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	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
2	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
3	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
4	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
5	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
6	NAXD Deficiency Associated with Perinatal Autoinflammation, Pancytopenia, Dermatitis, Colitis, and Cystic Encephalomalacia. Journal of Pediatric Neurology, 2021, 19, 105-108.	0.2	2
7	Outcome of chronic granulomatous disease â€•Conventional treatment vs stem cell transplantation. Pediatric Allergy and Immunology, 2021, 32, 576-585.	2.6	21
8	Ledipasvir/Sofosbuvir Eradicates Hepatitis C in an Immunodeficient STAT3-GOF Patient. Journal of Clinical Immunology, 2021, 41, 1365-1367.	3.8	2
9	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
10	Pulmonary granulomatosis of genetic origin. European Respiratory Review, 2021, 30, 200152.	7.1	4
11	Agammaglobulinemia with normal B-cell numbers in a patient lacking Bob1. Journal of Allergy and Clinical Immunology, 2021, 147, 1977-1980.	2.9	12
12	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
13	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
14	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.8	33
15	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
16	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
17	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
18	The expansion of human T-bet ^{high} CD21 ^{low} B cells is T cell dependent. Science Immunology, 2021, 6, eabh0891.	11.9	82

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19	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
20	Ruxolitinib Controls Lymphoproliferation and Diabetes in a STAT3-GOF Patient. Journal of Clinical Immunology, 2020, 40, 1207-1210.	3.8	10
21	Tuberculosis-Associated HLH in an 8-Month-Old Infant: A Case Report and Review. Frontiers in Pediatrics, 2020, 8, 556155.	1.9	4
22	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
23	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
24	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 54 2020, 40, 708-717.	3.8	9
25	Risk factors for mixed chimerism in children with hemophagocytic lymphohistiocytosis after reduced toxicity conditioning. Pediatric Blood and Cancer, 2020, 67, e28523.	1.5	8
26	Neuroinflammatory Disease as an Isolated Manifestation of Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2020, 40, 901-916.	3.8	33
27	T-zellulÃre und kombinierte Immundefekte bei Kindern und Jugendlichen. Springer Reference Medizin, 2020, , 1-24.	0.0	0
28	T-zellulÃre und kombinierte Immundefekte. Springer Reference Medizin, 2020, , 1003-1026.	0.0	0
29	The German National Registry of Primary Immunodeficiencies (2012â€“2017). Frontiers in Immunology, 2019, 10, 1272.	4.8	71
30	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
31	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
32	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
33	Increased proportions of Î³Î´ T lymphocytes in atypical SCID associate with disease manifestations. Clinical Immunology, 2019, 201, 30-34.	3.2	6
34	Secondary C1q Deficiency in Activated PI3KÎ´ Syndrome Type 2. Frontiers in Immunology, 2019, 10, 2589.	4.8	7
35	Pulmonary Manifestations of Genetic Disorders of Immune Regulation. , 2019, , 145-168.		0
36	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

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37	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
38	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
39	Variable impairment of platelet functions in patients with severe, genetically linked immune deficiencies. Haematologica, 2018, 103, 540-549.	3.5	36
40	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
41	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
42	Epigenetic immune cell counting in human blood samples for immunodiagnostics. Science Translational Medicine, 2018, 10, .	12.4	83
43	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
44	IgG4-related disease in autoimmune lymphoproliferative syndrome. Clinical Immunology, 2017, 180, 97-99.	3.2	5
45	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
46	Human RAD52 â€“ a novel player in DNA repair in cancer and immunodeficiency. Haematologica, 2017, 102, e69-e72.	3.5	7
47	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
48	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
49	Targeted Gene Panel Sequencing for Early-onset Inflammatory Bowel Disease and Chronic Diarrhea. Inflammatory Bowel Diseases, 2017, 23, 2109-2120.	1.9	33
50	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
51	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
52	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
53	Clinical and Molecular Heterogeneity of RTEL1 Deficiency. Frontiers in Immunology, 2017, 8, 449.	4.8	35
54	Genetic Disorders of Immune Regulation. , 2017, , 295-338.		4

