue of a germline ribosome assembly defect. <i>Nature Communications</i> , 2021, 12, 5044.	5.8	44
63	Functional characterization of the promoter for the human germ-line T cell receptor J β (TEA) transcript. <i>European Journal of Immunology</i> , 1993, 23, 1294-1298.	1.6	43
64	Requirement for XLF/Cernunnos in alignment-based gap filling by DNA polymerases δ and ϵ for nonhomologous end joining in human whole-cell extracts. <i>Nucleic Acids Research</i> , 2009, 37, 4055-4062.	6.5	43
65	Primary Microcephaly, Impaired DNA Replication, and Genomic Instability Caused by Compound Heterozygous ATR Mutations. <i>Human Mutation</i> , 2013, 34, 374-384.	1.1	43
66	PROMIS1: AAT-cell receptor β signature associated with immunodeficiencies caused by V(D)J recombination defects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 325-334.e2.	1.5	43
67	Artemis sheds new light on V(D)J recombination. <i>Immunological Reviews</i> , 2004, 200, 142-155.	2.8	40
68	PAXX and Xlf interplay revealed by impaired CNS development and immunodeficiency of double KO mice. <i>Cell Death and Differentiation</i> , 2018, 25, 444-452.	5.0	40
69	Restoration of Human B-cell Differentiation Into NOD-SCID Mice Engrafted With Gene-corrected CD34+ Cells Isolated From Artemis or RAG1-deficient Patients. <i>Molecular Therapy</i> , 2008, 16, 396-403.	3.7	39
70	Germ-line transcription and methylation status of the TCR-J β locus in its accessible configuration. <i>European Journal of Immunology</i> , 1997, 27, 1619-1625.	1.6	38
71	NHP2 deficiency impairs rRNA biogenesis and causes pulmonary fibrosis and H α veraala β Hreidarsson syndrome. <i>Human Molecular Genetics</i> , 2020, 29, 907-922.	1.4	38
72	Normal T cell receptor V β 2 usage in a primary immunodeficiency associated with HLA class II deficiency. <i>European Journal of Immunology</i> , 1993, 23, 928-934.	1.6	37

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73	Immunologic aspects of patients with disseminated bacille Calmette-Guerin disease in north-west of Iran. <i>Italian Journal of Pediatrics</i> , 2009, 35, 42.	1.0	34
74	Heterogeneous telomere defects in patients with severe forms of dyskeratosis congenita. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 129, 473-482.e3.	1.5	34
75	A nonsense mutation in the DNA repair factor Hebo causes mild bone marrow failure and microcephaly. <i>Journal of Experimental Medicine</i> , 2016, 213, 1011-1028.	4.2	34
76	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2013, 210, 2503-2513.	4.2	33
77	Impaired telomere integrity and rRNA biogenesis in PARN-deficient patients and knockout models. <i>EMBO Molecular Medicine</i> , 2019, 11, e10201.	3.3	31
78	High level transient gene expression in human lymphoid cells by SV40 large T antigen boost. <i>Nucleic Acids Research</i> , 1992, 20, 245-250.	6.5	30
79	Reduced immunoglobulin class switch recombination in the absence of Artemis. <i>Blood</i> , 2009, 114, 3601-3609.	0.6	29
80	Impaired lymphocyte function and differentiation in CTPS1-deficient patients result from a hypomorphic homozygous mutation. <i>JCI Insight</i> , 2020, 5, .	2.3	29
81	CD34-positive early human thymocytes: T cell receptor and cytokine receptor gene expression. <i>European Journal of Immunology</i> , 1995, 25, 2471-2478.	1.6	27
82	DNA repair and the immune system: From V(D)J recombination to aging lymphocytes. <i>European Journal of Immunology</i> , 2007, 37, S71-S82.	1.6	27
83	Congenital defects in V(D)J recombination. <i>British Medical Bulletin</i> , 2015, 114, 157-167.	2.7	27
84	DNA replication stress triggers rapid DNA replication fork breakage by Artemis and XPF. <i>PLoS Genetics</i> , 2018, 14, e1007541.	1.5	27
85	Lack of selective \hat{V}^2 deletion in peripheral CD4+ T cells of human immunodeficiency virus-infected infants. <i>European Journal of Immunology</i> , 1993, 23, 2041-2044.	1.6	26
86	The V(D)J Recombination/DNA Repair Factor Artemis Belongs to the Metallo- β -Lactamase Family and Constitutes a Critical Developmental Checkpoint of the Lymphoid System. <i>Annals of the New York Academy of Sciences</i> , 2003, 987, 150-157.	1.8	26
87	Human regulator of telomere elongation helicase 1 (RTEL1) is required for the nuclear and cytoplasmic trafficking of pre-U2 RNA. <i>Nucleic Acids Research</i> , 2015, 43, 1834-1847.	6.5	26
