Carsten Speckmann

List of Publications by Year
in descending order

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93 papers	5,212 citations	109321 35 h-index	91884 69 g-index
95 all docs	95 docs citations	95 times ranked	6516 citing authors

#	Article	IF	Citations
1	Autosomal dominant immune dysregulation syndrome in humans with CTLA4 mutations. Nature Medicine, 2014, 20, 1410-1416.	30.7	723
2	Clinical spectrum and features of activated phosphoinositide 3-kinase Î´ syndrome: AÂlarge patient cohort study. Journal of Allergy and Clinical Immunology, 2017, 139, 597-606.e4.	2.9	377
3	Phenotype, penetrance, and treatment of 133 cytotoxic T-lymphocyte antigen 4â€“insufficient subjects. Journal of Allergy and Clinical Immunology, 2018, 142, 1932-1946.	2.9	344
4	The extended phenotype of LPS-responsive beige-like anchor protein (LRBA) deficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 223-230.	2.9	247
5	Long-term follow-up of IPEX syndrome patients after different therapeutic strategies: An international multicenter retrospective study. Journal of Allergy and Clinical Immunology, 2018, 141, 1036-1049.e5.	2.9	233
6	Lethal hemophagocytic lymphohistiocytosis in Hermansky-Pudlak syndrome type II. Blood, 2006, 108, 81-87.	1.4	194
7	The syndrome of hemophagocytic lymphohistiocytosis in primary immunodeficiencies: implications for differential diagnosis and pathogenesis. Haematologica, 2015, 100, 978-988.	3.5	161
8	X-linked inhibitor of apoptosis (XIAP) deficiency: The spectrum of presenting manifestations beyond hemophagocytic lymphohistiocytosis. Clinical Immunology, 2013, 149, 133-141.	3.2	158
9	Clinical and immunological manifestations of patients with atypical severe combined immunodeficiency. Clinical Immunology, 2011, 141, 73-82.	3.2	157
10	Antiviral and Regulatory T Cell Immunity in a Patient with Stromal Interaction Molecule 1 Deficiency. Journal of Immunology, 2012, 188, 1523-1533.	0.8	156
11	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	6.2	148
12	Diseaseâ€causing mutations in the <scp>XIAP</scp> <scp>BIR</scp> 2 domain impair <scp>NOD</scp> 2â€dependent immune signalling. EMBO Molecular Medicine, 2013, 5, 1278-1295.	6.9	137
13	Atypical familial hemophagocytic lymphohistiocytosis due to mutations in UNC13D and STXBP2 overlaps with primary immunodeficiency diseases. Haematologica, 2010, 95, 2080-2087.	3.5	109
14	Epigenetic immune cell counting in human blood samples for immunodiagnostics. Science Translational Medicine, 2018, 10, .	12.4	83
15	The expansion of human T-bet ^{high} CD21 ^{low} B cells is T cell dependent. Science Immunology, 2021, 6, eabh0891.	11.9	82
16	Hyperactive mTOR pathway promotes lymphoproliferation and abnormal differentiation in autoimmune lymphoproliferative syndrome. Blood, 2016, 128, 227-238.	1.4	77
17	A new functional assay for the diagnosis of X-linked inhibitor of apoptosis (XIAP) deficiency. Clinical and Experimental Immunology, 2014, 176, 394-400.	2.6	75
18	A prospective study on the natural history of patients with profound combined immunodeficiency: An interim analysis. Journal of Allergy and Clinical Immunology, 2017, 139, 1302-1310.e4.	2.9	71

