Gulbu Uzel

List of Publications by Year
in descending order

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45 papers	4,585 citations	236925 25 h-index	233421 45 g-index
45 all docs	45 docs citations	45 times ranked	6933 citing authors

#	Article	IF	Citations
1	Immune dysregulation in human subjects with heterozygous germline mutations in CTLA4. Science, 2014, 345, 1623-1627.	12.6	745
2	Combined Immunodeficiency Associated withDOCK8Mutations. New England Journal of Medicine, 2009, 361, 2046-2055.	27.0	655
3	Second messenger role for Mg2+ revealed by human T-cell immunodeficiency. Nature, 2011, 475, 471-476.	27.8	465
4	Common Severe Infections in Chronic Granulomatous Disease. Clinical Infectious Diseases, 2015, 60, 1176-1183.	5.8	323
5	Coronavirus disease 2019 in patients with inborn errors of immunity: An international study. Journal of Allergy and Clinical Immunology, 2021, 147, 520-531.	2.9	278
6	Mg ²⁺ Regulates Cytotoxic Functions of NK and CD8 T Cells in Chronic EBV Infection Through NKG2D. Science, 2013, 341, 186-191.	12.6	269
7	First-in-human topical microbiome transplantation with Roseomonas mucosa for atopic dermatitis. JCI Insight, 2018, 3, .	5.0	208
8	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2015, 136, 993-1006.e1.	2.9	181
9	XMEN disease: a new primary immunodeficiency affecting Mg2+ regulation of immunity against Epstein-Barr virus. Blood, 2014, 123, 2148-2152.	1.4	147
10	Loss-of-function of the protein kinase C Î´ (PKCÎ´) causes a B-cell lymphoproliferative syndrome in humans. Blood, 2013, 121, 3117-3125.	1.4	138
11	PI3K pathway defects leading to immunodeficiency and immune dysregulation. Journal of Allergy and Clinical Immunology, 2019, 143, 1676-1687.	2.9	104
12	Somatic reversion in dedicator of cytokinesis 8 immunodeficiency modulates disease phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1667-1675.	2.9	82
13	Germline-activating mutations in PIK3CD compromise B cell development and function. Journal of Experimental Medicine, 2018, 215, 2073-2095.	8.5	79
14	Hyperactivated PI3KÎ´ promotes self and commensal reactivity at the expense of optimal humoral immunity. Nature Immunology, 2018, 19, 986-1000.	14.5	77
15	Defective glycosylation and multisystem abnormalities characterize the primary immunodeficiency XMEN disease. Journal of Clinical Investigation, 2019, 130, 507-522.	8.2	74
16	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. Journal of Experimental Medicine, 2015, 212, 855-864.	8.5	70
17	Dedicator of cytokinesis 8â€“deficient CD4 + TÂcells are biased to a T H 2 effector fate at the expense of T H 1 and T H 17Âcells. Journal of Allergy and Clinical Immunology, 2017, 139, 933-949.	2.9	69
18	Activating PIK3CD mutations impair human cytotoxic lymphocyte differentiation and function and EBV immunity. Journal of Allergy and Clinical Immunology, 2019, 143, 276-291.e6.	2.9	64

