Genevieve de Saint Basile

List of Publications by Year
in descending order

52 papers	5,707 citations	230014 27 h-index	274796 44 g-index
53 all docs	53 docs citations	53 times ranked	5813 citing authors

#	Article	IF	Citations
1	Genetic diseases predisposing to HLH. , 2020, , 549-572.		0
2	HAVCR2 mutations are associated with severe hemophagocytic syndrome in subcutaneous panniculitis-like T-cell lymphoma. Blood, 2020, 135, 1058-1061.	0.6	29
3	Efficacy of ruxolitinib in subcutaneous panniculitis-like T-cell lymphoma and hemophagocytic lymphohistiocytosis. Blood Advances, 2020, 4, 1383-1387.	2.5	21
4	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. Nature Genetics, 2018, 50, 1650-1657.	9.4	151
5	A RAB27A duplication in several cases of Griscelli syndrome type 2: An explanation for cases lacking a genetic diagnosis. Human Mutation, 2017, 38, 1355-1359.	1.1	9
6	Hemophagocytic syndrome: primary forms and predisposing conditions. Current Opinion in Immunology, 2017, 49, 20-26.	2.4	97
7	Ichthyosis as the dermatological phenotype associated withTTC7Amutations. British Journal of Dermatology, 2016, 175, 1061-1064.	1.4	17
8	Familial Lymphohistiocytosis. , 2016, , 400-406.		0
9	<scp>LYST</scp> Controls the Biogenesis of the Endosomal Compartment Required for Secretory Lysosome Function. Traffic, 2015, 16, 191-203.	1.3	63
10	Genetic Diseases Predisposing to HLH. , 2014, , 437-460.		4
11	Immune deficiencyâ€“related enteropathy-lymphocytopenia-alopecia syndrome results from tetratricopeptide repeat domain 7A deficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 1354-1364.e6.	1.5	66
12	The Discovery of the Familial Hemophagocytosis Syndromes. , 2014, , 139-149.		0
13	Familial Hemophagocytic Lymphohistiocytosis Type 5 in a Patient With Microvillous Inclusion Disease (MVID). American Journal of Clinical Pathology, 2012, 138, A289-A289.	0.4	0
14	Inherited defects causing hemophagocytic lymphohistiocytic syndrome. Annals of the New York Academy of Sciences, 2011, 1246, 64-76.	1.8	28
15	Molecular mechanisms of biogenesis and exocytosis of cytotoxic granules. Nature Reviews Immunology, 2010, 10, 568-579.	10.6	354
16	Inherited defects in lymphocyte cytotoxic activity. Immunological Reviews, 2010, 235, 10-23.	2.8	143
17	Neutralization of IFNÎ³ defeats haemophagocytosis in LCMVâ€infected perforinâ€and Rab27aâ€deficient mice. EMBO Molecular Medicine, 2009, 1, 112-124.	3.3	165
18	Munc18-2 deficiency causes familial hemophagocytic lymphohistiocytosis type 5 and impairs cytotoxic granule exocytosis in patient NK cells. Journal of Clinical Investigation, 2009, 119, 3765-3773.	3.9	301

