Patrick Revy

List of Publications by Year
in descending order

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76	8,357	76294	64755
papers	citations	40 h-index	79 g-index

82	82	82	9031
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Activation-Induced Cytidine Deaminase (AID) Deficiency Causes the Autosomal Recessive Form of the Hyper-IgM Syndrome (HIGM2). Cell, 2000, 102, 565-575.	13.5	1,489
2	Interferon-Î³ â€“Receptor Deficiency in an Infant with Fatal Bacille Calmetteâ€“GuÃ©rin Infection. New England Journal of Medicine, 1996, 335, 1956-1962.	13.9	832
3	XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. Nature, 2006, 444, 110-114.	13.7	649
4	Cernunnos, a Novel Nonhomologous End-Joining Factor, Is Mutated in Human Immunodeficiency with Microcephaly. Cell, 2006, 124, 287-299.	13.5	640
5	Human uracilâ€“DNA glycosylase deficiency associated with profoundly impaired immunoglobulin class-switch recombination. Nature Immunology, 2003, 4, 1023-1028.	7.0	573
6	Functional antigen-independent synapses formed between T cells and dendritic cells. Nature Immunology, 2001, 2, 925-931.	7.0	268
7	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
8	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. European Respiratory Journal, 2017, 49, 1602314.	3.1	154
9	Human RTEL1 deficiency causes Hoyeraalâ€“Hreidarsson syndrome with short telomeres and genome instability. Human Molecular Genetics, 2013, 22, 3239-3249.	1.4	150
10	Retinoids Regulate Survival and Antigen Presentation by Immature Dendritic Cells. Journal of Experimental Medicine, 2003, 198, 623-634.	4.2	143
11	Prevalence and characteristics ofTERTandTERCmutations in suspected genetic pulmonary fibrosis. European Respiratory Journal, 2016, 48, 1721-1731.	3.1	136
12	HeterozygousRTEL1mutations are associated with familial pulmonary fibrosis. European Respiratory Journal, 2015, 46, 474-485.	3.1	135
13	Whole-exome sequencing identifies Coronin-1A deficiency in 3 siblings with immunodeficiency and EBV-associated B-cell lymphoproliferation. Journal of Allergy and Clinical Immunology, 2013, 131, 1594-1603.e9.	1.5	127
14	Structural characterization of filaments formed by human Xrcc4â€“Cernunnos/XLF complex involved in nonhomologous DNA end-joining. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 12663-12668.	3.3	126
15	A homozygous mucosa-associated lymphoid tissue 1 (MALT1) mutation in a family with combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2013, 132, 151-158.	1.5	124
16	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
17	Hyper-IgM syndrome type 4 with a B lymphocyteâ€“intrinsic selective deficiency in Ig class-switch recombination. Journal of Clinical Investigation, 2003, 112, 136-142.	3.9	114
18	Cernunnos Interacts with the XRCC4Â·DNA-ligase IV Complex and Is Homologous to the Yeast Nonhomologous End-joining Factor Nej1*. Journal of Biological Chemistry, 2006, 281, 13857-13860.	1.6	112

