

# Michel J Massaad

## List of Publications by Year in descending order

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64  
papers

3,453  
citations

186265

28  
h-index

144013

57  
g-index

66  
all docs

66  
docs citations

66  
times ranked

4861  
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnosis and Treatment of a Patient With Severe Combined Immunodeficiency Due to a Novel Homozygous Mutation in the IL-7R $\alpha$ Chain. <i>Frontiers in Immunology</i> , 2022, 13, 867837.	4.8	5
2	Challenges in glioblastoma immunotherapy: mechanisms of resistance and therapeutic approaches to overcome them. <i>British Journal of Cancer</i> , 2022, 127, 976-987.	6.4	26
3	Combined immunodeficiency due to a mutation in the $\beta$ 1 subunit of the coat protein I complex. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	15
4	Hypomorphic variants in AK2 reveal the contribution of mitochondrial function to B-cell activation. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 192-202.	2.9	13
5	Wiskott-Aldrich Syndrome in four male siblings from a consanguineous family from Lebanon. <i>Clinical Immunology</i> , 2020, 219, 108573.	3.2	6
6	Frequency and Manifestations of Autoimmunity Among Children Registered in the Kuwait National Primary Immunodeficiency Registry. <i>Frontiers in Immunology</i> , 2020, 11, 1119.	4.8	19
7	Inherited human IFN- $\beta$ deficiency underlies mycobacterial disease. <i>Journal of Clinical Investigation</i> , 2020, 130, 3158-3171.	8.2	89
8	Combined immunodeficiency in a patient with c-Rel deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 606-608.e4.	2.9	32
9	Rethinking newborn screening for severe combined immunodeficiency: Lessons from an international partnership for patients with primary immunodeficiencies in Pakistan. <i>Clinical Immunology</i> , 2019, 202, 29-32.	3.2	2
10	Mechanisms of genotype-phenotype correlation in autosomal dominant anhidrotic ectodermal dysplasia with immune deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1060-1073.e3.	2.9	22
11	DNA recombination defects in Kuwait: Clinical, immunologic and genetic profile. <i>Clinical Immunology</i> , 2018, 187, 68-75.	3.2	11
12	Clinical, immunologic, and genetic spectrum of 696 patients with combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1450-1458.	2.9	90
13	The Lack of WIP Binding to Actin Results in Impaired B Cell Migration and Altered Humoral Immune Responses. <i>Cell Reports</i> , 2018, 24, 619-629.	6.4	17
14	Comprehensive Genetic Results for Primary Immunodeficiency Disorders in a Highly Consanguineous Population. <i>Frontiers in Immunology</i> , 2018, 9, 3146.	4.8	37
15	Janus kinase 3 deficiency caused by a homozygous synonymous exonic mutation that creates a dominant splice site. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 268-271.e6.	2.9	11
16	DOCK8 Deficiency Presenting as an IPEX-Like Disorder. <i>Journal of Clinical Immunology</i> , 2017, 37, 811-819.	3.8	39
17	A novel mutation in the JH4 domain of JAK3 causing severe combined immunodeficiency complicated by vertebral osteomyelitis. <i>Clinical Immunology</i> , 2017, 183, 198-200.	3.2	6
18	DOCK8 and STAT3 dependent inhibition of IgE isotype switching by TLR9 ligation in human B cells. <i>Clinical Immunology</i> , 2017, 183, 263-265.	3.2	13