#	Article	IF	Citations
19	Immunotherapy of Familial Hemophagocytic Lymphohistiocytosis With Antithymocyte Globulins: A Single-Center Retrospective Report of 38 Patients. Pediatrics, 2007, 120, e622-e628.	1.0	218
20	Molecular causes leading to defective melanosome transport in Griscelli syndrome. Experimental Dermatology, 2006, 15, 848-849.	1.4	0
21	XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. Nature, 2006, 444, 110-114.	13.7	649
22	Hematopoietic Stem Cell Transplantation in Hemophagocytic Lymphohistiocytosis: A Single-Center Report of 48 Patients. Pediatrics, 2006, 117, e743-e750.	1.0	230
23	Gene Therapy of X-Linked Severe Combined Immunodeficiency. , 2003, 215, 247-260.		18
24	Munc13-4 Is Essential for Cytolytic Granules Fusion and Is Mutated in a Form of Familial Hemophagocytic Lymphohistiocytosis (FHL3). Cell, 2003, 115, 461-473.	13.5	825
25	Griscelli syndrome restricted to hypopigmentation results from a melanophilin defect (GS3) or a MYO5A F-exon deletion (GS1). Journal of Clinical Investigation, 2003, 112, 450-456.	3.9	108
26	ChÃ©diak-Higashi and Griscelli syndromes. Immunology and Allergy Clinics of North America, 2002, 22, 301-317.	0.7	5
27	Functional consequences of perforin gene mutations in 22 patients with familial haemophagocytic lymphohistiocytosis. British Journal of Haematology, 2002, 117, 965-972.	1.2	128
28	Gene Therapy for Human Severe Combined Immunodeficiencies. Immunity, 2001, 15, 1-4.	6.6	35
29	The role of cytotoxicity in lymphocyte homeostasis. Current Opinion in Immunology, 2001, 13, 549-554.	2.4	79
30	Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. Human Mutation, 2001, 18, 255-263.	1.1	100
31	Eleven novel JAK3 mutations in patients with severe combined immunodeficiency?including the first patients with mutations in the kinase domain. Human Mutation, 2001, 18, 355-356.	1.1	28
32	Gene therapy of severe combined immunodeficiencies. Advances in Experimental Medicine and Biology, 2001, 495, 199-204.	0.8	5
33	Gene therapy of severe combined immunodeficiencies. Immunological Reviews, 2000, 178, 13-20.	2.8	18
34	Perforin: more than just an effector molecule. Trends in Immunology, 2000, 21, 254-256.	7.5	109
35	Response to Moretta et al. and Arnaout. Trends in Immunology, 2000, 21, 593-594.	7.5	1
36	Mutations in RAB27A cause Griscelli syndrome associated with haemophagocytic syndrome. Nature Genetics, 2000, 25, 173-176.	9.4	831

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37	An allelic variant of Griscelli disease: presentation with severe hypotonia, mental-motor retardation, and hypopigmentation consistent with Elejalde syndrome (neuroectodermal melanolysosomal) Tj ETQq1 1 0.784314 rgBT /Overlock 10	1.4	40
38	Two Genes Are Responsible for Griscelli Syndrome at the Same 15q21 Locus. Genomics, 2000, 63, 299-306.	1.3	106
39	Chediak-Higashi syndrome associated with maternal uniparental isodisomy of chromosome 1. European Journal of Human Genetics, 1999, 7, 633-637.	1.4	64
40	Linkage of Familial Hemophagocytic Lymphohistiocytosis to 10q21-22 and Evidence for Heterogeneity. American Journal of Human Genetics, 1999, 64, 172-179.	2.6	133
41	Localization of the Rab Escort Protein-2 (REP2) and Inositol 1,4,5-Trisphosphate 3-Kinase (ITPKB) Genes to Mouse Chromosome 1 byin SituHybridization and Precision of the Syntenic Regions between Mouse and Human 1q42â€“q44. Genomics, 1997, 43, 111-113.	1.3	4
42	Isolated X-linked thrombocytopenia in two unrelated families is associated with point mutations in the Wiskott-Aldrich syndrome protein gene. Journal of Pediatrics, 1996, 129, 56-62.	0.9	35
43	IL2RGbase: a database of Î³c-chain defects causing human X-SCID. Trends in Immunology, 1996, 17, 507-511.	7.5	76
44	Atypical X-Linked Severe Combined Immunodeficiency Due to Possible Spontaneous Reversion of the Genetic Defect in T Cells. New England Journal of Medicine, 1996, 335, 1563-1567.	13.9	259
45	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. Human Molecular Genetics, 1995, 4, 2403-2405.	1.4	28
46	Interleukin-2 (IL-2) receptor Î³ chain mutations in X-linked severe combined immunodeficiency disease result in the loss of high-affinity IL-2 receptor binding. European Journal of Immunology, 1994, 24, 475-479.	1.6	67
47	The murine interleukin-2 receptor Î³ chain gene: Organization, chromosomal localization and expression in the adult thymus. European Journal of Immunology, 1994, 24, 3014-3018.	1.6	19
48	Fine mapping of the human SCIDX1 locus at Xq12â€“13.1. Human Molecular Genetics, 1993, 2, 651-654.	1.4	20
49	Primary membrane T cell immunodeficiencies. Clinical Immunology and Immunopathology, 1991, 61, S56-S60.	2.1	3
50	X-linked immunodeficiencies: clues to genes involved in T- and B-cell differentiation. Trends in Immunology, 1991, 12, 456-461.	7.5	12
51	Small Molecule Inhibitors of Interferonâ€Induced JAKâ€STAT Signalling. Angewandte Chemie, 0, , .	1.6	0
52	Small Molecule Inhibitors of Interferonâ€Induced JAKâ€STAT Signalling. Angewandte Chemie - International Edition, 0, , .	7.2	5