Emmanuelle Jouanguy

List of Publications by Year
in descending order

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127 papers	21,549 citations	14614 66 h-index	15218 126 g-index
135 all docs	135 docs citations	135 times ranked	22087 citing authors

#	Article	IF	Citations
1	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. Science Immunology, 2023, 8, .	5.6	35
2	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	7.0	41
3	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. Journal of Investigative Dermatology, 2022, 142, 1265-1269.	0.3	10
4	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
5	Autoantibodies Neutralizing Type I Interferons in 20% of COVID-19 Deaths in a French Hospital. Journal of Clinical Immunology, 2022, 42, 459-470.	2.0	46
6	Type I interferons and SARS-CoV-2: from cells to organisms. Current Opinion in Immunology, 2022, 74, 172-182.	2.4	49
7	TIM3+ TRBV11-2 T cells and IFNÎ³ signature in patrolling monocytes and CD16+ NK cells delineate MIS-C. Journal of Experimental Medicine, 2022, 219, .	4.2	57
8	Whole-transcriptome sequencingâ€“based concomitant detection of viral and human genetic determinants of cutaneous lesions. JCI Insight, 2022, 7, .	2.3	6
9	Diagnosis of APS-1 in Two Siblings Following Life-Threatening COVID-19 Pneumonia. Journal of Clinical Immunology, 2022, 42, 749-752.	2.0	10
10	Recalcitrant Cutaneous Warts in a Family with Inherited ICOS Deficiency. Journal of Investigative Dermatology, 2022, 142, 2435-2445.	0.3	4
11	A loss-of-function IFNAR1 allele in Polynesia underlies severe viral diseases in homozygotes. Journal of Experimental Medicine, 2022, 219, .	4.2	28
12	Human NLRP1 is a sensor of pathogenic coronavirus 3CL proteases in lung epithelial cells. Molecular Cell, 2022, 82, 2385-2400.e9.	4.5	61
13	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
14	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
15	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
16	TLR3 controls constitutive IFN-Î² antiviral immunity in human fibroblasts and cortical neurons. Journal of Clinical Investigation, 2021, 131, .	3.9	64
17	SARS-CoV-2 induces human plasmacytoid predendritic cell diversification via UNC93B and IRAK4. Journal of Experimental Medicine, 2021, 218, .	4.2	107
18	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	33

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19	Distinct antibody repertoires against endemic human coronaviruses in children and adults. JCI Insight, 2021, 6, .	2.3	40
20	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	4.2	130
21	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
22	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	4.2	185
23	Insufficient type I IFN immunity underlies life-threatening COVID-19 pneumonia. Comptes Rendus - Biologies, 2021, 344, 19-25.	0.1	16
24	Humans with inherited TÂcell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. Cell, 2021, 184, 3812-3828.e30.	13.5	53
25	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	5.6	357
26	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
27	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
28	Human inborn errors of immunity to oncogenic viruses. Current Opinion in Immunology, 2021, 72, 277-285.	2.4	10
29	Human genetic and immunological dissection of papillomavirus-driven diseases: new insights into their pathogenesis. Current Opinion in Virology, 2021, 51, 9-15.	2.6	16
30	Monoclonal antibody-mediated neutralization of SARS-CoV-2 in an IRF9-deficient child. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	24
31	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. Med, 2020, 1, 14-20.	2.2	110
32	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
33	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
34	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
35	Severe COVID-19 in the young and healthy: monogenic inborn errors of immunity?. Nature Reviews Immunology, 2020, 20, 455-456.	10.6	47
36	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69