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19	Disseminated Mycobacterium malmoeense and Salmonella Infections Associated with a Novel Variant in NFKBIA. Journal of Clinical Immunology, 2017, 37, 415-418.	3.8	13
20	Epidermodysplasia verruciformis as a manifestation of ARTEMIS deficiency in a young adult. Journal of Allergy and Clinical Immunology, 2017, 139, 372-375.e4.	2.9	18
21	Cadherin 17 mutation associated with leaky severe combined immune deficiency is corrected by HSCT. Blood Advances, 2017, 1, 2083-2087.	5.2	3
22	Natural Killer Cells from Patients with Recombinase-Activating Gene and Non-Homologous End Joining Gene Defects Comprise a Higher Frequency of CD56bright NKG2A+++ Cells, and Yet Display Increased Degranulation and Higher Perforin Content. Frontiers in Immunology, 2017, 8, 798.	4.8	41
23	Chronic mucocutaneous candidiasis associated with an SH2 domain gain-of-function mutation that enhances STAT1 phosphorylation. Journal of Allergy and Clinical Immunology, 2016, 138, 297-299.	2.9	24
24	Hematopoietic stem cell transplantation outcomes for 11 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 852-859.e3.	2.9	48
25	Combined immunodeficiency due to a homozygous mutation in ORAI1 that deletes the C-terminus that interacts with STIM 1. Clinical Immunology, 2016, 166-167, 100-102.	3.2	11
26	Mutations in pyrin masquerading as a primary immunodeficiency. Clinical Immunology, 2016, 171, 65-66.	3.2	2
27	A missense mutation in TFRC, encoding transferrin receptor 1, causes combined immunodeficiency. Nature Genetics, 2016, 48, 74-78.	21.4	219
28	Spectrum of Phenotypes Associated with Mutations in LRBA. Journal of Clinical Immunology, 2016, 36, 33-45.	3.8	180
29	Recurrent viral infections associated with a homozygous CORO1A mutation that disrupts oligomerization and cytoskeletal association. Journal of Allergy and Clinical Immunology, 2016, 137, 879-888.e2.	2.9	41
30	Autoimmune lymphoproliferative syndrome caused by a homozygous FasL mutation that disrupts FasL assembly. Journal of Allergy and Clinical Immunology, 2016, 137, 324-327.e2.	2.9	13
31	Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. Journal of Clinical Investigation, 2016, 126, 4219-4236.	8.2	56
32	A DOCK8-WIP-WASp complex links T cell receptors to the actin cytoskeleton. Journal of Clinical Investigation, 2016, 126, 3837-3851.	8.2	93
33	The extended clinical phenotype of 64 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 402-412.	2.9	163
34	Defective lymphoid organogenesis underlies the immune deficiency caused by a heterozygous S32I mutation in I $\beta$ B1 $\pm$ . Journal of Experimental Medicine, 2015, 212, 185-202.	8.5	25
35	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	27.0	169
36	Mutations in Recombination Activating Gene 1 and 2 in patients with severe combined immunodeficiency disorders in Egypt. Clinical Immunology, 2015, 158, 167-173.	3.2	10

