

Emmanuelle Jouanguy

List of Publications by Year in descending order

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Version: 2024-02-01

127
papers

21,549
citations

14644

66
h-index

15249

126
g-index

135
all docs

135
docs citations

135
times ranked

22087
citing authors

#	ARTICLE	IF	CITATIONS
1	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,749
3	TLR3 Deficiency in Patients with Herpes Simplex Encephalitis. <i>Science</i> , 2007, 317, 1522-1527.	6.0	970
4	Interferon- β Receptor Deficiency in an Infant with Fatal Bacille Calmette-Guérin Infection. <i>New England Journal of Medicine</i> , 1996, 335, 1956-1962.	13.9	832
5	Impairment of Mycobacterial Immunity in Human Interleukin-12 Receptor Deficiency. <i>Science</i> , 1998, 280, 1432-1435.	6.0	787
6	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. <i>Journal of Experimental Medicine</i> , 2011, 208, 1635-1648.	4.2	739
7	Impaired response to interferon- β and lethal viral disease in human STAT1 deficiency. <i>Nature Genetics</i> , 2003, 33, 388-391.	9.4	720
8	Herpes Simplex Virus Encephalitis in Human UNC-93B Deficiency. <i>Science</i> , 2006, 314, 308-312.	6.0	674
9	The Jak-STAT signaling pathway is required but not sufficient for the antiviral response of drosophila. <i>Nature Immunology</i> , 2005, 6, 946-953.	7.0	569
10	A human IFNGR1 small deletion hotspot associated with dominant susceptibility to mycobacterial infection. <i>Nature Genetics</i> , 1999, 21, 370-378.	9.4	458
11	Human intracellular ISG15 prevents interferon- β over-amplification and auto-inflammation. <i>Nature</i> , 2015, 517, 89-93.	13.7	432
12	Inborn errors of IL-12/23- and IFN- γ -mediated immunity: molecular, cellular, and clinical features. <i>Seminars in Immunology</i> , 2006, 18, 347-361.	2.7	422
13	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	5.6	357
14	Human TRAF3 Adaptor Molecule Deficiency Leads to Impaired Toll-like Receptor 3 Response and Susceptibility to Herpes Simplex Encephalitis. <i>Immunity</i> , 2010, 33, 400-411.	6.6	304
15	IL-12 and IFN- γ in host defense against mycobacteria and salmonella in mice and men. <i>Current Opinion in Immunology</i> , 1999, 11, 346-351.	2.4	301
16	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. <i>Nature</i> , 2012, 491, 769-773.	13.7	288
17	Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. <i>Journal of Clinical Investigation</i> , 2012, 122, 821-832.	3.9	272
18	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN- α , IFN- β , and IFN- γ in host defense. <i>Immunological Reviews</i> , 2008, 226, 29-40.	2.8	271