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19	Analysis of class switch recombination and somatic hypermutation in patients affected with autosomal dominant hyper-IgM syndrome type 2. Clinical Immunology, 2005, 115, 277-285.	1.4	111
20	Severe hematologic complications after lung transplantation in patients with telomerase complex mutations. Journal of Heart and Lung Transplantation, 2015, 34, 538-546.	0.3	109
21	Unraveling the pathogenesis of Hoyeraalâ€“Hreidarsson syndrome, a complex telomere biology disorder. British Journal of Haematology, 2015, 170, 457-471.	1.2	105
22	The Block in Immunoglobulin Class Switch Recombination Caused by Activation-Induced Cytidine Deaminase Deficiency Occurs Prior to the Generation of DNA Double Strand Breaks in Switch Î¼ Region. Journal of Immunology, 2003, 171, 2504-2509.	0.4	84
23	Polymerase Îµ1 mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature (â€œFILS syndromeâ€). Journal of Experimental Medicine, 2012, 209, 2323-2330.	4.2	83
24	Hyper-immunoglobulin M syndromes caused by intrinsic B-lymphocyte defects. Immunological Reviews, 2005, 203, 67-79.	2.8	76
25	Function of Apollo (SNM1B) at telomere highlighted by a splice variant identified in a patient with Hoyeraalâ€“Hreidarsson syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 10097-10102.	3.3	76
26	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	4.2	75
27	Somatic genetic rescue in Mendelian haematopoietic diseases. Nature Reviews Genetics, 2019, 20, 582-598.	7.7	74
28	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967.	5.8	66
29	Cernunnos Deficiency Reduces Thymocyte Life Span and Alters the T Cell Repertoire in Mice and Humans. Molecular and Cellular Biology, 2013, 33, 701-711.	1.1	63
30	An inÂvivo genetic reversion highlights the crucial role of Myb-Like, SWIRM, and MPN domains 1 (MYSM1) in human hematopoiesis and lymphocyte differentiation. Journal of Allergy and Clinical Immunology, 2015, 136, 1619-1626.e5.	1.5	63
31	The Repair of DNA Damages/Modifications During the Maturation of the Immune System: Lessons from Human Primary Immunodeficiency Disorders and Animal Models. Advances in Immunology, 2005, 87, 237-295.	1.1	58
32	RAG2 and XLF/Cernunnos interplay reveals a novel role for the RAG complex in DNA repair. Nature Communications, 2016, 7, 10529.	5.8	57
33	Tâ€„cell adhesion lowers the threshold for antigen detection. European Journal of Immunology, 2003, 33, 1215-1223.	1.6	48
34	EFL1 mutations impair eIF6 release to cause Shwachman-Diamond syndrome. Blood, 2019, 134, 277-290.	0.6	48
35	A primary immunodeficiency characterized by defective immunoglobulin class switch recombination and impaired DNA repair. Journal of Experimental Medicine, 2007, 204, 1207-1216.	4.2	47
36	Interplay between Cernunnos-XLF and Nonhomologous End-joining Proteins at DNA Ends in the Cell. Journal of Biological Chemistry, 2007, 282, 31937-31943.	1.6	47

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37	Regulator of telomere length 1 (RTEL1) mutations are associated with heterogeneous pulmonary and extra-pulmonary phenotypes. European Respiratory Journal, 2019, 53, 1800508.	3.1	45
38	Delineation of the Xrcc4-interacting Region in the Globular Head Domain of Cernunnos/XLF. Journal of Biological Chemistry, 2010, 285, 26475-26483.	1.6	44
39	Cernunnos influences human immunoglobulin class switch recombination and may be associated with B cell lymphomagenesis. Journal of Experimental Medicine, 2012, 209, 291-305.	4.2	44
40	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	5.8	44
41	Requirement for XLF/Cernunnos in alignment-based gap filling by DNA polymerases Î» and Î¼ for nonhomologous end joining in human whole-cell extracts. Nucleic Acids Research, 2009, 37, 4055-4062.	6.5	43
42	Primary Microcephaly, Impaired DNA Replication, and Genomic Instability Caused by Compound HeterozygousATRMutations. Human Mutation, 2013, 34, 374-384.	1.1	43
43	PROMIDISÎ±: AÂT-cell receptor Î± signature associated with immunodeficiencies caused by V(D)J recombination defects. Journal of Allergy and Clinical Immunology, 2019, 143, 325-334.e2.	1.5	43
44	PAXX and Xlf interplay revealed by impaired CNS development and immunodeficiency of double KO mice. Cell Death and Differentiation, 2018, 25, 444-452.	5.0	40
45	NHP2 deficiency impairs rRNA biogenesis and causes pulmonary fibrosis and HÃ¸yeraalâ€“Hreidarsson syndrome. Human Molecular Genetics, 2020, 29, 907-922.	1.4	38
46	Imaging T-cell antigen recognition and comparing immunological and neuronal synapses. Immunology, 2001, 103, 417-425.	2.0	37
47	Human Models of Inherited Immunoglobulin Class Switch Recombination and Somatic Hypermutation Defects (Hyper-IgM Syndromes). Advances in Immunology, 2004, 82, 295-330.	1.1	37
48	Unraveling Ewing Sarcoma Tumorigenesis Originating from Patient-Derived Mesenchymal Stem Cells. Cancer Research, 2021, 81, 4994-5006.	0.4	35
49	Heterogeneous telomere defects in patients with severe forms of dyskeratosis congenita. Journal of Allergy and Clinical Immunology, 2012, 129, 473-482.e3.	1.5	34
50	A nonsense mutation in the DNA repair factor Hebo causes mild bone marrow failure and microcephaly. Journal of Experimental Medicine, 2016, 213, 1011-1028.	4.2	34
51	The Câ€terminal extension of human RTEL1, mutated in HoyeraalâHreidarsson syndrome, contains Harmoninâ€Nâ€like domains. Proteins: Structure, Function and Bioinformatics, 2014, 82, 897-903.	1.5	31
52	Impaired telomere integrity and rRNA biogenesis in PARNâ€deficient patients and knockâ€out models. EMBO Molecular Medicine, 2019, 11, e10201.	3.3	31
53	Gain-of-function mutations in RPA1 cause a syndrome with short telomeres and somatic genetic rescue. Blood, 2022, 139, 1039-1051.	0.6	29
54	DNA repair and the immune system: From V(D)J recombination to aging lymphocytes. European Journal of Immunology, 2007, 37, S71-S82.	1.6	27

