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	Gain-of-function mutations in RPA1 cause a syndrome with short telomeres and somatic genetic rescue. Blood, 2022, 139, 1039-1051.	0.6	29
2	Genotype-Phenotype Relationships in Inheritable Idiopathic Pulmonary Fibrosis: A Greek National Cohort Study. Respiration, 2022, 101, 531-543.	1.2	5
3	Inherited human Apollo deficiency causes severe bone marrow failure and developmental defects. Blood, 2022, 139, 2427-2440.	0.6	14
4	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
5	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	5.8	44
6	Unraveling Ewing Sarcoma Tumorigenesis Originating from Patient-Derived Mesenchymal Stem Cells. Cancer Research, 2021, 81, 4994-5006.	0.4	35
7	EFL1 deficiency: a little is better than nothing. Blood, 2021, 138, 2016-2018.	0.6	0
8	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
9	NHP2 deficiency impairs rRNA biogenesis and causes pulmonary fibrosis and HÃ¸yeraalâ€“Hreidarsson syndrome. Human Molecular Genetics, 2020, 29, 907-922.	1.4	38
10	First heterozygous NOP10 mutation in familial pulmonary fibrosis. European Respiratory Journal, 2020, 55, 1902465.	3.1	13
11	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
12	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
13	Myelodysplastic syndromes and idiopathic pulmonary fibrosis: a dangerous liaison. Respiratory Research, 2019, 20, 182.	1.4	7
14	Impaired telomere integrity and rRNA biogenesis in PARNâ€deficient patients and knockâ€out models. EMBO Molecular Medicine, 2019, 11, e10201.	3.3	31
15	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967.	5.8	66
16	EFL1 mutations impair eIF6 release to cause Shwachman-Diamond syndrome. Blood, 2019, 134, 277-290.	0.6	48
17	Somatic genetic rescue in Mendelian haematopoietic diseases. Nature Reviews Genetics, 2019, 20, 582-598.	7.7	74
18	Bloom syndrome protein restrains innate immune sensing of micronuclei by cGAS. Journal of Experimental Medicine, 2019, 216, 1199-1213.	4.2	75

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19	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
20	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
21	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
22	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. European Respiratory Journal, 2017, 49, 1602314.	3.1	154
23	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
24	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
25	Extended clinical and genetic spectrum associated with biallelic RTEL1 mutations. Blood Advances, 2016, 1, 36-46.	2.5	19
26	The Immunologic Complications and Genetic Origins of Telomere Disorders. , 2016, , 451-457.		0
27	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
28	Prevalence and characteristics ofTERTandTERCmutations in suspected genetic pulmonary fibrosis. European Respiratory Journal, 2016, 48, 1721-1731.	3.1	136
29	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
30	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
31	RAG2 and XLF/Cernunnos interplay reveals a novel role for the RAG complex in DNA repair. Nature Communications, 2016, 7, 10529.	5.8	57
32	Unraveling the pathogenesis of Hoyeraalâ€“Hreidarsson syndrome, a complex telomere biology disorder. British Journal of Haematology, 2015, 170, 457-471.	1.2	105
33	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
34	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
35	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
36	HeterozygousRTEL1mutations are associated with familial pulmonary fibrosis. European Respiratory Journal, 2015, 46, 474-485.	3.1	135

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37	The Câ€terminal extension of human RTEL1, mutated in Hoyeraalâ€Hreidarsson syndrome, contains Harmoninâ€Nâ€like domains. Proteins: Structure, Function and Bioinformatics, 2014, 82, 897-903.	1.5	31
38	Pulmonary fibrosis associated with TINF2 gene mutation: is somatic reversion required?. European Respiratory Journal, 2014, 44, 269-270.	3.1	14
39	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
40	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
41	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
42	Primary Microcephaly, Impaired DNA Replication, and Genomic Instability Caused by Compound HeterozygousATRMutations. Human Mutation, 2013, 34, 374-384.	1.1	43
43	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
44	Human RTEL1 deficiency causes Hoyeraalâ€“Hreidarsson syndrome with short telomeres and genome instability. Human Molecular Genetics, 2013, 22, 3239-3249.	1.4	150
45	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
46	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
47	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
48	Two SCID cases with Cernunnosâ€XLF deficiency successfully treated by hematopoietic stem cell transplantation. Pediatric Transplantation, 2012, 16, E167-71.	0.5	22
49	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
50	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
51	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
52	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
53	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
54	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

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55	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
56	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
57	Cernunnos, a Novel Nonhomologous End-Joining Factor, Is Mutated in Human Immunodeficiency with Microcephaly. Cell, 2006, 124, 287-299.	13.5	640
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	XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. Nature, 2006, 444, 110-114.	13.7	649
60	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
61	Hyper-immunoglobulin M syndromes caused by intrinsic B-lymphocyte defects. Immunological Reviews, 2005, 203, 67-79.	2.8	76
62	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
63	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
64	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
65	Tâ€„cell adhesion lowers the threshold for antigen detection. European Journal of Immunology, 2003, 33, 1215-1223.	1.6	48
66	Human uracilâ€“DNA glycosylase deficiency associated with profoundly impaired immunoglobulin class-switch recombination. Nature Immunology, 2003, 4, 1023-1028.	7.0	573
67	Retinoids Regulate Survival and Antigen Presentation by Immature Dendritic Cells. Journal of Experimental Medicine, 2003, 198, 623-634.	4.2	143
68	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
69	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
70	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
71	Imaging T-cell antigen recognition and comparing immunological and neuronal synapses. Immunology, 2001, 103, 417-425.	2.0	37
72	Functional antigen-independent synapses formed between T cells and dendritic cells. Nature Immunology, 2001, 2, 925-931.	7.0	268

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73	A Syndrome Involving Intrauterine Growth Retardation, Microcephaly, Cerebellar Hypoplasia, B Lymphocyte Deficiency, and Progressive Pancytopenia. Pediatrics, 2000, 105, e39-e39.	1.0	25
74	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
75	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
76	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