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19	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. <i>Journal of Experimental Medicine</i> , 2010, 207, 2307-2312.	4.2	268
20	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	267
21	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. <i>Journal of Experimental Medicine</i> , 2006, 203, 1745-1759.	4.2	264
22	Herpes simplex virus encephalitis in a patient with complete TLR3 deficiency: TLR3 is otherwise redundant in protective immunity. <i>Journal of Experimental Medicine</i> , 2011, 208, 2083-2098.	4.2	262
23	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. <i>Journal of Clinical Investigation</i> , 2011, 121, 4889-4902.	3.9	254
24	Human TLR-7-, -8-, and -9-Mediated Induction of IFN- β and - γ Is IRAK-4 Dependent and Redundant for Protective Immunity to Viruses. <i>Immunity</i> , 2005, 23, 465-478.	6.6	245
25	Immunological conditions of children with BCG disseminated infection. <i>Lancet, The</i> , 1995, 346, 581.	6.3	219
26	Human genetic and immunological determinants of critical COVID-19 pneumonia. <i>Nature</i> , 2022, 603, 587-598.	13.7	216
27	IRAK-4- and MyD88-Dependent Pathways Are Essential for the Removal of Developing Autoreactive B Cells in Humans. <i>Immunity</i> , 2008, 29, 746-757.	6.6	201
28	Human Complete Stat-1 Deficiency Is Associated with Defective Type I and II IFN Responses In Vitro but Immunity to Some Low Virulence Viruses In Vivo. <i>Journal of Immunology</i> , 2006, 176, 5078-5083.	0.4	191
29	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , 2020, 181, 1194-1199.	13.5	185
30	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	185
31	A novel immunodeficiency associated with hypomorphic RAG1 mutations and CMV infection. <i>Journal of Clinical Investigation</i> , 2005, 115, 3291-3299.	3.9	177
32	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. <i>Journal of Experimental Medicine</i> , 2011, 208, 2305-2320.	4.2	175
33	Partial Interferon- β Receptor Signaling Chain Deficiency in a Patient with Bacille Calmette-Guérin and Mycobacterium abscessus Infection. <i>Journal of Infectious Diseases</i> , 2000, 181, 379-384.	1.9	171
34	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. <i>PLoS Genetics</i> , 2006, 2, e131.	1.5	171
35	Idiopathic Disseminated Bacillus Calmette-Guérin Infection: A French National Retrospective Study. <i>Pediatrics</i> , 1996, 98, 774-778.	1.0	170
36	A partial form of recessive STAT1 deficiency in humans. <i>Journal of Clinical Investigation</i> , 2009, 119, 1502-1514.	3.9	167

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37	Recurrent Staphylococcal Cellulitis and Subcutaneous Abscesses in a Child with Autoantibodies against IL-6. <i>Journal of Immunology</i> , 2008, 180, 647-654.	0.4	154
38	Human interferon-g-mediated immunity is a genetically controlled continuous trait that determines the outcome of mycobacterial invasion. <i>Immunological Reviews</i> , 2000, 178, 129-137.	2.8	153
39	In a novel form of IFN- β receptor 1 deficiency, cell surface receptors fail to bind IFN- β . <i>Journal of Clinical Investigation</i> , 2000, 105, 1429-1436.	3.9	149
40	Human Toll-like receptor-dependent induction of interferons in protective immunity to viruses. <i>Immunological Reviews</i> , 2007, 220, 225-236.	2.8	147
41	Genome-Wide Association Study Identifies Variants Associated With Progression of Liver Fibrosis From HCV Infection. <i>Gastroenterology</i> , 2012, 143, 1244-1252.e12.	0.6	142
42	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. <i>Journal of Clinical Investigation</i> , 2012, 122, 3239-3247.	3.9	134
43	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	130
44	Pherokine-2 and -3. Two Drosophila molecules related to pheromone/odor-binding proteins induced by viral and bacterial infections. <i>FEBS Journal</i> , 2003, 270, 3398-3407.	0.2	128
45	TLR3 deficiency in herpes simplex encephalitis. <i>Neurology</i> , 2014, 83, 1888-1897.	1.5	128
46	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. <i>Current Opinion in Immunology</i> , 2008, 20, 39-48.	2.4	127
47	Inherited IFNAR1 deficiency in otherwise healthy patients with adverse reaction to measles and yellow fever live vaccines. <i>Journal of Experimental Medicine</i> , 2019, 216, 2057-2070.	4.2	127
48	Inherited MST1 Deficiency Underlies Susceptibility to EV-HPV Infections. <i>PLoS ONE</i> , 2012, 7, e44010.	1.1	125
49	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. <i>Journal of Experimental Medicine</i> , 2013, 210, 1743-1759.	4.2	119
50	Dual T cell and B cell intrinsic deficiency in humans with biallelic <i>RLTPR</i> mutations. <i>Journal of Experimental Medicine</i> , 2016, 213, 2413-2435.	4.2	117
51	CORRELATION OF GRANULOMA STRUCTURE WITH CLINICAL OUTCOME DEFINES TWO TYPES OF IDIOPATHIC DISSEMINATED BCG INFECTION. , 1997, 181, 25-30.		116
52	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 1991-2006.	3.9	115
53	A Novel Primary Immunodeficiency with Specific Natural-Killer Cell Deficiency Maps to the Centromeric Region of Chromosome 8. <i>American Journal of Human Genetics</i> , 2006, 78, 721-727.	2.6	113
54	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. <i>Med</i> , 2020, 1, 14-20.	2.2	110

