# Genevieve de Saint Basile

List of Publications by Year
in descending order



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|---|---|---|---|
| 52 papers | 5,707 citations | 201658 27 h-index | 243610 44 g-index |
| 53 all docs | 53 docs citations | 53 times ranked | 5409 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. | 1.4 | 29 |
| 3 | Efficacy of ruxolitinib in subcutaneous panniculitis-like T-cell lymphoma and hemophagocytic lymphohistiocytosis. Blood Advances, 2020, 4, 1383-1387. | 5.2 | 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. | 21.4 | 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. | 2.5 | 9 |
| 6 | Hemophagocytic syndrome: primary forms and predisposing conditions. Current Opinion in Immunology, 2017, 49, 20-26. | 5.5 | 97 |
| 7 | Ichthyosis as the dermatological phenotype associated withTTC7Amutations. British Journal of Dermatology, 2016, 175, 1061-1064. | 1.5 | 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. | 2.7 | 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. | 2.9 | 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. | 0.7 | 0 |
| 14 | Inherited defects causing hemophagocytic lymphohistiocytic syndrome. Annals of the New York Academy of Sciences, 2011, 1246, 64-76. | 3.8 | 28 |
| 15 | Molecular mechanisms of biogenesis and exocytosis of cytotoxic granules. Nature Reviews Immunology, 2010, 10, 568-579. | 22.7 | 354 |
| 16 | Inherited defects in lymphocyte cytotoxic activity. Immunological Reviews, 2010, 235, 10-23. | 6.0 | 143 |
| 17 | Neutralization of IFNÎ³ defeats haemophagocytosis in LCMVâ€infected perforinâ€and Rab27aâ€deficient mice. EMBO Molecular Medicine, 2009, 1, 112-124. | 6.9 | 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. | 8.2 | 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. | 2.1 | 218 |
| 20 | Molecular causes leading to defective melanosome transport in Griscelli syndrome. Experimental Dermatology, 2006, 15, 848-849. | 2.9 | 0 |
| 21 | XIAP deficiency in humans causes an X-linked lymphoproliferative syndrome. Nature, 2006, 444, 110-114. | 27.8 | 649 |
| 22 | Hematopoietic Stem Cell Transplantation in Hemophagocytic Lymphohistiocytosis: A Single-Center Report of 48 Patients. Pediatrics, 2006, 117, e743-e750. | 2.1 | 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. | 28.9 | 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. | 8.2 | 108 |
| 26 | ChÃ©diak-Higashi and Griscelli syndromes. Immunology and Allergy Clinics of North America, 2002, 22, 301-317. | 1.9 | 5 |
| 27 | Functional consequences of perforin gene mutations in 22 patients with familial haemophagocytic lymphohistiocytosis. British Journal of Haematology, 2002, 117, 965-972. | 2.5 | 128 |
| 28 | Gene Therapy for Human Severe Combined Immunodeficiencies. Immunity, 2001, 15, 1-4. | 14.3 | 35 |
| 29 | The role of cytotoxicity in lymphocyte homeostasis. Current Opinion in Immunology, 2001, 13, 549-554. | 5.5 | 79 |
| 30 | Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. Human Mutation, 2001, 18, 255-263. | 2.5 | 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. | 2.5 | 28 |
| 32 | Gene therapy of severe combined immunodeficiencies. Advances in Experimental Medicine and Biology, 2001, 495, 199-204. | 1.6 | 5 |
| 33 | Gene therapy of severe combined immunodeficiencies. Immunological Reviews, 2000, 178, 13-20. | 6.0 | 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. | 21.4 | 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 | 3.6 | 40 |
| 38 | Two Genes Are Responsible for Griscelli Syndrome at the Same 15q21 Locus. Genomics, 2000, 63, 299-306. | 2.9 | 106 |
| 39 | Chediak-Higashi syndrome associated with maternal uniparental isodisomy of chromosome 1. European Journal of Human Genetics, 1999, 7, 633-637. | 2.8 | 64 |
| 40 | Linkage of Familial Hemophagocytic Lymphohistiocytosis to 10q21-22 and Evidence for Heterogeneity. American Journal of Human Genetics, 1999, 64, 172-179. | 6.2 | 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. | 2.9 | 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. | 1.8 | 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. | 27.0 | 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. | 2.9 | 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. | 2.9 | 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. | 2.9 | 19 |
| 48 | Fine mapping of the human SCIDX1 locus at Xq12â€“13.1. Human Molecular Genetics, 1993, 2, 651-654. | 2.9 | 20 |
| 49 | Primary membrane T cell immunodeficiencies. Clinical Immunology and Immunopathology, 1991, 61, S56-S60. | 2.0 | 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, , . | 2.0 | 0 |
| 52 | Small Molecule Inhibitors of Interferonâ€Induced JAKâ€STAT Signalling. Angewandte Chemie - International Edition, 0, , . | 13.8 | 5 |