88	Translesion DNA synthesis-assisted non-homologous end-joining of complex double-strand breaks prevents loss of DNA sequences in mammalian cells. <i>Nucleic Acids Research</i> , 2009, 37, 6737-6745.	6.5	25
89	Tissue-specific Activity of the \hat{I}^3c Chain Gene Promoter Depends upon an Ets Binding Site and Is Regulated by GA-binding Protein. <i>Journal of Biological Chemistry</i> , 1996, 271, 14849-14855.	1.6	24
90	Cernunnos-XLF, a recently identified non-homologous end-joining factor required for the development of the immune system. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2006, 6, 416-420.	1.1	24

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91	T early alpha (TEA) regulates initial TCRVAJA rearrangements and leads to TCRJA coincidence. European Journal of Immunology, 2001, 31, 2080-2086.	1.6	23
92	Human and animal models of V(D)J recombination deficiency. Current Opinion in Immunology, 2003, 15, 592-598.	2.4	23
93	Self-tolerance to host and donor following HLA-mismatched bone marrow transplantation. European Journal of Immunology, 1986, 16, 117-122.	1.6	22
94	Activation induces apoptosis in Herpesvirus saimiri-transformed T cells independent of CD95 (Fas). Tj ETQq0 0 0 rgBT/Overlock 10 Tf 50	1.6	22
95	Two SCID cases with Cernunnos/XLF deficiency successfully treated by hematopoietic stem cell transplantation. Pediatric Transplantation, 2012, 16, E167-71.	0.5	22
96	Atypical combined immunodeficiency due to Artemis defect: A case presenting as hyperimmunoglobulin M syndrome and with LGLL. Molecular Immunology, 2013, 56, 354-357.	1.0	21
97	Coupling DNA Damage and Repair: an Essential Safeguard during Programmed DNA Double-Strand Breaks?. Trends in Cell Biology, 2020, 30, 87-96.	3.6	20
98	The differentiation of human pro-thymocytes along the TCR- α/β pathway in vitro is accompanied by the site-specific deletion of the TCR- γ locus. International Immunology, 1991, 3, 1301-1305.	1.8	19
99	A histidine in the β -CASP domain of Artemis is critical for its full in vitro and in vivo functions. DNA Repair, 2009, 8, 202-208.	1.3	19
100	Expanding the SRI domain family: A common scaffold for binding the phosphorylated C-terminal domain of RNA polymerase II. FEBS Letters, 2014, 588, 4431-4437.	1.3	19
101	Extended clinical and genetic spectrum associated with biallelic RTEL1 mutations. Blood Advances, 2016, 1, 36-46.	2.5	19
102	Impaired Replication Stress Response in Cells from Immunodeficiency Patients Carrying Cernunnos/XLF Mutations. PLoS ONE, 2009, 4, e4516.	1.1	19
103	A short peptide at the C terminus is responsible for the nuclear localization of RAG2. European Journal of Immunology, 2002, 32, 2068.	1.6	18
104	Late-onset combined immune deficiency due to $\langle scp \rangle$ LIGIV $\langle /scp \rangle$ mutations in a 12-year-old patient. Pediatric Allergy and Immunology, 2017, 28, 203-206.	1.1	18
105	Biosafety Studies of a Clinically Applicable Lentiviral Vector for the Gene Therapy of Artemis-SCID. Molecular Therapy - Methods and Clinical Development, 2019, 15, 232-245.	1.8	18
106	Somatic reversion of pathogenic DOCK8 variants alters lymphocyte differentiation and function to effectively cure DOCK8 deficiency. Journal of Clinical Investigation, 2021, 131, .	3.9	18
107	A common $\hat{V}2\hat{D}2\hat{D}13$ T cell receptor gene rearrangement in precursor B acute lymphoblastic leukaemia. British Journal of Haematology, 1991, 79, 44-49.	1.2	17
108	Diagnosis of 22q11.2 Deletion Syndrome and Artemis Deficiency in Two Children with T-B-NK+ Immunodeficiency. Journal of Clinical Immunology, 2012, 32, 1141-1144.	2.0	17

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109	V(D)J recombination and DNA repair: lessons from human immune deficiencies and other animal models. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2002, 2, 473-479.	1.1	16
110	TEA regulates local TCR- β accessibility through histone acetylation. <i>European Journal of Immunology</i> , 2003, 33, 2216-2222.	1.6	16
111	The C-Terminal Domain of Cernunnos/XLF Is Dispensable for DNA Repair In Vivo. <i>Molecular and Cellular Biology</i> , 2009, 29, 1116-1122.	1.1	16
112	When natural mutants do not fit our expectations: the intriguing case of patients with $\langle i \rangle \langle scp \rangle XRCC \langle /scp \rangle 4 \langle /i \rangle$ mutations revealed by whole-exome sequencing. <i>EMBO Molecular Medicine</i> , 2015, 7, 862-864.	3.3	16