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55	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	3.3	110
56	Inborn errors of anti-viral interferon immunity in humans. Current Opinion in Virology, 2011, 1, 487-496.	2.6	109
57	SARS-CoV-2 induces human plasmacytoid dendritic cell diversification via UNC93B and IRAK4. Journal of Experimental Medicine, 2021, 218, .	4.2	107
58	Mendelian susceptibility to mycobacterial infection in man. Current Opinion in Immunology, 1998, 10, 413-417.	2.4	106
59	Genetic susceptibility to herpes simplex virus 1 encephalitis in mice and humans. Current Opinion in Allergy and Clinical Immunology, 2007, 7, 495-505.	1.1	101
60	SARS-CoV-2-related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
61	A Causative Relationship between Mutant IFNGR1 Alleles and Impaired Cellular Response to IFN γ in a Compound Heterozygous Child. American Journal of Human Genetics, 1998, 62, 723-727.	2.6	97
62	The human CIB1-EVER1-EVER2 complex governs keratinocyte-intrinsic immunity to β 2-papillomaviruses. Journal of Experimental Medicine, 2018, 215, 2289-2310.	4.2	92
63	Homozygous <i>NLRP1</i> gain-of-function mutation in siblings with a syndromic form of recurrent respiratory papillomatosis. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 19055-19063.	3.3	92
64	Age-Dependent Mendelian Predisposition to Herpes Simplex Virus Type 1 Encephalitis in Childhood. Journal of Pediatrics, 2010, 157, 623-629.e1.	0.9	85
65	Compound Heterozygous CORO1A Mutations in Siblings with a Mucocutaneous-Immunodeficiency Syndrome of Epidermodysplasia Verruciformis-HPV, Molluscum Contagiosum and Granulomatous Tuberculoid Leprosy. Journal of Clinical Immunology, 2014, 34, 871-890.	2.0	78
66	Osteopontin Expression Correlates with Clinical Outcome in Patients with Mycobacterial Infection. American Journal of Pathology, 2000, 157, 37-42.	1.9	73
67	Infections in IFNGR-1-Deficient Children. Journal of Interferon and Cytokine Research, 1997, 17, 583-587.	0.5	71
68	Inherited IL-18BP deficiency in human fulminant viral hepatitis. Journal of Experimental Medicine, 2019, 216, 1777-1790.	4.2	70
69	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69
70	NEMO is a key component of NF- κ B and IRF-3-dependent TLR3-mediated immunity to herpes simplex virus. Journal of Allergy and Clinical Immunology, 2011, 128, 610-617.e4.	1.5	66
71	Primary immunodeficiencies of protective immunity to primary infections. Clinical Immunology, 2010, 135, 204-209.	1.4	65
72	TLR3 controls constitutive IFN- β antiviral immunity in human fibroblasts and cortical neurons. Journal of Clinical Investigation, 2021, 131, .	3.9	64

