

# Timothy Ronan Leahy

## List of Publications by Year in descending order

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Version: 2024-02-01

57  
papers

2,424  
citations

331670

21  
h-index

214800

47  
g-index

57  
all docs

57  
docs citations

57  
times ranked

4120  
citing authors

#	ARTICLE	IF	CITATIONS
1	Phosphoinositide 3-Kinase Î Gene Mutation Predisposes to Respiratory Infection and Airway Damage. <i>Science</i> , 2013, 342, 866-871.	12.6	541
2	Clinical spectrum and features of activated phosphoinositide 3-kinase Î syndrome: A large patient cohort study. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 597-606.e4.	2.9	377
3	Human IFNAR2 deficiency: Lessons for antiviral immunity. <i>Science Translational Medicine</i> , 2015, 7, 307ra154.	12.4	190
4	Clinical and Immunological Phenotype of Patients With Primary Immunodeficiency Due to Damaging Mutations in NFKB2. <i>Frontiers in Immunology</i> , 2019, 10, 297.	4.8	117
5	Hypomorphic caspase activation and recruitment domain 11 (CARD11) mutations associated with diverse immunologic phenotypes with or without atopic disease. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1482-1495.	2.9	116
6	Thymus transplantation for complete DiGeorge syndrome: European experience. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 1660-1670.e16.	2.9	108
7	An essential role for the Zn <sup>2+</sup> transporter ZIP7 in B cell development. <i>Nature Immunology</i> , 2019, 20, 350-361.	14.5	92
8	Somatic STAT5b gain-of-function mutations in early onset nonclonal eosinophilia, urticaria, dermatitis, and diarrhea. <i>Blood</i> , 2017, 129, 650-653.	1.4	74
9	Early-onset autoimmunity associated with SOCS1 haploinsufficiency. <i>Nature Communications</i> , 2020, 11, 5341.	12.8	74
10	Changes in airway inflammation during pulmonary exacerbations in patients with cystic fibrosis and primary ciliary dyskinesia. <i>European Respiratory Journal</i> , 2016, 47, 829-836.	6.7	66
11	Functional characterization of the human dendritic cell immunodeficiency associated with the IRF8K108E mutation. <i>Blood</i> , 2014, 124, 1894-1904.	1.4	65
12	Increased systemic inflammation in children with Down syndrome. <i>Cytokine</i> , 2020, 127, 154938.	3.2	49
13	Randomized controlled trial of biofilm antimicrobial susceptibility testing in cystic fibrosis patients. <i>Journal of Cystic Fibrosis</i> , 2015, 14, 262-266.	0.7	45
14	Multiorgan involvement and management in children with Down syndrome. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2020, 109, 1096-1111.	1.5	40
15	Interferon signature in patients with STAT1 gain-of-function mutation is epigenetically determined. <i>European Journal of Immunology</i> , 2019, 49, 790-800.	2.9	39
16	Haploidentical T-cell alpha beta receptor and CD19-depleted stem cell transplant for Wiskott-Aldrich syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 1199-1201.	2.9	36
17	Targeted Gene Panel Sequencing for Early-onset Inflammatory Bowel Disease and Chronic Diarrhea. <i>Inflammatory Bowel Diseases</i> , 2017, 23, 2109-2120.	1.9	33
18	Mumps Outbreaks in Vaccinated Populations—Is It Time to Re-assess the Clinical Efficacy of Vaccines?. <i>Frontiers in Immunology</i> , 2020, 11, 2089.	4.8	30

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19	<sc>CD</sc>57 identifies T cells with functional senescence before terminal differentiation and relative telomere shortening in patients with activated <sc>PI</sc>3 kinase delta syndrome. Immunology and Cell Biology, 2018, 96, 1060-1071.	2.3	29
20	VARICELLA ZOSTER VIRUS ASSOCIATED ACUTE ASEPTIC MENINGITIS WITHOUT EXANTHEM IN AN IMMUNOCOMPETENT 14-YEAR-OLD BOY. Pediatric Infectious Disease Journal, 2008, 27, 362-363.	2.0	27
21	Altered endotoxin responsiveness in healthy children with Down syndrome. BMC Immunology, 2018, 19, 31.	2.2	26
22	Novel Gain-of-Function Mutation in Stat1 Sumoylation Site Leads to CMC/CID Phenotype Responsive to Ruxolitinib. Journal of Clinical Immunology, 2019, 39, 776-785.	3.8	21
23	Interleukin-15 is associated with disease severity in viral bronchiolitis. European Respiratory Journal, 2016, 47, 212-222.	6.7	19
24	Epidemiology of borderline oxacillin-resistant <i>Staphylococcus aureus</i> in Pediatric cystic fibrosis. Pediatric Pulmonology, 2011, 46, 489-496.	2.0	18
25	Incomplete Kawasaki Disease Associated with Complicated Streptococcus pyogenes Pneumonia: A Case Report. Canadian Journal of Infectious Diseases and Medical Microbiology, 2012, 23, 137-139.	1.9	18
26	Altered Toll-Like Receptor Signalling in Children with Down Syndrome. Mediators of Inflammation, 2019, 2019, 1-13.	3.0	18
27	Emerging Role of the NLRP3 Inflammasome and Interleukin-1 $\beta$ in Neonates. Neonatology, 2020, 117, 545-554.	2.0	18
28	Asplenia in ATR-X syndrome: A second report. American Journal of Medical Genetics, Part A, 2005, 139A, 37-39.	1.2	15
29	Aortic Calcification in a Patient with a Gain-of-Function STAT1 Mutation. Journal of Clinical Immunology, 2018, 38, 468-470.	3.8	15
30	<sc>HIV</sc> virological suppression influences response to the <sc>AS</sc>03 adjuvanted monovalent pandemic influenza <sc>AH</sc>1<sc>N</sc>1 vaccine in <sc>HIV</sc>-infected children. Influenza and Other Respiratory Viruses, 2014, 8, 360-366.	3.4	12
31	Molecular identification of phaeohyphomycosis due to Alternaria infectoria in a patient with acute myeloid leukemia—a case report. Diagnostic Microbiology and Infectious Disease, 2010, 66, 318-321.	1.8	10
32	Idiopathic Suppurative Pylephlebitis: Interventional Radiological Diagnosis and Management. CardioVascular and Interventional Radiology, 2009, 32, 1304-1307.	2.0	9
33	Does vaccine dose predict response to the monovalent pandemic H1N1 influenza a vaccine in children with acute lymphoblastic leukemia? A single-centre study. Pediatric Blood and Cancer, 2013, 60, 1656-1661.	1.5	9
34	Autosomal Dominant Hyper IgE Syndrome — Treatment Strategies and Clinical Outcomes. Journal of Clinical Immunology, 2016, 36, 107-109.	3.8	9
35	T-replete HLA-matched grafts vs T-depleted HLA-mismatched grafts in inborn errors of immunity. Blood Advances, 2022, 6, 1319-1328.	5.2	8
36	Treatment-recalcitrant laryngeal sarcoidosis responsive to sirolimus. BMJ Case Reports, 2020, 13, e235372.	0.5	7