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37	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. New England Journal of Medicine, 2020, 382, 437-445.	13.9	38
38	Human genetic basis of fulminant viral hepatitis. Human Genetics, 2020, 139, 877-884.	1.8	10
39	Human inborn errors of immunity to herpes viruses. Current Opinion in Immunology, 2020, 62, 106-122.	2.4	60
40	Immunodeficiencies at the Interface of Innate and Adaptive Immunity. , 2019, , 509-522.e1.		0
41	Inherited IFNAR1 deficiency in otherwise healthy patients with adverse reaction to measles and yellow fever live vaccines. Journal of Experimental Medicine, 2019, 216, 2057-2070.	4.2	127
42	Candidate Predisposition Variants in Kaposi Sarcoma as Detected by Whole-Genome Sequencing. Open Forum Infectious Diseases, 2019, 6, ofz337.	0.4	5
43	Dominant negative CARD11 mutations: Beyond atopy. Journal of Allergy and Clinical Immunology, 2019, 143, 1345-1347.	1.5	8
44	Inherited IL-18BP deficiency in human fulminant viral hepatitis. Journal of Experimental Medicine, 2019, 216, 1777-1790.	4.2	70
45	Life-Threatening Infections Due to Live-Attenuated Vaccines: Early Manifestations of Inborn Errors of Immunity. Journal of Clinical Immunology, 2019, 39, 376-390.	2.0	50
46	Human inborn errors of immunity to infection affecting cells other than leukocytes: from the immune system to the whole organism. Current Opinion in Immunology, 2019, 59, 88-100.	2.4	44
47	A CIB1 Splice-Site Founder Mutation in Families withÂTypical Epidermodysplasia Verruciformis. Journal of Investigative Dermatology, 2019, 139, 1195-1198.	0.3	19
48	Inherited Interleukin 2â€“Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. Clinical Infectious Diseases, 2019, 68, 1938-1941.	2.9	22
49	Identification of an Endoglin Variant Associated With HCV-Related Liver Fibrosis Progression by Next-Generation Sequencing. Frontiers in Genetics, 2019, 10, 1024.	1.1	6
50	Homozygous NLRP1 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
51	Epidermodysplasia Verruciformis: Genetic Heterogeneity and EVER1 and EVER2 Mutations Revealed by Genome-Wide Analysis. Journal of Investigative Dermatology, 2019, 139, 241-244.	0.3	19
52	The human CIB1â€“EVER1â€“EVER2 complex governs keratinocyte-intrinsic immunity to Î²-papillomaviruses. Journal of Experimental Medicine, 2018, 215, 2289-2310.	4.2	92
53	Epidermodysplasia Verruciformis: Inborn Errors of Immunity to Human Beta-Papillomaviruses. Frontiers in Microbiology, 2018, 9, 1222.	1.5	56
54	Recurrent elevated liver transaminases and acute liver failure in two siblings with novel bi-allelic mutations of NBAS. European Journal of Medical Genetics, 2017, 60, 426-432.	0.7	31

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55	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. Journal of Clinical Investigation, 2017, 127, 1991-2006.	3.9	115
56	Kaposi Sarcoma of Childhood: Inborn or Acquired Immunodeficiency to Oncogenic HHVâ€8. Pediatric Blood and Cancer, 2016, 63, 392-397.	0.8	50
57	Dual T cellâ€“ and B cellâ€“intrinsic deficiency in humans with biallelic RLTPR mutations. Journal of Experimental Medicine, 2016, 213, 2413-2435.	4.2	117
58	Whole-exome sequencing to analyze population structure, parental inbreeding, and familial linkage. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 6713-6718.	3.3	53
59	A homozygous mutation of RTEL1 in a child presenting with an apparently isolated natural killer cell deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 1113-1114.	1.5	37
60	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
61	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
62	TLR3 deficiency in herpes simplex encephalitis. Neurology, 2014, 83, 1888-1897.	1.5	128
63	EVER2 Deficiency is Associated with Mild T-cell Abnormalities. Journal of Clinical Immunology, 2013, 33, 14-21.	2.0	38
64	Inborn errors of the development of human natural killer cells. Current Opinion in Allergy and Clinical Immunology, 2013, 13, 589-595.	1.1	24
65	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. Journal of Experimental Medicine, 2013, 210, 1743-1759.	4.2	119
66	Inherited disorders of IFN-Î³-, IFN-Î±/Î²/Î»-, and NF-ÎºB-mediated immunity. , 2013, , 454-464.		1
67	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. Nature, 2012, 491, 769-773.	13.7	288
68	Genome-Wide Association Study Identifies Variants Associated With Progression of Liver Fibrosis From HCV Infection. Gastroenterology, 2012, 143, 1244-1252.e12.	0.6	142
69	Inherited MST1 Deficiency Underlies Susceptibility to EV-HPV Infections. PLoS ONE, 2012, 7, e44010.	1.1	125
70	Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. Journal of Clinical Investigation, 2012, 122, 821-832.	3.9	272
71	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. Journal of Clinical Investigation, 2012, 122, 3239-3247.	3.9	134
72	NEMO is a key component of NF-ÎºBâ€“ and IRF-3â€“dependent TLR3-mediated immunity to herpes simplex virus. Journal of Allergy and Clinical Immunology, 2011, 128, 610-617.e4.	1.5	66