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73	Human NLRP1 is a sensor of pathogenic coronavirus 3CL proteases in lung epithelial cells. <i>Molecular Cell</i> , 2022, 82, 2385-2400.e9.	4.5	61
74	Human inborn errors of immunity to herpes viruses. <i>Current Opinion in Immunology</i> , 2020, 62, 106-122.	2.4	60
75	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	59
76	Human primary immunodeficiencies of type I interferons. <i>Biochimie</i> , 2007, 89, 878-883.	1.3	57
77	A role for interleukin-12/23 in the maturation of human natural killer and CD56+ T cells in vivo. <i>Blood</i> , 2008, 111, 5008-5016.	0.6	57
78	TIM3+ TRBV11-2 T cells and IFN γ signature in patrolling monocytes and CD16+ NK cells delineate MIS-C. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	57
79	Epidermodysplasia Verruciformis: Inborn Errors of Immunity to Human Beta-Papillomaviruses. <i>Frontiers in Microbiology</i> , 2018, 9, 1222.	1.5	56
80	Whole-exome sequencing to analyze population structure, parental inbreeding, and familial linkage. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 6713-6718.	3.3	53
81	Humans with inherited T β cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. <i>Cell</i> , 2021, 184, 3812-3828.e30.	13.5	53
82	Recurrent <i>Mycobacterium avium</i> Osteomyelitis Associated With a Novel Dominant Interferon Gamma Receptor Mutation. <i>Pediatrics</i> , 2001, 107, e47-e47.	1.0	51
83	Kaposi Sarcoma of Childhood: Inborn or Acquired Immunodeficiency to Oncogenic HHV-8. <i>Pediatric Blood and Cancer</i> , 2016, 63, 392-397.	0.8	50
84	Life-Threatening Infections Due to Live-Attenuated Vaccines: Early Manifestations of Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2019, 39, 376-390.	2.0	50
85	Type I interferons and SARS-CoV-2: from cells to organisms. <i>Current Opinion in Immunology</i> , 2022, 74, 172-182.	2.4	49
86	Classic Kaposi Sarcoma in 3 Unrelated Turkish Children Born to Consanguineous Kindreds. <i>Pediatrics</i> , 2010, 125, e704-e708.	1.0	47
87	Severe COVID-19 in the young and healthy: monogenic inborn errors of immunity?. <i>Nature Reviews Immunology</i> , 2020, 20, 455-456.	10.6	47
88	Autoantibodies Neutralizing Type I Interferons in 20% of COVID-19 Deaths in a French Hospital. <i>Journal of Clinical Immunology</i> , 2022, 42, 459-470.	2.0	46
89	Surface Expression of the IFN- γ R2 Chain Is Regulated by Intracellular Trafficking in Human T Lymphocytes. <i>Journal of Immunology</i> , 2000, 164, 201-207.	0.4	44
90	A Novel Developmental and Immunodeficiency Syndrome Associated With Intrauterine Growth Retardation and a Lack of Natural Killer Cells. <i>Pediatrics</i> , 2004, 113, 136-141.	1.0	44

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91	Human inborn errors of immunity to infection affecting cells other than leukocytes: from the immune system to the whole organism. <i>Current Opinion in Immunology</i> , 2019, 59, 88-100.	2.4	44
92	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. <i>Nature Immunology</i> , 2022, 23, 159-164.	7.0	41
93	Distinct antibody repertoires against endemic human coronaviruses in children and adults. <i>JCI Insight</i> , 2021, 6, .	2.3	40
94	EVER2 Deficiency is Associated with Mild T-cell Abnormalities. <i>Journal of Clinical Immunology</i> , 2013, 33, 14-21.	2.0	38
95	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. <i>New England Journal of Medicine</i> , 2020, 382, 437-445.	13.9	38
96	A homozygous mutation of RTEL1 in a child presenting with an apparently isolated natural killer cell deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1113-1114.	1.5	37
97	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. <i>Science Immunology</i> , 2023, 8, .	5.6	35
98	Interferon- β receptor deficiency mimicking Langerhansâ€™ cell histiocytosis. <i>Journal of Pediatrics</i> , 2001, 139, 600-603.	0.9	33
99	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	33
100	Biochemically deleterious human <i>NFKB1</i> variants underlie an autosomal dominant form of common variable immunodeficiency. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	32
101	Familial NK Cell Deficiency Associated with Impaired IL-2- and IL-15-Dependent Survival of Lymphocytes. <i>Journal of Immunology</i> , 2006, 177, 8835-8843.	0.4	31
102	Recurrent elevated liver transaminases and acute liver failure in two siblings with novel bi-allelic mutations of NBAS. <i>European Journal of Medical Genetics</i> , 2017, 60, 426-432.	0.7	31
103	Nonpathogenic Common Variants of IFNGR1 and IFNGR2 in Association with Total Serum IgE Levels. <i>Biochemical and Biophysical Research Communications</i> , 1999, 263, 425-429.	1.0	30
104	A loss-of-function <i>IFNAR1</i> allele in Polynesia underlies severe viral diseases in homozygotes. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	28
105	Inborn errors of the development of human natural killer cells. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2013, 13, 589-595.	1.1	24
106	Monoclonal antibody-mediated neutralization of SARS-CoV-2 in an IRF9-deficient child. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	24
107	Requirement for both IL-12 and IFN- β signaling pathways in optimal IFN- β production by human T cells. <i>European Journal of Immunology</i> , 2002, 32, 693.	1.6	23
108	Inherited Interleukin 2â€™-Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. <i>Clinical Infectious Diseases</i> , 2019, 68, 1938-1941.	2.9	22

