Capucine Picard

List of Publications by Citations

Source: https://exaly.com/author-pdf/1815315/capucine-picard-publications-by-citations.pdf
Version: 2024-04-17

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.
The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

360 papers	34,424 citations	105 h-index	176 g-index
384 ext. papers	40,897 ext. citations	9.5 avg, IF	6.45 L-index

#	Paper	IF	Citations
360	Chronic mucocutaneous candidiasis in humans with inborn errors of interleukin-17 immunity. *Science*, **2011**, 332, 65-8	33.3	1309
359	Human CD14dim monocytes patrol and sense nucleic acids and viruses via TLR7 and TLR8 receptors. *Immunity*, **2010**, 33, 375-86	32.3	862
358	TLR3 deficiency in patients with herpes simplex encephalitis. *Science*, **2007**, 317, 1522-7	33.3	842
357	Pyogenic bacterial infections in humans with IRAK-4 deficiency. *Science*, **2003**, 299, 2076-9	33.3	737
356	Pyogenic bacterial infections in humans with MyD88 deficiency. *Science*, **2008**, 321, 691-6	33.3	608
355	Herpes simplex virus encephalitis in human UNC-93B deficiency. *Science*, **2006**, 314, 308-12	33.3	601
354	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2011**, 208, 1635-48	16.6	599
353	Autoantibodies against IL-17A, IL-17F, and IL-22 in patients with chronic mucocutaneous candidiasis and autoimmune polyendocrine syndrome type I. *Journal of Experimental Medicine*, **2010**, 207, 291-7	16.6	556
352	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. *Journal of Clinical Immunology*, **2018**, 38, 96-128	5.7	510
351	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. *Journal of Clinical Immunology*, **2020**, 40, 24-64	5.7	497
350	Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015. *Journal of Clinical Immunology*, **2015**, 35, 696-726	5.7	478
349	Efficacy of gene therapy for X-linked severe combined immunodeficiency. *New England Journal of Medicine*, **2010**, 363, 355-64	59.2	471
348	Phosphoinositide 3-kinase gene mutation predisposes to respiratory infection and airway damage. *Science*, **2013**, 342, 866-71	33.3	424
347	STIM1 mutation associated with a syndrome of immunodeficiency and autoimmunity. *New England Journal of Medicine*, **2009**, 360, 1971-80	59.2	395
346	Inborn errors of IL-12/23- and IFN-gamma-mediated immunity: molecular, cellular, and clinical features. *Seminars in Immunology*, **2006**, 18, 347-61	10.7	366
345	Mutations in STAT3 and IL12RB1 impair the development of human IL-17-producing T cells. *Journal of Experimental Medicine*, **2008**, 205, 1543-50	16.6	361
344	Clinical features of dominant and recessive interferon gamma receptor 1 deficiencies. *Lancet, The*, **2004**, 364, 2113-21	40	359

343	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2018**, 38, 129-143	5.7	345
342	Selective predisposition to bacterial infections in IRAK-4-deficient children: IRAK-4-dependent TLRs are otherwise redundant in protective immunity. *Journal of Experimental Medicine*, **2007**, 204, 2407-22	16.6	329
341	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. *Nature Immunology*, **2012**, 13, 1178-86	19.1	320
340	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. *Blood*, **2016**, 127, 3154-64	2.2	314
339	Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency. *Frontiers in Immunology*, **2014**, 5, 162	8.4	309
338	Clinical features and outcome of patients with IRAK-4 and MyD88 deficiency. *Medicine (United States)*, **2010**, 89, 403-425	1.8	297
337	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. *Journal of Clinical Investigation*, **2014**, 124, 5516-20	15.9	294
336	IMMUNODEFICIENCIES. Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. *Science*, **2015**, 349, 606-613	33.3	291
335	Infectious diseases in patients with IRAK-4, MyD88, NEMO, or IB deficiency. *Clinical Microbiology Reviews*, **2011**, 24, 490-7	34	286
334	Infectious disease. Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. *Science*, **2015**, 348, 448-53	33.3	285
333	A modified retrovirus vector for X-linked severe combined immunodeficiency. *New England Journal of Medicine*, **2014**, 371, 1407-17	59.2	278
332	Revisiting human IL-12R deficiency: a survey of 141 patients from 30 countries. *Medicine (United States)*, **2010**, 89, 381-402	1.8	277
331	Clinical similarities and differences of patients with X-linked lymphoproliferative syndrome type 1 (XLP-1/SAP deficiency) versus type 2 (XLP-2/XIAP deficiency). *Blood*, **2011**, 117, 1522-9	2.2	272
330	Munc18-2 deficiency causes familial hemophagocytic lymphohistiocytosis type 5 and impairs cytotoxic granule exocytosis in patient NK cells. *Journal of Clinical Investigation*, **2009**, 119, 3765-73	15.9	267
329	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification. *Journal of Clinical Immunology*, **2020**, 40, 66-81	5.7	267
328	Human TRAF3 adaptor molecule deficiency leads to impaired Toll-like receptor 3 response and susceptibility to herpes simplex encephalitis. *Immunity*, **2010**, 33, 400-11	32.3	262
327	Clinical spectrum and features of activated phosphoinositide 3-kinase syndrome: A large patient cohort study. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 597-606.e4	11.5	251
326	The transmembrane activator TACI triggers immunoglobulin class switching by activating B cells through the adaptor MyD88. *Nature Immunology*, **2010**, 11, 836-45	19.1	251