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55	XIAP deficiency and MEFV variants resulting in an autoinflammatory lymphoproliferative syndrome. BMJ Case Reports, 2016, 2016, bcr2016216922.	0.5	9
56	Preserved effector functions of human ORAI1- and STIM1-deficient neutrophils. Journal of Allergy and Clinical Immunology, 2016, 137, 1587-1591.e7.	2.9	16
57	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
58	Disturbed B-lymphocyte selection in autoimmune lymphoproliferative syndrome. Blood, 2016, 127, 2193-2202.	1.4	25
59	Hyperactive mTOR pathway promotes lymphoproliferation and abnormal differentiation in autoimmune lymphoproliferative syndrome. Blood, 2016, 128, 227-238.	1.4	77
60	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
61	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
62	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
63	Gray platelet syndrome can mimic autoimmune lymphoproliferative syndrome. Blood, 2015, 126, 1967-1969.	1.4	21
64	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
65	The syndrome of hemophagocytic lymphohistiocytosis in primary immunodeficiencies: implications for differential diagnosis and pathogenesis. Haematologica, 2015, 100, 978-988.	3.5	161
66	SYK expression endows human ZAP70-deficient CD8 T cells with residual TCR signaling. Clinical Immunology, 2015, 161, 103-109.	3.2	38
67	Symptomatic Males and Female Carriers in a Large Caucasian Kindred with XIAP Deficiency. Journal of Clinical Immunology, 2015, 35, 439-444.	3.8	29
68	Reversible pancytopenia and immunodeficiency in a patient with hereditary folate malabsorption. Pediatric Blood and Cancer, 2015, 62, 1091-1094.	1.5	15
69	A prospective outcome study of patients with profound combined immunodeficiency (P-CID). LymphoSign Journal, 2015, 2, 91-106.	0.2	2
70	Hyperactive mTOR Pathway Promotes Lymphoproliferation and Abnormal Differentiation in Human Autoimmune Lymphoproliferative Syndrome. Blood, 2015, 126, 1020-1020.	1.4	1
71	Autoinflammatory Diseases Predominantly Affecting the Gastrointestinal Tract. , 2014, , 573-584.		1
72	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

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73	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
74	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
75	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
76	Autosomal dominant immune dysregulation syndrome in humans with CTLA4 mutations. Nature Medicine, 2014, 20, 1410-1416.	30.7	723
77	XIAP deficiency is a mendelian cause of late-onset IBD. Gut, 2014, 63, 1031-1032.	12.1	38
78	RAG1 deficiency with a shifting B cell phenotype and chromosomal instability. Clinical Immunology, 2014, 152, 65-67.	3.2	3
79	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
80	T-zellulÃre und kombinierte Immundefekte. , 2014, , 704-720.		0
81	X-linked inhibitor of apoptosis (XIAP) deficiency: The spectrum of presenting manifestations beyond hemophagocytic lymphohistiocytosis. Clinical Immunology, 2013, 149, 133-141.	3.2	158
82	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
83	Sequential decisions on FAS sequencing guided by biomarkers in patients with lymphoproliferation and autoimmune cytopenia. Haematologica, 2013, 98, 1948-1955.	3.5	29
84	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
85	Platelet secretion defect in a patient with stromal interaction molecule 1 deficiency. Blood, 2013, 122, 3696-3698.	1.4	11
86	Rheumatologische und immunologische Krankheitsbilder. , 2013, , 731-744.		0
87	Delayed-onset adenosine deaminase deficiency: Strategies for an early diagnosis. Journal of Allergy and Clinical Immunology, 2012, 130, 991-994.	2.9	44
88	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
89	Clinical and immunological manifestations of patients with atypical severe combined immunodeficiency. Clinical Immunology, 2011, 141, 73-82.	3.2	157
90	Atypical familial hemophagocytic lymphohistiocytosis due to mutations in UNC13D and STXBP2 overlaps with primary immunodeficiency diseases. Haematologica, 2010, 95, 2080-2087.	3.5	109

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91	Chronic Inflammatory Bowel Disease as Key Manifestation of Atypical ARTEMIS Deficiency. Journal of Clinical Immunology, 2010, 30, 314-320.	3.8	42
92	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
93	Lethal hemophagocytic lymphohistiocytosis in Hermansky-Pudlak syndrome type II. Blood, 2006, 108, 81-87.	1.4	194