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109	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	21
110	Interferon λ receptor 2 gene variants are associated with liver fibrosis in patients with chronic hepatitis C infection. <i>Gut</i> , 2010, 59, 1120-1126.	6.1	19
111	A CIB1 Splice-Site Founder Mutation in Families with Atypical Epidermodysplasia Verruciformis. <i>Journal of Investigative Dermatology</i> , 2019, 139, 1195-1198.	0.3	19
112	Epidermodysplasia Verruciformis: Genetic Heterogeneity and EVER1 and EVER2 Mutations Revealed by Genome-Wide Analysis. <i>Journal of Investigative Dermatology</i> , 2019, 139, 241-244.	0.3	19
113	Insufficient type I IFN immunity underlies life-threatening COVID-19 pneumonia. <i>Comptes Rendus - Biologies</i> , 2021, 344, 19-25.	0.1	16
114	Human genetic and immunological dissection of papillomavirus-driven diseases: new insights into their pathogenesis. <i>Current Opinion in Virology</i> , 2021, 51, 9-15.	2.6	16
115	Human genetic basis of fulminant viral hepatitis. <i>Human Genetics</i> , 2020, 139, 877-884.	1.8	10
116	Human inborn errors of immunity to oncogenic viruses. <i>Current Opinion in Immunology</i> , 2021, 72, 277-285.	2.4	10
117	Recalcitrant Warts, Epidermodysplasia Verruciformis, and the Tree-Man Syndrome: Phenotypic Spectrum of Cutaneous Human Papillomavirus Infections at the Intersection of Genetic Variability of Viral and Human Genomes. <i>Journal of Investigative Dermatology</i> , 2022, 142, 1265-1269.	0.3	10
118	Diagnosis of APS-1 in Two Siblings Following Life-Threatening COVID-19 Pneumonia. <i>Journal of Clinical Immunology</i> , 2022, 42, 749-752.	2.0	10
119	Dominant negative CARD11 mutations: Beyond atopy. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1345-1347.	1.5	8
120	Identification of an Endoglin Variant Associated With HCV-Related Liver Fibrosis Progression by Next-Generation Sequencing. <i>Frontiers in Genetics</i> , 2019, 10, 1024.	1.1	6
121	Whole-transcriptome sequencing-based concomitant detection of viral and human genetic determinants of cutaneous lesions. <i>JCI Insight</i> , 2022, 7, .	2.3	6
122	A virus finds its natural killer. <i>Nature Genetics</i> , 2001, 28, 7-9.	9.4	5
123	Candidate Predisposition Variants in Kaposi Sarcoma as Detected by Whole-Genome Sequencing. <i>Open Forum Infectious Diseases</i> , 2019, 6, ofz337.	0.4	5
124	Recalcitrant Cutaneous Warts in a Family with Inherited ICOS Deficiency. <i>Journal of Investigative Dermatology</i> , 2022, 142, 2435-2445.	0.3	4
125	Title is missing!. <i>Nature Genetics</i> , 2001, 28, 7-9.	9.4	2
126	Inherited disorders of IFN- λ ³ , IFN- λ ^{1/2/1} , and NF- κ B-mediated immunity. , 2013, , 454-464.		1

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127	Immunodeficiencies at the Interface of Innate and Adaptive Immunity. , 2019, , 509-522.e1.		0