325	Outcomes following gene therapy in patients with severe Wiskott-Aldrich syndrome. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1550-63	27.4	245
324	Deep dermatophytosis and inherited CARD9 deficiency. *New England Journal of Medicine*, **2013**, 369, 1704-1714	59.2	245
323	ORAI1 deficiency and lack of store-operated Ca2+ entry cause immunodeficiency, myopathy, and ectodermal dysplasia. *Journal of Allergy and Clinical Immunology*, **2009**, 124, 1311-1318.e7	11.5	238
322	Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. *Blood*, **2011**, 118, 1675-84	2.2	236
321	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. *Journal of Experimental Medicine*, **2010**, 207, 2307-12	16.6	236
320	Inherited interleukin-12 deficiency: IL12B genotype and clinical phenotype of 13 patients from six kindreds. *American Journal of Human Genetics*, **2002**, 70, 336-48	11	229
319	Human TLR-7-, -8-, and -9-mediated induction of IFN-alpha/beta and -lambda Is IRAK-4 dependent and redundant for protective immunity to viruses. *Immunity*, **2005**, 23, 465-78	32.3	228
318	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. *Journal of Clinical Investigation*, **2011**, 121, 4889-902	15.9	227
317	Herpes simplex virus encephalitis in a patient with complete TLR3 deficiency: TLR3 is otherwise redundant in protective immunity. *Journal of Experimental Medicine*, **2011**, 208, 2083-98	16.6	223
316	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. *Journal of Experimental Medicine*, **2006**, 203, 1745-59	16.6	222
315	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN-alpha/beta, IFN-gamma, and IFN-lambda in host defense. *Immunological Reviews*, **2008**, 226, 29-40	11.3	220
314	MST1 mutations in autosomal recessive primary immunodeficiency characterized by defective naive T-cell survival. *Blood*, **2012**, 119, 3458-68	2.2	210
313	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. *Journal of Experimental Medicine*, **2015**, 212, 1641-62	16.6	209
312	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. *Current Opinion in Allergy and Clinical Immunology*, **2012**, 12, 616-22	3.3	208
311	An ACT1 mutation selectively abolishes interleukin-17 responses in humans with chronic mucocutaneous candidiasis. *Immunity*, **2013**, 39, 676-86	32.3	204
310	Autosomal dominant STAT3 deficiency and hyper-IgE syndrome: molecular, cellular, and clinical features from a French national survey. *Medicine (United States)*, **2012**, 91, e1-e19	1.8	203
309	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. *Nature Immunology*, **2011**, 12, 213-21	19.1	202
308	Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. *Journal of Clinical Investigation*, **2012**, 122, 821-32	15.9	201

307	Hematopoietic stem cell transplantation in hemophagocytic lymphohistiocytosis: a single-center report of 48 patients. *Pediatrics*, **2006**, 117, e743-50	7.4	200
306	Evidence of innate lymphoid cell redundancy in humans. *Nature Immunology*, **2016**, 17, 1291-1299	19.1	196
305	DOCK8 deficiency: clinical and immunological phenotype and treatment options - a review of 136 patients. *Journal of Clinical Immunology*, **2015**, 35, 189-98	5.7	196
304	Long-term outcome after hematopoietic stem cell transplantation of a single-center cohort of 90 patients with severe combined immunodeficiency. *Blood*, **2009**, 113, 4114-24	2.2	184
303	Immunotherapy of familial hemophagocytic lymphohistiocytosis with antithymocyte globulins: a single-center retrospective report of 38 patients. *Pediatrics*, **2007**, 120, e622-8	7.4	180
302	IRAK-4- and MyD88-dependent pathways are essential for the removal of developing autoreactive B cells in humans. *Immunity*, **2008**, 29, 746-57	32.3	178
301	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. *Nature Genetics*, **2011**, 43, 127-31	36.3	173
300	Human adenylate kinase 2 deficiency causes a profound hematopoietic defect associated with sensorineural deafness. *Nature Genetics*, **2009**, 41, 106-11	36.3	173
299	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. *Journal of Experimental Medicine*, **2015**, 212, 939-51	16.6	171
298	Interleukin (IL)-12 and IL-23 are key cytokines for immunity against Salmonella in humans. *Journal of Infectious Diseases*, **2004**, 190, 1755-7	7	169
297	A human immunodeficiency caused by mutations in the PIK3R1 gene. *Journal of Clinical Investigation*, **2014**, 124, 3923-8	15.9	166
296	Inherited disorders of NF-kappaB-mediated immunity in man. *Current Opinion in Immunology*, **2004**, 16, 34-41	7.8	166
295	Clinical and immunologic phenotype associated with activated phosphoinositide 3-kinase syndrome 2: A´cohort study. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 210-218.e9	11.5	163
294	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2015**, 35, 727-38	5.7	160
293	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. *Nature Genetics*, **2007**, 39, 454-6	36.3	152
292	Primary immunodeficiencies underlying fungal infections. *Current Opinion in Pediatrics*, **2013**, 25, 736-47	3.2	149
291	Long-term remissions of severe pemphigus after rituximab therapy are associated with prolonged failure of desmoglein B cell response. *Science Translational Medicine*, **2013**, 5, 175ra30	17.5	146
290	Ribosomal protein SA haploinsufficiency in humans with isolated congenital asplenia. *Science*, **2013**, 340, 976-8	33.3	145