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19	The German National Registry of Primary Immunodeficiencies (2012â€“2017). Frontiers in Immunology, 2019, 10, 1272.	4.8	71
20	Tandem mass spectrometry, but not T-cell receptor excision circle analysis, identifies newborns with late-onset adenosine deaminase deficiency. Journal of Allergy and Clinical Immunology, 2013, 131, 1604-1610.	2.9	65
21	ORAI1 mutations abolishing store-operated Ca2+ entry cause anhidrotic ectodermal dysplasia with immunodeficiency. Journal of Allergy and Clinical Immunology, 2018, 142, 1297-1310.e11.	2.9	62
22	Hematopoietic stem cell transplantation for CD40 ligand deficiency: Results from an EBMT/ESID-IEWP-SCETIDE-PIDTC study. Journal of Allergy and Clinical Immunology, 2019, 143, 2238-2253.	2.9	60
23	Clinical and immunologic consequences of a somatic reversion in a patient with X-linked severe combined immunodeficiency. Blood, 2008, 112, 4090-4097.	1.4	59
24	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots. Journal of Allergy and Clinical Immunology, 2014, 134, 155-159.e3.	2.9	56
25	Abnormally differentiated CD4+ or CD8+ T cells with phenotypic and genetic features of double negative T cells in human Fas deficiency. Blood, 2014, 124, 851-860.	1.4	54
26	Hematopoietic cell transplantation in severe combined immunodeficiency: The SCETIDE 2006-2014 European cohort. Journal of Allergy and Clinical Immunology, 2022, 149, 1744-1754.e8.	2.9	51
27	Evolution of disease activity and biomarkers on and off rapamycin in 28 patients with autoimmune lymphoproliferative syndrome. Haematologica, 2017, 102, e52-e56.	3.5	49
28	Activated <scp>PI</scp>3KÎ´ syndrome type 2: Two patients, a novel mutation, and review of the literature. Pediatric Allergy and Immunology, 2016, 27, 640-644.	2.6	46
29	Delayed-onset adenosine deaminase deficiency: Strategies for an early diagnosis. Journal of Allergy and Clinical Immunology, 2012, 130, 991-994.	2.9	44
30	Chronic Inflammatory Bowel Disease as Key Manifestation of Atypical ARTEMIS Deficiency. Journal of Clinical Immunology, 2010, 30, 314-320.	3.8	42
31	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 2303-2306.	2.9	40
32	Patients with T^{+/low} NK⁺ ILâ€2 receptor Î³ chain deficiency have differentiallyâ€impaired cytokine signaling resulting in severe combined immunodeficiency. European Journal of Immunology, 2014, 44, 3129-3140.	2.9	39
33	XIAP deficiency is a mendelian cause of late-onset IBD. Gut, 2014, 63, 1031-1032.	12.1	38
34	SYK expression endows human ZAP70-deficient CD8 T cells with residual TCR signaling. Clinical Immunology, 2015, 161, 103-109.	3.2	38
35	Transplantation from a symptomatic carrier sister restores host defenses but does not prevent colitis in NEMO deficiency. Clinical Immunology, 2016, 164, 52-56.	3.2	38
36	Omenn syndrome associated with a functional reversion due to a somatic second-site mutation in CARD11 deficiency. Blood, 2015, 126, 1658-1669.	1.4	37

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37	Effective Immunological Guidance of Genetic Analyses Including Exome Sequencing in Patients Evaluated for Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2017, 37, 770-780.	3.8	37
38	Variable impairment of platelet functions in patients with severe, genetically linked immune deficiencies. Haematologica, 2018, 103, 540-549.	3.5	36
39	Clinical and Molecular Heterogeneity of RTEL1 Deficiency. Frontiers in Immunology, 2017, 8, 449.	4.8	35
40	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
41	Targeted Gene Panel Sequencing for Early-onset Inflammatory Bowel Disease and Chronic Diarrhea. Inflammatory Bowel Diseases, 2017, 23, 2109-2120.	1.9	33
42	Neuroinflammatory Disease as an Isolated Manifestation of Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2020, 40, 901-916.	3.8	33
43	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.8	33
44	SARS-CoV-2 in children with cancer or after haematopoietic stem cell transplant: An analysis of 131 patients. European Journal of Cancer, 2021, 159, 78-86.	2.8	32
45	Sequential decisions on FAS sequencing guided by biomarkers in patients with lymphoproliferation and autoimmune cytopenia. Haematologica, 2013, 98, 1948-1955.	3.5	29
46	Symptomatic Males and Female Carriers in a Large Caucasian Kindred with XIAP Deficiency. Journal of Clinical Immunology, 2015, 35, 439-444.	3.8	29
47	X-linked Inhibitor of Apoptosis Complicated by Granulomatous Lymphocytic Interstitial Lung Disease (GLILD) and Granulomatous Hepatitis. Journal of Clinical Immunology, 2016, 36, 733-738.	3.8	25
48	Disturbed B-lymphocyte selection in autoimmune lymphoproliferative syndrome. Blood, 2016, 127, 2193-2202.	1.4	25
49	A distinct CD38+CD45RA+ population of CD4+, CD8+, and double-negative T cells is controlled by FAS. Journal of Experimental Medicine, 2021, 218, .	8.5	25
50	Hemophagocytic lymphohistiocytosis as presenting manifestation of profound combined immunodeficiency due to an ORAI1 mutation. Journal of Allergy and Clinical Immunology, 2017, 140, 1721-1724.	2.9	23
51	A RAB27A 5â€² untranslated region structural variant associated with late-onset hemophagocytic lymphohistiocytosis and normal pigmentation. Journal of Allergy and Clinical Immunology, 2018, 142, 317-321.e8.	2.9	22
52	Gray platelet syndrome can mimic autoimmune lymphoproliferative syndrome. Blood, 2015, 126, 1967-1969.	1.4	21
53	Outcome of chronic granulomatous disease â€•Conventional treatment vs stem cell transplantation. Pediatric Allergy and Immunology, 2021, 32, 576-585.	2.6	21
54	Newborn screening for severe combined immunodeficiency using a novel and simplified method to measure T-cell excision circles (TREC). Clinical Immunology, 2017, 175, 51-55.	3.2	20