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37	The Rho GTPase Cdc42 Is Essential for the Activation and Function of Mature B Cells. <i>Journal of Immunology</i> , 2015, 194, 4750-4758.	0.8	26
38	A novel mutation in ICOS presenting as hypogammaglobulinemia with susceptibility to opportunistic pathogens. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 794-797.e1.	2.9	26
39	Regulatory T-cell deficiency and immune dysregulation, polyendocrinopathy, enteropathy, X-linked-like disorder caused by loss-of-function mutations in LRBA. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 217-227.e9.	2.9	223
40	Flow cytometry diagnosis of dedicator of cytokinesis 8 (DOCK8) deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 221-223.e7.	2.9	62
41	A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1099-1108.e12.	2.9	132
42	Binding of WIP to Actin Is Essential for T Cell Actin Cytoskeleton Integrity and Tissue Homing. <i>Molecular and Cellular Biology</i> , 2014, 34, 4343-4354.	2.3	21
43	A novel mutation in FOXP1 resulting in SCID: A case report and literature review. <i>Clinical Immunology</i> , 2014, 155, 30-32.	3.2	32
44	Binding of the WASP/N-WASP-Interacting Protein WIP to Actin Regulates Focal Adhesion Assembly and Adhesion. <i>Molecular and Cellular Biology</i> , 2014, 34, 2600-2610.	2.3	18
45	Lessons in gene hunting: A RAG1 mutation presenting with agammaglobulinemia and absence of B cells. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 983-985.e1.	2.9	22
46	Successful Hematopoietic Stem Cell Transplant For CD40 Deficiency Manifesting As Hyper-IgM Syndrome With Absent CD40 Expression and Marked Lymphocytosis. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, AB95.	2.9	1
47	A novel disease-causing CD40L mutation reduces expression of CD40 ligand, but preserves CD40 binding capacity. <i>Clinical Immunology</i> , 2014, 153, 288-291.	3.2	9
48	Successful interferon-alpha 2b therapy for unremitting warts in a patient with DOCK8 deficiency. <i>Clinical Immunology</i> , 2014, 153, 104-108.	3.2	29
49	T-cell receptor ligation causes Wiskott-Aldrich syndrome protein degradation and F-actin assembly downregulation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 648-655.e1.	2.9	42
50	Intronic SH2D1A mutation with impaired SAP expression and agammaglobulinemia. <i>Clinical Immunology</i> , 2013, 146, 84-89.	3.2	6
51	Wiskott-Aldrich syndrome: a comprehensive review. <i>Annals of the New York Academy of Sciences</i> , 2013, 1285, 26-43.	3.8	297
52	A homozygous mucosa-associated lymphoid tissue 1 (MALT1) mutation in a family with combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 151-158.	2.9	124
53	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. <i>Journal of Experimental Medicine</i> , 2012, 209, 29-34.	8.5	158
54	B cell-intrinsic deficiency of the Wiskott-Aldrich syndrome protein (WASP) causes severe abnormalities of the peripheral B-cell compartment in mice. <i>Blood</i> , 2012, 119, 2819-2828.	1.4	99

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55	A novel homozygous mutation in recombination activating gene 2 in 2 relatives with different clinical phenotypes: Omenn syndrome and hyper-IgM syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 1414-1416.	2.9	43
56	DOCK8 functions as an adaptor that links TLR-MyD88 signaling to B cell activation. <i>Nature Immunology</i> , 2012, 13, 612-620.	14.5	205
57	Clinical, immunologic and genetic profiles of DOCK8-deficient patients in Kuwait. <i>Clinical Immunology</i> , 2012, 143, 266-272.	3.2	60
58	A peptide derived from the Wiskott-Aldrich syndrome (WAS) protein-interacting protein (WIP) restores WAS protein level and actin cytoskeleton reorganization in lymphocytes from patients with WAS mutations that disrupt WIP binding. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 998-1005.e2.	2.9	25
59	Cdc42 interacting protein 4 (CIP4) is essential for integrin-dependent T-cell trafficking. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 16252-16256.	7.1	23
60	WIP is critical for T cell responsiveness to IL-2. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 7519-7524.	7.1	22
61	A novel anti-WIP monoclonal antibody detects an isoform of WIP that lacks the WASP binding domain. <i>Biochemical and Biophysical Research Communications</i> , 2007, 353, 875-881.	2.1	20
62	Defective nuclear translocation of nuclear factor of activated T cells and extracellular signal-regulated kinase underlies deficient IL-2 gene expression in Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2005, 116, 1364-1371.	2.9	56
63	The Yeast Split-Ubiquitin Membrane Protein Two-Hybrid Screen Identifies BAP31 as a Regulator of the Turnover of Endoplasmic Reticulum-Associated Protein Tyrosine Phosphatase-Like B. <i>Molecular and Cellular Biology</i> , 2004, 24, 2767-2778.	2.3	58
64	The processing of $\alpha$ 1,2-mannosidase of <i>Saccharomyces cerevisiae</i> depends on Rer1p for its localization in the endoplasmic reticulum. <i>European Journal of Cell Biology</i> , 1999, 78, 435-440.	3.6	20