289	Inherited CARD9 deficiency in otherwise healthy children and adults with Candida species-induced meningoencephalitis, colitis, or both. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1558-68.e2	11.5	143
288	Immunodeficiency due to mutations in ORAI1 and STIM1. *Clinical Immunology*, **2010**, 135, 169-82	9	143
287	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. *Journal of Experimental Medicine*, **2011**, 208, 2305-20	16.6	140
286	Novel STAT1 alleles in otherwise healthy patients with mycobacterial disease. *PLoS Genetics*, **2006**, 2, e131	6	138
285	Human interferon-gamma-mediated immunity is a genetically controlled continuous trait that determines the outcome of mycobacterial invasion. *Immunological Reviews*, **2000**, 178, 129-37	11.3	133
284	CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation. *Nature*, **2014**, 510, 288-92	50.4	131
283	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2015**, 212, 619-31	16.6	130
282	Immunity to infection in IL-17-deficient mice and humans. *European Journal of Immunology*, **2012**, 42, 2246-54	6.1	130
281	Autoimmune and inflammatory manifestations occur frequently in patients with primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 1388-1393.e8	11.5	129
280	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 993-1006.e1	11.5	126
279	Intrinsic antiproliferative activity of the innate sensor STING in T lymphocytes. *Journal of Experimental Medicine*, **2017**, 214, 1769-1785	16.6	125
278	Somatic diversification in the absence of antigen-driven responses is the hallmark of the IgM+ IgD+ CD27+ B cell repertoire in infants. *Journal of Experimental Medicine*, **2008**, 205, 1331-42	16.6	125
277	Recurrent staphylococcal cellulitis and subcutaneous abscesses in a child with autoantibodies against IL-6. *Journal of Immunology*, **2008**, 180, 647-54	5.3	122
276	Human Toll-like receptor-dependent induction of interferons in protective immunity to viruses. *Immunological Reviews*, **2007**, 220, 225-36	11.3	122
275	Experimental and natural infections in MyD88- and IRAK-4-deficient mice and humans. *European Journal of Immunology*, **2012**, 42, 3126-35	6.1	121
274	Naive and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells. *Journal of Experimental Medicine*, **2013**, 210, 2739-53	16.6	121
273	Inherited IL-12p40 deficiency: genetic, immunologic, and clinical features of 49 patients from 30 kindreds. *Medicine (United States)*, **2013**, 92, 109-122	1.8	121
272	A survey of 90 patients with autoimmune lymphoproliferative syndrome related to TNFRSF6 mutation. *Blood*, **2011**, 118, 4798-807	2.2	119