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37	Erythema induratum of Bazin and episcleritis in a 6 year old girl. Archives of Disease in Childhood, 2005, 90, 1132-1132.	1.9	6
38	Immunodeficiency in a Child with 22q11.2 Microduplication Syndrome. Journal of Clinical Immunology, 2016, 36, 418-419.	3.8	6
39	Topical cidofovir for the treatment of recalcitrant viral warts and molluscum contagiosum in Jacobsen syndrome. Pediatric Dermatology, 2020, 37, 1191-1192.	0.9	5
40	Developing integrated care in the context of rare chromosomal conditions: 22q11 Deletion Syndrome; A parent/clinician collaboration.. International Journal of Integrated Care, 2017, 17, 215.	0.2	5
41	Imported childhood malaria: the Dublin experience, 1999â€“2006. Irish Journal of Medical Science, 2009, 178, 329-332.	1.5	4
42	Haploidentical CD3 TCRÎ±Î² and CD19-depleted second stem cell transplant for steroid-resistant acute skin graft versus host disease. Journal of Allergy and Clinical Immunology, 2016, 138, 603-605.e1.	2.9	4
43	Severe combined immunodeficiency syndrome presenting with disseminated BCG infection. Archives of Disease in Childhood, 2015, 100, 891-891.	1.9	3
44	Double Trouble? CMC with a Mutation in both AIRE and STAT1. Journal of Clinical Immunology, 2018, 38, 635-637.	3.8	3
45	Persistent pruritic subcutaneous nodules at injection sites and other delayed type hypersensitivity reactions to aluminium adsorbed vaccines in Irish children: A case series. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 2692-2693.	1.5	3
46	Adenosine Deaminase Deficient SCID with Myocardial Hypertrophy. Journal of Clinical Immunology, 2021, 41, 1128-1130.	3.8	3
47	Viral Bronchiolitis is Associated With Altered Cytokine Gene Expression and Lymphocyte Activation Status. Pediatric Infectious Disease Journal, 2016, 35, e326-e338.	2.0	2
48	X-linked agammaglobulinaemia (XLA) presenting with neutropenia and Pseudomonasaeruginosa cellulitis. Archives of Disease in Childhood, 2016, 101, 1106-1106.	1.9	0
49	Acute Genital Swelling Heraldng C1 Esterase Inhibitor Deficiency in a Child. Pediatric Dermatology, 2017, 34, e277-e278.	0.9	0
50	Expanding the Spectrum of Post-malaria Neurologic Syndrome in the Pediatric Population. Pediatric Infectious Disease Journal, 2018, 37, 499-500.	2.0	0
51	P373â€“...You give me fever! â€“ the autoinflammatory clinic in an irish tertiary paediatric hospital. , 2019, , .		0
52	P376â€“...Parechovirus infection as the presenting illness in an infant with SCID. , 2019, , .		0
53	GP107â€“...Integrated care for 22q11 deletion syndrome in ireland â€“ meeting childrenâ€™s needs through enhanced care co-ordination. , 2019, , .		0
54	GP194â€“...Periodic fever-the irish PFAPA story so far. , 2019, , .		0

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55	OC29â€¦Respiratory syncytial virus hospital admission and outcome in ireland among infants born less than 32 weeks gestation in the era of changing palivizumab prescription guidelines. , 2019, , .		0
56	P362â€¦Delayed type hypersensitivity reactions to aluminium-adsorbed vaccines: a case series. , 2019, , .		0
57	P374â€¦Laryngeal sarcoidosis responsive to treatment with sirolimus: a first case report. , 2019, , .		0