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55	Profound immunodeficiency with severe skin disease explained by concomitant novel CARMIL2 and PLEC1 loss-of-function mutations. Clinical Immunology, 2019, 208, 108228.	3.2	20
56	Hematopoietic stem cell transplantation for adolescents and adults with inborn errors of immunity: an EBMT IEWP study. Blood, 2022, 140, 1635-1649.	1.4	20
57	Preserved effector functions of human ORAI1- and STIM1-deficient neutrophils. Journal of Allergy and Clinical Immunology, 2016, 137, 1587-1591.e7.	2.9	16
58	Reversible pancytopenia and immunodeficiency in a patient with hereditary folate malabsorption. Pediatric Blood and Cancer, 2015, 62, 1091-1094.	1.5	15
59	High Levels of IL-18 and IFN-Î³ in Chronically Inflamed Tissue in Chronic Granulomatous Disease. Frontiers in Immunology, 2019, 10, 2236.	4.8	15
60	Phenotype, genotype, treatment, and survival outcomes in patients with X-linked inhibitor of apoptosis deficiency. Journal of Allergy and Clinical Immunology, 2022, 150, 456-466.	2.9	15
61	Daratumumab therapy for post-HSCT immune-mediated cytopenia: experiences from two pediatric cases and review of literature. Molecular and Cellular Pediatrics, 2021, 8, 5.	1.8	13
62	The <scp>BEACH</scp> protein <scp>LRBA</scp> is required for hair bundle maintenance in cochlear hair cells and for hearing. EMBO Reports, 2017, 18, 2015-2029.	4.5	12
63	Agammaglobulinemia with normal B-cell numbers in a patient lacking Bob1. Journal of Allergy and Clinical Immunology, 2021, 147, 1977-1980.	2.9	12
64	Platelet secretion defect in a patient with stromal interaction molecule 1 deficiency. Blood, 2013, 122, 3696-3698.	1.4	11
65	Rubella vaccineâ€“induced granulomas are a novel phenotype with incomplete penetrance of genetic defects in cytotoxicity. Journal of Allergy and Clinical Immunology, 2022, 149, 388-399.e4.	2.9	11
66	Ruxolitinib Controls Lymphoproliferation and Diabetes in a STAT3-GOF Patient. Journal of Clinical Immunology, 2020, 40, 1207-1210.	3.8	10
67	XIAP deficiency and MEFV variants resulting in an autoinflammatory lymphoproliferative syndrome. BMJ Case Reports, 2016, 2016, bcr2016216922.	0.5	9
68	Incidence of SCID in Germany from 2014 to 2015 an ESPED* Survey on Behalf of the API*** Erhebungseinheit fÃ¼r Seltene PÃdiatrische Erkrankungen in Deutschland (German Paediatric) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 22 2020, 40, 708-717.	3.8	9
69	Reduced-Intensity/Reduced-Toxicity Conditioning Approaches Are Tolerated in XIAP Deficiency but Patients Fare Poorly with Acute GVHD. Journal of Clinical Immunology, 2021, , 1.	3.8	9
70	Risk factors for mixed chimerism in children with hemophagocytic lymphohistiocytosis after reduced toxicity conditioning. Pediatric Blood and Cancer, 2020, 67, e28523.	1.5	8
71	Human RAD52 â€“ a novel player in DNA repair in cancer and immunodeficiency. Haematologica, 2017, 102, e69-e72.	3.5	7
72	Secondary C1q Deficiency in Activated PI3KÎ´ Syndrome Type 2. Frontiers in Immunology, 2019, 10, 2589.	4.8	7