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73	Inborn errors of anti-viral interferon immunity in humans. Current Opinion in Virology, 2011, 1, 487-496.	2.6	109
74	Herpes simplex virus encephalitis in a patient with complete TLR3 deficiency: TLR3 is otherwise redundant in protective immunity. Journal of Experimental Medicine, 2011, 208, 2083-2098.	4.2	262
75	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. Journal of Experimental Medicine, 2011, 208, 2305-2320.	4.2	175
76	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2011, 208, 1635-1648.	4.2	739
77	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. Journal of Clinical Investigation, 2011, 121, 4889-4902.	3.9	254
78	Age-Dependent Mendelian Predisposition to Herpes Simplex Virus Type 1 Encephalitis in Childhood. Journal of Pediatrics, 2010, 157, 623-629.e1.	0.9	85
79	Primary immunodeficiencies of protective immunity to primary infections. Clinical Immunology, 2010, 135, 204-209.	1.4	65
80	Human TRAF3 Adaptor Molecule Deficiency Leads to Impaired Toll-like Receptor 3 Response and Susceptibility to Herpes Simplex Encephalitis. Immunity, 2010, 33, 400-411.	6.6	304
81	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. Journal of Experimental Medicine, 2010, 207, 2307-2312.	4.2	268
82	Classic Kaposi Sarcoma in 3 Unrelated Turkish Children Born to Consanguineous Kindreds. Pediatrics, 2010, 125, e704-e708.	1.0	47
83	Interferon Â receptor 2 gene variants are associated with liver fibrosis in patients with chronic hepatitis C infection. Gut, 2010, 59, 1120-1126.	6.1	19
84	A partial form of recessive STAT1 deficiency in humans. Journal of Clinical Investigation, 2009, 119, 1502-1514.	3.9	167
85	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. Current Opinion in Immunology, 2008, 20, 39-48.	2.4	127
86	Inborn errors of interferon (IFN)â€mediated immunity in humans: insights into the respective roles of IFNâ€Î±/Î², IFNâ€Î³, and IFNâ€Î» in host defense. Immunological Reviews, 2008, 226, 29-40.	2.8	271
87	IRAK-4- and MyD88-Dependent Pathways Are Essential for the Removal of Developing Autoreactive B Cells in Humans. Immunity, 2008, 29, 746-757.	6.6	201
88	Recurrent Staphylococcal Cellulitis and Subcutaneous Abscesses in a Child with Autoantibodies against IL-6. Journal of Immunology, 2008, 180, 647-654.	0.4	154
89	A role for interleukin-12/23 in the maturation of human natural killer and CD56+ T cells in vivo. Blood, 2008, 111, 5008-5016.	0.6	57
90	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

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91	Human primary immunodeficiencies of type I interferons. Biochimie, 2007, 89, 878-883.	1.3	57
92	TLR3 Deficiency in Patients with Herpes Simplex Encephalitis. Science, 2007, 317, 1522-1527.	6.0	970
93	Human Tollâ€like receptorâ€dependent induction of interferons in protective immunity to viruses. Immunological Reviews, 2007, 220, 225-236.	2.8	147
94	A Novel Primary Immunodeficiency with Specific Natural-Killer Cell Deficiency Maps to the Centromeric Region of Chromosome 8. American Journal of Human Genetics, 2006, 78, 721-727.	2.6	113
95	Herpes Simplex Virus Encephalitis in Human UNC-93B Deficiency. Science, 2006, 314, 308-312.	6.0	674
96	Inborn errors of IL-12/23- and IFN-Î³-mediated immunity: molecular, cellular, and clinical features. Seminars in Immunology, 2006, 18, 347-361.	2.7	422
97	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. PLoS Genetics, 2006, 2, e131.	1.5	171
98	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. Journal of Experimental Medicine, 2006, 203, 1745-1759.	4.2	264
99	Familial NK Cell Deficiency Associated with Impaired IL-2- and IL-15-Dependent Survival of Lymphocytes. Journal of Immunology, 2006, 177, 8835-8843.	0.4	31
100	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. Journal of Immunology, 2006, 176, 5078-5083.	0.4	191
101	The Jak-STAT signaling pathway is required but not sufficient for the antiviral response of drosophila. Nature Immunology, 2005, 6, 946-953.	7.0	569
102	Human TLR-7-, -8-, and -9-Mediated Induction of IFN-Î±/Î² and -Î» Is IRAK-4 Dependent and Redundant for Protective Immunity to Viruses. Immunity, 2005, 23, 465-478.	6.6	245
103	A novel immunodeficiency associated with hypomorphic RAG1 mutations and CMV infection. Journal of Clinical Investigation, 2005, 115, 3291-3299.	3.9	177
104	A Novel Developmental and Immunodeficiency Syndrome Associated With Intrauterine Growth Retardation and a Lack of Natural Killer Cells. Pediatrics, 2004, 113, 136-141.	1.0	44
105	Pherokine-2 and -3. Two Drosophila molecules related to pheromone/odor-binding proteins induced by viral and bacterial infections. FEBS Journal, 2003, 270, 3398-3407.	0.2	128
106	Impaired response to interferon-Î±/Î² and lethal viral disease in human STAT1 deficiency. Nature Genetics, 2003, 33, 388-391.	9.4	720
107	Requirement for both IL-12 and IFN-Î³ signaling pathways in optimal IFN-Î³ production by human T cells. European Journal of Immunology, 2002, 32, 693.	1.6	23
108	Interferon-Î³ receptor deficiency mimicking Langerhansâ€™ cell histiocytosis. Journal of Pediatrics, 2001, 139, 600-603.	0.9	33

