

Genevieve de Saint Basile

List of Publications by Year in descending order

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52
papers

5,707
citations

201674

27
h-index

243625

44
g-index

53
all docs

53
docs citations

53
times ranked

5409
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in RAB27A cause Griscelli syndrome associated with haemophagocytic syndrome. <i>Nature Genetics</i> , 2000, 25, 173-176.	21.4	831
2	Munc13-4 Is Essential for Cytolytic Granules Fusion and Is Mutated in a Form of Familial Hemophagocytic Lymphohistiocytosis (FHL3). <i>Cell</i> , 2003, 115, 461-473.	28.9	825
3	XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. <i>Nature</i> , 2006, 444, 110-114.	27.8	649
4	Molecular mechanisms of biogenesis and exocytosis of cytotoxic granules. <i>Nature Reviews Immunology</i> , 2010, 10, 568-579.	22.7	354
5	Munc18-2 deficiency causes familial hemophagocytic lymphohistiocytosis type 5 and impairs cytotoxic granule exocytosis in patient NK cells. <i>Journal of Clinical Investigation</i> , 2009, 119, 3765-3773.	8.2	301
6	Atypical X-Linked Severe Combined Immunodeficiency Due to Possible Spontaneous Reversion of the Genetic Defect in T Cells. <i>New England Journal of Medicine</i> , 1996, 335, 1563-1567.	27.0	259
7	Hematopoietic Stem Cell Transplantation in Hemophagocytic Lymphohistiocytosis: A Single-Center Report of 48 Patients. <i>Pediatrics</i> , 2006, 117, e743-e750.	2.1	230
8	Immunotherapy of Familial Hemophagocytic Lymphohistiocytosis With Antithymocyte Globulins: A Single-Center Retrospective Report of 38 Patients. <i>Pediatrics</i> , 2007, 120, e622-e628.	2.1	218
9	Neutralization of IFN γ defeats haemophagocytosis in LCMV-infected perforin and Rab27a-deficient mice. <i>EMBO Molecular Medicine</i> , 2009, 1, 112-124.	6.9	165
10	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. <i>Nature Genetics</i> , 2018, 50, 1650-1657.	21.4	151
11	Inherited defects in lymphocyte cytotoxic activity. <i>Immunological Reviews</i> , 2010, 235, 10-23.	6.0	143
12	Linkage of Familial Hemophagocytic Lymphohistiocytosis to 10q21-22 and Evidence for Heterogeneity. <i>American Journal of Human Genetics</i> , 1999, 64, 172-179.	6.2	133
13	Functional consequences of perforin gene mutations in 22 patients with familial haemophagocytic lymphohistiocytosis. <i>British Journal of Haematology</i> , 2002, 117, 965-972.	2.5	128
14	Perforin: more than just an effector molecule. <i>Trends in Immunology</i> , 2000, 21, 254-256.	7.5	109
15	Griscelli syndrome restricted to hypopigmentation results from a melanophilin defect (GS3) or a MYO5A F-exon deletion (GS1). <i>Journal of Clinical Investigation</i> , 2003, 112, 450-456.	8.2	108
16	Two Genes Are Responsible for Griscelli Syndrome at the Same 15q21 Locus. <i>Genomics</i> , 2000, 63, 299-306.	2.9	106
17	Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. <i>Human Mutation</i> , 2001, 18, 255-263.	2.5	100
18	Hemophagocytic syndrome: primary forms and predisposing conditions. <i>Current Opinion in Immunology</i> , 2017, 49, 20-26.	5.5	97