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73	Increased proportions of Î³Î´ T lymphocytes in atypical SCID associate with disease manifestations. Clinical Immunology, 2019, 201, 30-34.	3.2	6
74	IgG4-related disease in autoimmune lymphoproliferative syndrome. Clinical Immunology, 2017, 180, 97-99.	3.2	5
75	Functional flow cytometry of monocytes for routine diagnosis of innate primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2020, 145, 434-437.e4.	2.9	5
76	Long-term robustness of a T-cell system emerging from somatic rescue of a genetic block in T-cell development. EBioMedicine, 2020, 59, 102961.	6.1	5
77	Activation-induced deaminase is critical for the establishment of DNA methylation patterns prior to the germinal center reaction. Nucleic Acids Research, 2021, 49, 5057-5073.	14.5	5
78	Curative Treatment of POMP-Related Autoinflammation and Immune Dysregulation (PRAID) by Hematopoietic Stem Cell Transplantation. Journal of Clinical Immunology, 2021, 41, 1664-1667.	3.8	5
79	Tuberculosis-Associated HLH in an 8-Month-Old Infant: A Case Report and Review. Frontiers in Pediatrics, 2020, 8, 556155.	1.9	4
80	Lipid Apheresis to Manage Severe Hypertriglyceridemia during Induction Therapy in a Child with Acute Lymphoblastic Leukemia. Pediatric Hematology and Oncology, 2020, 37, 530-538.	0.8	4
81	Pulmonary granulomatosis of genetic origin. European Respiratory Review, 2021, 30, 200152.	7.1	4
82	Genetic Disorders of Immune Regulation. , 2017, , 295-338.		4
83	RAG1 deficiency with a shifting B cell phenotype and chromosomal instability. Clinical Immunology, 2014, 152, 65-67.	3.2	3
84	NAXD Deficiency Associated with Perinatal Autoinflammation, Pancytopenia, Dermatitis, Colitis, and Cystic Encephalomalacia. Journal of Pediatric Neurology, 2021, 19, 105-108.	0.2	2
85	Ledipasvir/Sofosbuvir Eradicates Hepatitis C in an Immunodeficient STAT3-GOF Patient. Journal of Clinical Immunology, 2021, 41, 1365-1367.	3.8	2
86	A prospective outcome study of patients with profound combined immunodeficiency (P-CID). LymphoSign Journal, 2015, 2, 91-106.	0.2	2
87	Autoinflammatory Diseases Predominantly Affecting the Gastrointestinal Tract. , 2014, , 573-584.		1
88	Hyperactive mTOR Pathway Promotes Lymphoproliferation and Abnormal Differentiation in Human Autoimmune Lymphoproliferative Syndrome. Blood, 2015, 126, 1020-1020.	1.4	1
89	Pulmonary Manifestations of Genetic Disorders of Immune Regulation. , 2019, , 145-168.		0
90	Rheumatologische und immunologische Krankheitsbilder. , 2013, , 731-744.		0

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91	T-zellulÃre und kombinierte Immundefekte. , 2014, , 704-720.		0
92	T-zellulÃre und kombinierte Immundefekte bei Kindern und Jugendlichen. Springer Reference Medizin, 2020, , 1-24.	0.0	0
93	T-zellulÃre und kombinierte Immundefekte. Springer Reference Medizin, 2020, , 1003-1026.	0.0	0