271	Bacillus Calmette Guerin triggers the IL-12/IFN-gamma axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and T lymphocytes. *European Journal of Immunology*, **2004**, 34, 3276-84	6.1	119
270	SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID. *Blood*, **2014**, 123, 281-9	2.2	115
269	Inherited disorders of human Toll-like receptor signaling: immunological implications. *Immunological Reviews*, **2005**, 203, 10-20	11.3	115
268	Human RTEL1 deficiency causes Hoyeraal-Hreidarsson syndrome with short telomeres and genome instability. *Human Molecular Genetics*, **2013**, 22, 3239-49	5.6	113
267	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. *Current Opinion in Immunology*, **2008**, 20, 39-48	7.8	113
266	Mutational, functional, and expression studies of the TCF4 gene in Pitt-Hopkins syndrome. *Human Mutation*, **2009**, 30, 669-76	4.7	112
265	Inborn errors of mucocutaneous immunity to Candida albicans in humans: a role for IL-17 cytokines?. *Current Opinion in Immunology*, **2010**, 22, 467-74	7.8	112
264	Juvenile myelomonocytic leukemia displays mutations in components of the RAS pathway and the PRC2 network. *Nature Genetics*, **2015**, 47, 1334-40	36.3	111
263	Whole-exome sequencing identifies Coronin-1A deficiency in 3 siblings with immunodeficiency and EBV-associated B-cell lymphoproliferation. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 1594-603	11.5	111
262	Type I interferon-mediated autoinflammation due to DNase II deficiency. *Nature Communications*, **2017**, 8, 2176	17.4	111
261	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. *Journal of Medical Genetics*, **2007**, 44, 16-23	5.8	111
260	Exome and genome sequencing for inborn errors of immunity. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 957-969	11.5	111
259	Mutations in CDCA7 and HELLS cause immunodeficiency-centromeric instability-facial anomalies syndrome. *Nature Communications*, **2015**, 6, 7870	17.4	110
258	FAS-L, IL-10, and double-negative CD4- CD8- TCR alpha/beta+ T cells are reliable markers of autoimmune lymphoproliferative syndrome (ALPS) associated with FAS loss of function. *Blood*, **2009**, 113, 3027-30	2.2	110
257	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. *Journal of Clinical Investigation*, **2012**, 122, 3239-47	15.9	109
256	Morbidity and mortality from ataxia-telangiectasia are associated with ATM genotype. *Journal of Allergy and Clinical Immunology*, **2011**, 128, 382-9.e1	11.5	105
255	Primary immunodeficiencies associated with pneumococcal disease. *Current Opinion in Allergy and Clinical Immunology*, **2003**, 3, 451-9	3.3	104
254	Inherited CARD9 deficiency in 2 unrelated patients with invasive Exophiala infection. *Journal of Infectious Diseases*, **2015**, 211, 1241-50	7	101

253	Inherited MST1 deficiency underlies susceptibility to EV-HPV infections. *PLoS ONE*, **2012**, 7, e44010	3.7	101
252	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. *Journal of Experimental Medicine*, **2013**, 210, 1743-59	16.6	99
251	HHV-8-associated Kaposi sarcoma in a child with IFNgammaR1 deficiency. *Journal of Pediatrics*, **2004**, 144, 519-23	3.6	92
250	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. *Journal of Clinical Investigation*, **2017**, 127, 3543-3556	15.9	91
249	Disease Evolution and Response to Rapamycin in Activated Phosphoinositide 3-Kinase δ Syndrome: The European Society for Immunodeficiencies-Activated Phosphoinositide 3-Kinase δ Syndrome Registry. *Frontiers in Immunology*, **2018**, 9, 543	8.4	88
248	A Mendelian predisposition to B-cell lymphoma caused by IL-10R deficiency. *Blood*, **2013**, 122, 3713-22	2.2	88
247	First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases. *Blood*, **2011**, 117, 688-96	2.2	88
246	Inherited CD70 deficiency in humans reveals a critical role for the CD70-CD27 pathway in immunity to Epstein-Barr virus infection. *Journal of Experimental Medicine*, **2017**, 214, 73-89	16.6	87
245	Onset of autoimmune lymphoproliferative syndrome (ALPS) in humans as a consequence of genetic defect accumulation. *Journal of Clinical Investigation*, **2011**, 121, 106-12	15.9	87
244	Occurrence of B-cell lymphomas in patients with activated phosphoinositide 3-kinase δ syndrome. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 233-6	11.5	85
243	Interleukin receptor-associated kinase (IRAK-4) deficiency associated with bacterial infections and failure to sustain antibody responses. *Journal of Pediatrics*, **2004**, 144, 524-6	3.6	85
242	IL-21 signalling via STAT3 primes human naive B cells to respond to IL-2 to enhance their differentiation into plasmablasts. *Blood*, **2013**, 122, 3940-50	2.2	84
241	Major histocompatibility complex class II expression deficiency caused by a RFXANK founder mutation: a survey of 35 patients. *Blood*, **2011**, 118, 5108-18	2.2	84
240	Characterization of Crohn disease in X-linked inhibitor of apoptosis-deficient male patients and female symptomatic carriers. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 1131-41.e9	11.5	83
239	Hematopoietic stem cell transplantation for complete IFN-gamma receptor 1 deficiency: a multi-institutional survey. *Journal of Pediatrics*, **2004**, 145, 806-12	3.6	83
238	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. *Science Immunology*, **2018**, 3,	28	82
237	Human iNKT and MAIT cells exhibit a PLZF-dependent proapoptotic propensity that is counterbalanced by XIAP. *Blood*, **2013**, 121, 614-23	2.2	82
236	Clinical features of Candidiasis in patients with inherited interleukin 12 receptor β1 deficiency. *Clinical Infectious Diseases*, **2014**, 58, 204-13	11.6	81