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19	The role of cytotoxicity in lymphocyte homeostasis. <i>Current Opinion in Immunology</i> , 2001, 13, 549-554.	5.5	79
20	IL2RGbase: a database of β -chain defects causing human X-SCID. <i>Trends in Immunology</i> , 1996, 17, 507-511.	7.5	76
21	Interleukin-2 (IL-2) receptor β chain mutations in X-linked severe combined immunodeficiency disease result in the loss of high-affinity IL-2 receptor binding. <i>European Journal of Immunology</i> , 1994, 24, 475-479.	2.9	67
22	Immune deficiency-related enteropathy-lymphocytopenia-alopecia syndrome results from tetratricopeptide repeat domain 7A deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 1354-1364.e6.	2.9	66
23	Chediak-Higashi syndrome associated with maternal uniparental isodisomy of chromosome 1. <i>European Journal of Human Genetics</i> , 1999, 7, 633-637.	2.8	64
24	<scp>LYST</scp> Controls the Biogenesis of the Endosomal Compartment Required for Secretory Lysosome Function. <i>Traffic</i> , 2015, 16, 191-203.	2.7	63
25	An allelic variant of Griscelli disease: presentation with severe hypotonia, mental-motor retardation, and hypopigmentation consistent with Elejalde syndrome (neuroectodermal melanolyosomal) Tj ETQq1 1 0.7843146rgBT /Overlock 10	1.4	29
26	Isolated X-linked thrombocytopenia in two unrelated families is associated with point mutations in the Wiskott-Aldrich syndrome protein gene. <i>Journal of Pediatrics</i> , 1996, 129, 56-62.	1.8	35
27	Gene Therapy for Human Severe Combined Immunodeficiencies. <i>Immunity</i> , 2001, 15, 1-4.	14.3	35
28	HAVCR2 mutations are associated with severe hemophagocytic syndrome in subcutaneous panniculitis-like T-cell lymphoma. <i>Blood</i> , 2020, 135, 1058-1061.	1.4	29
29	Improved oligonucleotide primer set for molecular diagnosis of X-linked agammaglobulinaemia: predominance of amino acid substitutions in the catalytic domain of Bruton's tyrosine kinase. <i>Human Molecular Genetics</i> , 1995, 4, 2403-2405.	2.9	28
30	Eleven novel JAK3 mutations in patients with severe combined immunodeficiency?including the first patients with mutations in the kinase domain. <i>Human Mutation</i> , 2001, 18, 355-356.	2.5	28
31	Inherited defects causing hemophagocytic lymphohistiocytic syndrome. <i>Annals of the New York Academy of Sciences</i> , 2011, 1246, 64-76.	3.8	28
32	Efficacy of ruxolitinib in subcutaneous panniculitis-like T-cell lymphoma and hemophagocytic lymphohistiocytosis. <i>Blood Advances</i> , 2020, 4, 1383-1387.	5.2	21
33	Fine mapping of the human SCIDX1 locus at Xq12-13.1. <i>Human Molecular Genetics</i> , 1993, 2, 651-654.	2.9	20
34	The murine interleukin-2 receptor β chain gene: Organization, chromosomal localization and expression in the adult thymus. <i>European Journal of Immunology</i> , 1994, 24, 3014-3018.	2.9	19
35	Gene therapy of severe combined immunodeficiencies. <i>Immunological Reviews</i> , 2000, 178, 13-20.	6.0	18
36	Gene Therapy of X-Linked Severe Combined Immunodeficiency. , 2003, 215, 247-260.		18

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37	Ichthyosis as the dermatological phenotype associated with <i>TTC7A</i> mutations. <i>British Journal of Dermatology</i> , 2016, 175, 1061-1064.	1.5	17
38	X-linked immunodeficiencies: clues to genes involved in T- and B-cell differentiation. <i>Trends in Immunology</i> , 1991, 12, 456-461.	7.5	12
39	A <i>RAB27A</i> duplication in several cases of Griscelli syndrome type 2: An explanation for cases lacking a genetic diagnosis. <i>Human Mutation</i> , 2017, 38, 1355-1359.	2.5	9
40	Chédiak-Higashi and Griscelli syndromes. <i>Immunology and Allergy Clinics of North America</i> , 2002, 22, 301-317.	1.9	5
41	Gene therapy of severe combined immunodeficiencies. <i>Advances in Experimental Medicine and Biology</i> , 2001, 495, 199-204.	1.6	5
42	Small Molecule Inhibitors of Interferon-Induced JAK-STAT Signalling. <i>Angewandte Chemie - International Edition</i> , 0, , .	13.8	5
43	Localization of the Rab Escort Protein-2 (REP2) and Inositol 1,4,5-Trisphosphate 3-Kinase (ITPKB) Genes to Mouse Chromosome 1 by in Situ Hybridization and Precision of the Syntenic Regions between Mouse and Human 1q42-q44. <i>Genomics</i> , 1997, 43, 111-113.	2.9	4
44	Genetic Diseases Predisposing to HLH. , 2014, , 437-460.		4
45	Primary membrane T cell immunodeficiencies. <i>Clinical Immunology and Immunopathology</i> , 1991, 61, S56-S60.	2.0	3
46	Response to Moretta et al. and Arnaout. <i>Trends in Immunology</i> , 2000, 21, 593-594.	7.5	1
47	Molecular causes leading to defective melanosome transport in Griscelli syndrome. <i>Experimental Dermatology</i> , 2006, 15, 848-849.	2.9	0
48	Familial Hemophagocytic Lymphohistiocytosis Type 5 in a Patient With Microvillous Inclusion Disease (MVID). <i>American Journal of Clinical Pathology</i> , 2012, 138, A289-A289.	0.7	0
49	Genetic diseases predisposing to HLH. , 2020, , 549-572.		0
50	The Discovery of the Familial Hemophagocytosis Syndromes. , 2014, , 139-149.		0
51	Familial Lymphohistiocytosis. , 2016, , 400-406.		0
52	Small Molecule Inhibitors of Interferon-Induced JAK-STAT Signalling. <i>Angewandte Chemie</i> , 0, , .	2.0	0