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19	Identification of a Novel Mutation in MAGT1 and Progressive Multifocal Leucoencephalopathy in a 58-Year-Old Man with XMEN Disease. Journal of Clinical Immunology, 2015, 35, 112-118.	3.8	52
20	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. Journal of Allergy and Clinical Immunology, 2019, 144, 236-253.	2.9	44
21	STAT1 Gain-of-Function Mutations Cause High Total STAT1 Levels With Normal Dephosphorylation. Frontiers in Immunology, 2019, 10, 1433.	4.8	41
22	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. Journal of Allergy and Clinical Immunology, 2022, 149, 410-421.e7.	2.9	34
23	Activated PI3KÎ´ breaches multiple B cell tolerance checkpoints and causes autoantibody production. Journal of Experimental Medicine, 2020, 217, .	8.5	33
24	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	8.5	32
25	Genetic Defects in Phosphoinositide 3-Kinase Î´ Influence CD8+ T Cell Survival, Differentiation, and Function. Frontiers in Immunology, 2018, 9, 1758.	4.8	29
26	Successful Bone Marrow Transplantation for XMEN: Hemorrhagic Risk Uncovered. Journal of Clinical Immunology, 2019, 39, 1-3.	3.8	29
27	Prospective Study of a Novel, Radiation-Free, Reduced-Intensity Bone Marrow Transplantation Platform for Primary Immunodeficiency Diseases. Biology of Blood and Marrow Transplantation, 2020, 26, 94-106.	2.0	28
28	Immune Dysregulation and Disease Pathogenesis due to Activating Mutations in PIK3CDâ€"the Goldilocksâ€™ Effect. Journal of Clinical Immunology, 2019, 39, 148-158.	3.8	26
29	Diversity of XMEN Disease: Description of 2 Novel Variants and Analysis of the Lymphocyte Phenotype. Journal of Clinical Immunology, 2020, 40, 299-309.	3.8	25
30	Genomic Spectrum and Phenotypic Heterogeneity of Human IL-21 Receptor Deficiency. Journal of Clinical Immunology, 2021, 41, 1272-1290.	3.8	25
31	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	8.5	23
32	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	5.0	23
33	Haploinsufficiency of immune checkpoint receptor CTLA4 induces a distinct neuroinflammatory disorder. Journal of Clinical Investigation, 2020, 130, 5551-5561.	8.2	18
34	Primary immunodeficiencies: novel genes and unusual presentations. Hematology American Society of Hematology Education Program, 2019, 2019, 443-448.	2.5	18
35	Disseminated and Congenital Toxoplasmosis in a Mother and Child With Activated PI3-Kinase Î´ Syndrome Type 2 (APDS2): Case Report and a Literature Review of Toxoplasma Infections in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 77.	4.8	16
36	Persistent nodal histoplasmosis in nuclear factor kappa B essential modulator deficiency: Report of a case and review of infection in primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2016, 138, 903-905.	2.9	14

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37	Pathologic Findings in NEMO Deficiency: A Surgical and Autopsy Survey. Pediatric and Developmental Pathology, 2015, 18, 387-400.	1.0	10
38	Nodular regenerative hyperplasia in X-linked agammaglobulinemia: An underestimated and severe complication. Journal of Allergy and Clinical Immunology, 2022, 149, 400-409.e3.	2.9	8
39	Clinical Challenges: Identification of Patients With Novel Primary Immunodeficiency Syndromes. Journal of Pediatric Hematology/Oncology, 2018, 40, e319-e322.	0.6	6
40	Patients With Natural Killer (NK) Cell Chronic Active Epstein-Barr Virus Have Immature NK Cells and Hyperactivation of PI3K/Akt/mTOR and STAT1 Pathways. Journal of Infectious Diseases, 2020, 222, 1170-1179.	4.0	5
41	Cushing syndrome and glucocorticoids: T-cell lymphopenia, apoptosis, and rescue by IL-21. Journal of Allergy and Clinical Immunology, 2022, 149, 302-314.	2.9	4
42	Hyper-IgE Syndrome due to an Elusive Novel Intronic Homozygous Variant in DOCK8. Journal of Clinical Immunology, 2022, 42, 119-129.	3.8	4
43	Autophagy-associated immune dysregulation and hyperplasia in a patient with compound heterozygous mutations in ATG9A. Autophagy, 2023, 19, 678-691.	9.1	4
44	Mapping Out Autoimmunity Control in Primary Immune Regulatory Disorders. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 653-659.	3.8	3
45	Recurrent Nodular Regenerative Hyperplasia Following Liver Transplantation in Common Variable Immunodeficiency. Hepatology, 2021, 74, 1698-1701.	7.3	3