235	Impaired interferon gamma-mediated immunity and susceptibility to mycobacterial infection in childhood. *Pediatric Research*, **2001**, 50, 8-13	3.2	80
234	Reduced expression of FOXP3 and regulatory T-cell function in severe forms of early-onset autoimmune enteropathy. *Gastroenterology*, **2010**, 139, 770-8	13.3	79
233	Primary T-cell immunodeficiency with immunodysregulation caused by autosomal recessive LCK deficiency. *Journal of Allergy and Clinical Immunology*, **2012**, 130, 1144-1152.e11	11.5	78
232	Inherited disorders of IL-12- and IFNgamma-mediated immunity: a molecular genetics update. *Molecular Immunology*, **2002**, 38, 903-9	4.3	77
231	Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency. *Journal of Clinical Investigation*, **2018**, 128, 3071-3087	15.9	77
230	Mycobacterial disease in patients with chronic granulomatous disease: A´retrospective analysis of 71 cases. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 241-248.e3	11.5	76
229	Dual T cell- and B cell-intrinsic deficiency in humans with biallelic RLTPR mutations. *Journal of Experimental Medicine*, **2016**, 213, 2413-2435	16.6	75
228	Partial recessive IFN-γR1 deficiency: genetic, immunological and clinical features of 14 patients from 11 kindreds. *Human Molecular Genetics*, **2011**, 20, 1509-23	5.6	75
227	Hypomorphic mutation of ZAP70 in human results in a late onset immunodeficiency and no autoimmunity. *European Journal of Immunology*, **2009**, 39, 1966-76	6.1	73
226	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. *Journal of Clinical Investigation*, **2017**, 127, 1991-2006	15.9	73
225	IgM+IgD+CD27+ B cells are markedly reduced in IRAK-4-, MyD88-, and TIRAP- but not UNC-93B-deficient patients. *Blood*, **2012**, 120, 4992-5001	2.2	69
224	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. *Nature Immunology*, **2018**, 19, 973-985	19.1	67
223	Genetic lessons learned from X-linked Mendelian susceptibility to mycobacterial diseases. *Annals of the New York Academy of Sciences*, **2011**, 1246, 92-101	6.5	67
222	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. *Journal of Clinical Immunology*, **2021**, 41, 666-679	5.7	66
221	Characteristics and outcome of early-onset, severe forms of Wiskott-Aldrich syndrome. *Blood*, **2013** , 121, 1510-6	2.2	65
220	SARS-CoV-2 induces human plasmacytoid predendritic cell diversification via UNC93B and IRAK4. *Journal of Experimental Medicine*, **2021**, 218,	16.6	65
219	Prevention of infections during primary immunodeficiency. *Clinical Infectious Diseases*, **2014**, 59, 1462-70	11.6	62
218	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic /NEMO mutations. *Blood*, **2017**, 130, 1456-1467	2.2	61

217	Macrophages induce differentiation of plasma cells through CXCL10/IP-10. *Journal of Experimental Medicine*, **2012**, 209, 1813-23, S1-2	16.6	60
216	A fast procedure for the detection of defects in Toll-like receptor signaling. *Pediatrics*, **2006**, 118, 2498-503	7.4	60
215	Pediatric Evans syndrome is associated with a high frequency of potentially damaging variants in immune genes. *Blood*, **2019**, 134, 9-21	2.2	58
214	Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. *Cell*, **2017**, 168, 789-800.e10	56.2	57
213	Primary immunodeficiencies of protective immunity to primary infections. *Clinical Immunology*, **2010**, 135, 204-9	9	57
212	NEMO mutations in 2 unrelated boys with severe infections and conical teeth. *Pediatrics*, **2005**, 115, e615-9	7.4	57
211	Clinical, immunologic, and genetic spectrum of 696 patients with combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2018**, 141, 1450-1458	11.5	56
210	Invasive pneumococcal disease in children can reveal a primary immunodeficiency. *Clinical Infectious Diseases*, **2014**, 59, 244-51	11.6	56
209	Human primary immunodeficiencies of type I interferons. *Biochimie*, **2007**, 89, 878-83	4.6	56
208	Successful allogeneic hematopoietic stem cell transplantation for DOCK8 deficiency. *Journal of Allergy and Clinical Immunology*, **2011**, 128, 420-22.e2	11.5	55
207	Polymerase ϕ mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature ("FILS syndrome"). *Journal of Experimental Medicine*, **2012**, 209, 2323-30	16.6	55
206	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). *Nature Genetics*, **2008**, 40, 284-6	36.3	55
205	Mendelian traits that confer predisposition or resistance to specific infections in humans. *Current Opinion in Immunology*, **2006**, 18, 383-90	7.8	55
204	Shigella sonnei meningitis due to interleukin-1 receptor-associated kinase-4 deficiency: first association with a primary immune deficiency. *Clinical Infectious Diseases*, **2005**, 40, 1227-31	11.6	55
203	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. *Journal of Experimental Medicine*, **2015**, 212, 855-64	16.6	54
202	A novel gain-of-function IKBA mutation underlies ectodermal dysplasia with immunodeficiency and polyendocrinopathy. *Journal of Clinical Immunology*, **2013**, 33, 1088-99	5.7	54
201	Laboratory diagnosis of specific antibody deficiency to pneumococcal capsular polysaccharide antigens by multiplexed bead assay. *Clinical Immunology*, **2010**, 134, 198-205	9	54
200	High levels of interferon gamma in the plasma of children with complete interferon gamma receptor deficiency. *Pediatrics*, **2001**, 107, E48	7.4	54