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55	Human regulator of telomere elongation helicase 1 (RTEL1) is required for the nuclear and cytoplasmic trafficking of pre-U2 RNA. Nucleic Acids Research, 2015, 43, 1834-1847.	6.5	26
56	Normal CD40-mediated activation of monocytes and dendritic cells from patients with hyper-IgM syndrome due to a CD40 pathway defect in B cells. European Journal of Immunology, 1998, 28, 3648-3654.	1.6	25
57	A Syndrome Involving Intrauterine Growth Retardation, Microcephaly, Cerebellar Hypoplasia, B Lymphocyte Deficiency, and Progressive Pancytopenia. Pediatrics, 2000, 105, e39-e39.	1.0	25
58	Cernunnos-XLF, a recently identified non-homologous end-joining factor required for the development of the immune system. Current Opinion in Allergy and Clinical Immunology, 2006, 6, 416-420.	1.1	24
59	Two SCID cases with Cernunnosâ€XLF deficiency successfully treated by hematopoietic stem cell transplantation. Pediatric Transplantation, 2012, 16, E167-71.	0.5	22
60	Full length RTEL1 is required for the elongation of the single-stranded telomeric overhang by telomerase. Nucleic Acids Research, 2020, 48, 7239-7251.	6.5	20
61	Expanding the SRI domain family: A common scaffold for binding the phosphorylated Câ€terminal domain of RNA polymerase II. FEBS Letters, 2014, 588, 4431-4437.	1.3	19
62	Extended clinical and genetic spectrum associated with biallelic RTEL1 mutations. Blood Advances, 2016, 1, 36-46.	2.5	19
63	Replication stress triggered by nucleotide pool imbalance drives DNA damage and cGAS-STING pathway activation in NAFLD. Developmental Cell, 2022, 57, 1728-1741.e6.	3.1	17
64	The C-Terminal Domain of Cernunnos/XLF Is Dispensable for DNA Repair In Vivo. Molecular and Cellular Biology, 2009, 29, 1116-1122.	1.1	16
65	Mutations in XLF/NHEJ1/Cernunnos gene results in downregulation of telomerase genes expression and telomere shortening. Human Molecular Genetics, 2017, 26, 1900-1914.	1.4	16
66	Hyper-immunoglobulin-M syndromes caused by an intrinsic B cell defect. Current Opinion in Allergy and Clinical Immunology, 2003, 3, 421-425.	1.1	14
67	Pulmonary fibrosis associated with TINF2 gene mutation: is somatic reversion required?. European Respiratory Journal, 2014, 44, 269-270.	3.1	14
68	Mutations of the RTEL1 Helicase in a Hoyeraal-Hreidarsson Syndrome Patient Highlight the Importance of the ARCH Domain. Human Mutation, 2016, 37, 469-472.	1.1	14
69	Inherited human Apollo deficiency causes severe bone marrow failure and developmental defects. Blood, 2022, 139, 2427-2440.	0.6	14
70	First heterozygous NOP10 mutation in familial pulmonary fibrosis. European Respiratory Journal, 2020, 55, 1902465.	3.1	13
71	Myelodysplastic syndromes and idiopathic pulmonary fibrosis: a dangerous liaison. Respiratory Research, 2019, 20, 182.	1.4	7
72	A Disease-Causing Single Amino Acid Deletion in the Coiled-Coil Domain of RAD50 Impairs MRE11 Complex Functions in Yeast and Humans. Cell Reports, 2020, 33, 108559.	2.9	7

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73	Genotype-Phenotype Relationships in Inheritable Idiopathic Pulmonary Fibrosis: A Greek National Cohort Study. Respiration, 2022, 101, 531-543.	1.2	5
74	Reduced recruitment of 53BP1 during interstrand crosslink repair is associated with genetically inherited attenuation of mitomycin C sensitivity in a family with Fanconi anemia. Oncotarget, 2018, 9, 3779-3793.	0.8	2
75	The Immunologic Complications and Genetic Origins of Telomere Disorders. , 2016, , 451-457.		0
76	EFL1 deficiency: a little is better than nothing. Blood, 2021, 138, 2016-2018.	0.6	0