113	Mutations in XLF/NHEJ1/Cernunnos gene results in downregulation of telomerase genes expression and telomere shortening. <i>Human Molecular Genetics</i> , 2017, 26, 1900-1914.	1.4	16
114	Cernunnos/Xlf Deficiency Results in Suboptimal V(D)J Recombination and Impaired Lymphoid Development in Mice. <i>Frontiers in Immunology</i> , 2019, 10, 443.	2.2	16
115	The Expanding Spectrum of Human coronin 1A deficiency. <i>Current Allergy and Asthma Reports</i> , 2014, 14, 481.	2.4	15
116	Variable correction of Artemis deficiency by I-Sce1-meganuclease-assisted homologous recombination in murine hematopoietic stem cells. <i>Gene Therapy</i> , 2014, 21, 529-532.	2.3	14
117	Mutations of the RTEL1 Helicase in a Hoyeraal-Hreidarsson Syndrome Patient Highlight the Importance of the ARCH Domain. <i>Human Mutation</i> , 2016, 37, 469-472.	1.1	14
118	Inherited human Apollo deficiency causes severe bone marrow failure and developmental defects. <i>Blood</i> , 2022, 139, 2427-2440.	0.6	14
119	A case report of a patient with microcephaly, facial dysmorphism, chromosomal radiosensitivity and telomere length alterations closely resembling "Nijmegen breakage syndrome" phenotype. <i>European Journal of Medical Genetics</i> , 2007, 50, 176-187.	0.7	13
120	Distinct effects of DNA-PKcs and Artemis inactivation on signal joint formation in vivo. <i>Molecular Immunology</i> , 2008, 45, 3383-3391.	1.0	12
121	Tetratricopeptide repeat domain 7A is a nuclear factor that modulates transcription and chromatin structure. <i>Cell Discovery</i> , 2018, 4, 61.	3.1	10
122	An in vivo study of the impact of deficiency in the DNA repair proteins PAXX and XLF on development and maturation of the hemolymphoid system. <i>Journal of Biological Chemistry</i> , 2020, 295, 2398-2406.	1.6	8
123	CD8 Memory Cells Develop Unique DNA Repair Mechanisms Favoring Productive Division. <i>PLoS ONE</i> , 2015, 10, e0140849.	1.1	8
124	Severe combined immunodeficiency caused by a new homozygous RAG1 mutation with progressive encephalopathy. <i>Hematology/ Oncology and Stem Cell Therapy</i> , 2014, 7, 44-49.	0.6	7
125	$\langle scp \rangle DNA \langle /scp \rangle$ ligase $\langle scp \rangle IV \langle /scp \rangle$ deficiency: Immunoglobulin class deficiency depends on the genotype. <i>Pediatric Allergy and Immunology</i> , 2017, 28, 298-303.	1.1	7
126	Severe combined immune deficiency. , 2020, , 153-205.		7

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127	Indispensable epigenetic control of thymic epithelial cell development and function by polycomb repressive complex 2. <i>Nature Communications</i> , 2021, 12, 3933.	5.8	7
128	A Disease-Causing Single Amino Acid Deletion in the Coiled-Coil Domain of RAD50 Impairs MRE11 Complex Functions in Yeast and Humans. <i>Cell Reports</i> , 2020, 33, 108559.	2.9	7
129	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 408-411.e8.	1.5	6
130	Higher chromosome stability in embryonic neural stem and progenitor cells than in fibroblasts in response to acute or chronic genotoxic stress. <i>DNA Repair</i> , 2020, 88, 102801.	1.3	6
131	Clinical effects of mutations to CD95 (Fas): relevance to autoimmunity?. <i>Seminars in Immunopathology</i> , 1998, 19, 301-310.	4.0	5
132	Lymphopoiesis in transgenic mice over-expressing Artemis. <i>Gene Therapy</i> , 2016, 23, 176-186.	2.3	5
133	An XRCC4 mutant mouse, a model for human X4 syndrome, reveals interplays with Xlf, PAXX, and ATM in lymphoid development. <i>ELife</i> , 2021, 10, .	2.8	4
134	Primary membrane T cell immunodeficiencies. <i>Clinical Immunology and Immunopathology</i> , 1991, 61, S56-S60.	2.1	3
135	A novel radiosensitive SCID patient with a pronounced G2/M sensitivity. <i>DNA Repair</i> , 2010, 9, 365-373.	1.3	3
136	Passera ou ne passera pas€”accessibility is key. <i>Nature Immunology</i> , 2006, 7, 1019-1021.	7.0	2
137	Reduced recruitment of 53BP1 during interstrand crosslink repair is associated with genetically inherited attenuation of mitomycin C sensitivity in a family with Fanconi anemia. <i>Oncotarget</i> , 2018, 9, 3779-3793.	0.8	2
138	Severe Combined Immunodeficiencies. , 2014, , 87-141.		1
139	Severe Combined Immune Deficiency with Absence of B and T Lymphocytes (T ⁺ B ⁻ NK ⁺ SCIDs): The Key Function of V(D)J Recombination for Lymphocyte Development. , 2016, , 369-377.		0