199	Phenotypic complementation of genetic immunodeficiency by chronic herpesvirus infection. *ELife*, **2015**, 4,	8.9	54
198	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. *Human Molecular Genetics*, **2013**, 22, 769-81	5.6	53
197	Isolated congenital asplenia: a French nationwide retrospective survey of 20 cases. *Journal of Pediatrics*, **2011**, 158, 142-8, 148.e1	3.6	53
196	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1578-88.e5	11.5	52
195	A polysaccharide virulence factor of a human fungal pathogen induces neutrophil apoptosis via NK cells. *Journal of Immunology*, **2014**, 192, 5332-42	5.3	52
194	Faster T-cell development following gene therapy compared with haploidentical HSCT in the treatment of SCID-X1. *Blood*, **2015**, 125, 3563-9	2.2	52
193	TLR-mediated inflammatory responses to Streptococcus pneumoniae are highly dependent on surface expression of bacterial lipoproteins. *Journal of Immunology*, **2014**, 193, 3736-45	5.3	52
192	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. *Journal of Experimental Medicine*, **2008**, 205, 1729-37	16.6	52
191	A narrow repertoire of transcriptional modules responsive to pyogenic bacteria is impaired in patients carrying loss-of-function mutations in MYD88 or IRAK4. *Nature Immunology*, **2014**, 15, 1134-42	19.1	51
190	ZAP70: a master regulator of adaptive immunity. *Seminars in Immunopathology*, **2010**, 32, 107-16	12	51
189	Unique and shared signaling pathways cooperate to regulate the differentiation of human CD4+ T cells into distinct effector subsets. *Journal of Experimental Medicine*, **2016**, 213, 1589-608	16.6	51
188	Haploidentical Hematopoietic Stem Cell Transplantation with Post-Transplant Cyclophosphamide for Primary Immunodeficiencies and Inherited Disorders in Children. *Biology of Blood and Marrow Transplantation*, **2019**, 25, 1363-1373	4.7	50
187	Trends of pneumococcal meningitis in children after introduction of the 13-valent pneumococcal conjugate vaccine in France. *Pediatric Infectious Disease Journal*, **2014**, 33, 1216-21	3.4	49
186	Signal transducer and activator of transcription 3 (STAT3) mutations underlying autosomal dominant hyper-IgE syndrome impair human CD8(+) T-cell memory formation and function. *Journal of Allergy and Clinical Immunology*, **2013**, 132, 400-11.e9	11.5	48
185	Immune deficiency-related enteropathy-lymphocytopenia-alopecia syndrome results from tetratricopeptide repeat domain 7A deficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 1354-1364.e6	11.5	48
184	Successful allogeneic hemopoietic stem cell transplantation in a child who had anhidrotic ectodermal dysplasia with immunodeficiency. *Pediatrics*, **2006**, 118, e205-11	7.4	48
183	Self-reactive VH4-34-expressing IgG B cells recognize commensal bacteria. *Journal of Experimental Medicine*, **2017**, 214, 1991-2003	16.6	46
182	CD45RA depletion in HLA-mismatched allogeneic hematopoietic stem cell transplantation for primary combined immunodeficiency: A preliminary study. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1303-9.e1-3	11.5	46