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109	RecurrentMycobacterium aviumOsteomyelitis Associated With a Novel Dominant Interferon Gamma Receptor Mutation. Pediatrics, 2001, 107, e47-e47.	1.0	51
110	A virus finds its natural killer. Nature Genetics, 2001, 28, 7-9.	9.4	5
111	Title is missing!. Nature Genetics, 2001, 28, 7-9.	9.4	2
112	Human interferon-g-mediated immunity is a genetically controlled continuous trait that determines the outcome of mycobacterial invasion. Immunological Reviews, 2000, 178, 129-137.	2.8	153
113	Surface Expression of the IFN-Î³R2 Chain Is Regulated by Intracellular Trafficking in Human T Lymphocytes. Journal of Immunology, 2000, 164, 201-207.	0.4	44
114	Partial Interferonâ€Î³ Receptor Signaling Chain Deficiency in a Patient with Bacille Calmetteâ€GuÃ©rin andMycobacterium abscessusInfection. Journal of Infectious Diseases, 2000, 181, 379-384.	1.9	171
115	Osteopontin Expression Correlates with Clinical Outcome in Patients with Mycobacterial Infection. American Journal of Pathology, 2000, 157, 37-42.	1.9	73
116	In a novel form of IFN-Î³ receptor 1 deficiency, cell surface receptors fail to bind IFN-Î³. Journal of Clinical Investigation, 2000, 105, 1429-1436.	3.9	149
117	IL-12 and IFN-Î³ in host defense against mycobacteria and salmonella in mice and men. Current Opinion in Immunology, 1999, 11, 346-351.	2.4	301
118	A human IFNGR1 small deletion hotspot associated with dominant susceptibility to mycobacterial infection. Nature Genetics, 1999, 21, 370-378.	9.4	458
119	Nonpathogenic Common Variants of IFNGR1 and IFNGR2 in Association with Total Serum IgE Levels. Biochemical and Biophysical Research Communications, 1999, 263, 425-429.	1.0	30
120	Mendelian susceptibility to mycobacterial infection in man. Current Opinion in Immunology, 1998, 10, 413-417.	2.4	106
121	A Causative Relationship between Mutant IFNgR1 Alleles and Impaired Cellular Response to IFNÎ³ in a Compound Heterozygous Child. American Journal of Human Genetics, 1998, 62, 723-727.	2.6	97
122	Impairment of Mycobacterial Immunity in Human Interleukin-12 Receptor Deficiency. Science, 1998, 280, 1432-1435.	6.0	787
123	Infections in IFNGR-1-Deficient Children. Journal of Interferon and Cytokine Research, 1997, 17, 583-587.	0.5	71
124	CORRELATION OF GRANULOMA STRUCTURE WITH CLINICAL OUTCOME DEFINES TWO TYPES OF IDIOPATHIC DISSEMINATED BCG INFECTION. , 1997, 181, 25-30.		116
125	Interferon-Î³ â€“Receptor Deficiency in an Infant with Fatal Bacille Calmetteâ€“GuÃ©rin Infection. New England Journal of Medicine, 1996, 335, 1956-1962.	13.9	832
126	Idiopathic Disseminated Bacillus Calmette-Guèrin Infection: A French National Retrospective Study. Pediatrics, 1996, 98, 774-778.	1.0	170

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127	Immunological conditions of children with BCG disseminated infection. Lancet, The, 1995, 346, 581.	6.3	219