181	Human IBD Gain of Function: a Severe and Syndromic Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 397-412	5.7	45
180	X-linked primary immunodeficiency associated with hemizygous mutations in the moesin (MSN) gene. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 1681-1689.e8	11.5	45
179	New mechanism of X-linked anhidrotic ectodermal dysplasia with immunodeficiency: impairment of ubiquitin binding despite normal folding of NEMO protein. *Blood*, **2011**, 118, 926-35	2.2	45
178	Rituximab therapy for childhood Evans syndrome. *Haematologica*, **2007**, 92, 1691-4	6.6	45
177	Autosomal recessive interleukin-1 receptor-associated kinase 4 deficiency in fourth-degree relatives. *Journal of Pediatrics*, **2006**, 148, 549-51	3.6	45
176	An in vivo genetic reversion highlights the crucial role of Myb-Like, SWIRM, and MPN domains 1 (MYSM1) in human hematopoiesis and lymphocyte differentiation. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1619-1626.e5	11.5	44
175	Loss of RASGRP1 in humans impairs T-cell expansion leading to Epstein-Barr virus susceptibility. *EMBO Molecular Medicine*, **2018**, 10, 188-199	12	44
174	From idiopathic infectious diseases to novel primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2005**, 116, 426-30	11.5	44
173	A novel X-linked recessive form of Mendelian susceptibility to mycobaterial disease. *Journal of Medical Genetics*, **2007**, 44, e65	5.8	44
172	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	11.5	43
171	Recurrent Mycobacterium avium osteomyelitis associated with a novel dominant interferon gamma receptor mutation. *Pediatrics*, **2001**, 107, E47	7.4	43
170	ORAI1 mutations abolishing store-operated Ca entry cause anhidrotic ectodermal dysplasia with immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1297-1310.e11	11.5	42
169	Protective effect of IgM against colonization of the respiratory tract by nontypeable Haemophilus influenzae in patients with hypogammaglobulinemia. *Journal of Allergy and Clinical Immunology*, **2012**, 129, 770-7	11.5	42
168	B-cell subpopulations in children: National reference values. *Immunity, Inflammation and Disease*, **2014**, 2, 131-40	2.4	40
167	Frequent and widespread vascular abnormalities in human signal transducer and activator of transcription 3 deficiency. *Circulation: Cardiovascular Genetics*, **2012**, 5, 25-34		40
166	IRAK-4 mutation (Q293X): rapid detection and characterization of defective post-transcriptional TLR/IL-1R responses in human myeloid and non-myeloid cells. *Journal of Immunology*, **2006**, 177, 8202-11	5.3	40
165	Human leucocyte antigen-identical haematopoietic stem cell transplantation in major histocompatiblity complex class II immunodeficiency: reduced survival correlates with an increased incidence of acute graft-versus-host disease and pre-existing viral infections. *British Journal of Haematology*, **2006**, 134, 510-6	4.5	39
164	Septicemia without sepsis: inherited disorders of nuclear factor-kappa B-mediated inflammation. *Clinical Infectious Diseases*, **2005**, 41 Suppl 7, S436-9	11.6	39

163	Hematopoietic Stem Cell Transplantation as Treatment for Patients with DOCK8 Deficiency. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 848-855	5.4	39
162	Diagnostic Yield of Next-generation Sequencing in Very Early-onset Inflammatory Bowel Diseases: A Multicentre Study. *Journal of Crohn's and Colitis*, **2018**, 12, 1104-1112	1.5	39
161	A novel hypomorphic mutation in STIM1 results in a late-onset immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 816-819.e4	11.5	38
160	Autoimmune lymphoproliferative syndrome caused by a homozygous null FAS ligand (FASLG) mutation. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 486-90	11.5	38
159	Contribution of high-throughput DNA sequencing to the study of primary immunodeficiencies. *European Journal of Immunology*, **2014**, 44, 2854-61	6.1	38
158	Classic Kaposi sarcoma in 3 unrelated Turkish children born to consanguineous kindreds. *Pediatrics*, **2010**, 125, e704-8	7.4	38
157	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, 6713-8	11.5	37
156	Primary microcephaly, impaired DNA replication, and genomic instability caused by compound heterozygous ATR mutations. *Human Mutation*, **2013**, 34, 374-84	4.7	37
155	Clinical spectrum and long-term follow-up of 14 cases with G6PC3 mutations from the French Severe Congenital Neutropenia Registry. *Orphanet Journal of Rare Diseases*, **2014**, 9, 183	4.2	37
154	A novel developmental and immunodeficiency syndrome associated with intrauterine growth retardation and a lack of natural killer cells. *Pediatrics*, **2004**, 113, 136-41	7.4	37
153	Pediatric-onset Evans syndrome: Heterogeneous presentation and high frequency of monogenic disorders including LRBA and CTLA4 mutations. *Clinical Immunology*, **2018**, 188, 52-57	9	36
152	Interleukin 1/Toll-like receptor-induced autophosphorylation activates interleukin 1 receptor-associated kinase 4 and controls cytokine induction in a cell type-specific manner. *Journal of Biological Chemistry*, **2014**, 289, 10865-10875	5.4	36
151	Defective anti-polysaccharide response and splenic marginal zone disorganization in ALPS patients. *Blood*, **2014**, 124, 1597-609	2.2	36
150	IRAK-4 and MyD88 deficiencies impair IgM responses against T-independent bacterial antigens. *Blood*, **2014**, 124, 3561-71	2.2	36
149	Human X-linked variable immunodeficiency caused by a hypomorphic mutation in XIAP in association with a rare polymorphism in CD40LG. *Blood*, **2011**, 118, 252-61	2.2	35
148	Concomitant and deficiencies cause chronic active Epstein-Barr virus infection of T cells. *Journal of Experimental Medicine*, **2019**, 216, 2800-2818	16.6	33
147	EVER2 deficiency is associated with mild T-cell abnormalities. *Journal of Clinical Immunology*, **2013**, 33, 14-21	5.7	33
146	Three novel ZBTB24 mutations identified in Japanese and Cape Verdean type 2 ICF syndrome patients. *Journal of Human Genetics*, **2013**, 58, 455-60	4.3	33

145	Kaposi's sarcoma in a child with Wiskott-Aldrich syndrome. *European Journal of Pediatrics*, **2006**, 165, 453-7	4.1	32
144	Genetic, Cellular and Clinical Features of ICF Syndrome: a French National Survey. *Journal of Clinical Immunology*, **2016**, 36, 149-59	5.7	32
143	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E514-E523	11.5	31
142	Perforin-dependent apoptosis functionally compensates Fas deficiency in activation-induced cell death of human T lymphocytes. *Blood*, **2007**, 110, 4285-92	2.2	31
141	Evolution of disease activity and biomarkers on and off rapamycin in 28 patients with autoimmune lymphoproliferative syndrome. *Haematologica*, **2017**, 102, e52-e56	6.6	30
140	Founder effect for a 26-bp deletion in the RFXANK gene in North African major histocompatibility complex class II-deficient patients belonging to complementation group B. *Immunogenetics*, **2000**, 51, 261-7	3.2	30
139	Comparative methylome analysis of ICF patients identifies heterochromatin loci that require ZBTB24, CDCA7 and HELLS for their methylated state. *Human Molecular Genetics*, **2018**, 27, 2409-2424	5.6	28
138	Inherited human IRAK-4 deficiency: an update. *Immunologic Research*, **2007**, 38, 347-52	4.3	28
137	Early-onset autoimmunity associated with SOCS1 haploinsufficiency. *Nature Communications*, **2020**, 11, 5341	17.4	28
136	Multicentric Castleman disease in an HHV8-infected child born to consanguineous parents with systematic review. *Pediatrics*, **2012**, 129, e199-203	7.4	27
135	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12Rβ1 Deficiency. *Journal of Clinical Immunology*, **2018**, 38, 617-627	5.7	26
134	ICON: the early diagnosis of congenital immunodeficiencies. *Journal of Clinical Immunology*, **2014**, 34, 398-424	5.7	26
133	Inherited disorders of cytokines. *Current Opinion in Pediatrics*, **2004**, 16, 648-58	3.2	26
132	Mutations in the adaptor-binding domain and associated linker region of p110δ cause Activated PI3K-δ syndrome 1 (APDS1). *Haematologica*, **2017**, 102, e278-e281	6.6	25
131	PROMIDIS: A T-cell receptor δ signature associated with immunodeficiencies caused by V(D)J recombination defects. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 325-334.e2	11.5	25
130	Granulomatous inflammation in cartilage-hair hypoplasia: risks and benefits of anti-TNF-α mAbs. *Journal of Allergy and Clinical Immunology*, **2011**, 128, 847-53	11.5	25
129	Clinical disease caused by Klebsiella in 2 unrelated patients with interleukin 12 receptor beta1 deficiency. *Pediatrics*, **2010**, 126, e971-6	7.4	25
128	Three novel mutations of the CIITA gene in MHC class II-deficient patients with a severe immunodeficiency. *Immunogenetics*, **2002**, 53, 821-9	3.2	25

127	Heritable defects of the human TLR signalling pathways. *Journal of Endotoxin Research*, **2005**, 11, 220-4		25
126	Strains Responsible for Invasive Meningococcal Disease in Patients With Terminal Complement Pathway Deficiencies. *Journal of Infectious Diseases*, **2017**, 215, 1331-1338	7	24
125	Incomplete penetrance for isolated congenital asplenia in humans with mutations in translated and untranslated exons. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E8007-E8016	11.5	24
124	Very late-onset group B Streptococcus meningitis, sepsis, and systemic shigellosis due to interleukin-1 receptor-associated kinase-4 deficiency. *Clinical Infectious Diseases*, **2009**, 49, 1393-6	11.6	23
123	Hematopoietic stem cell transplantation and other management strategies for MHC class II deficiency. *Immunology and Allergy Clinics of North America*, **2010**, 30, 173-8	3.3	22
122	Inherited defects in the interferon-gamma receptor or interleukin-12 signalling pathways are not sufficient to cause allergic disease in children. *European Journal of Pediatrics*, **2005**, 164, 741-7	4.1	22
121	Loss of ARHGEF1 causes a human primary antibody deficiency. *Journal of Clinical Investigation*, **2019** , 129, 1047-1060	15.9	22
120	Clinical and economic aspects of newborn screening for severe combined immunodeficiency: DEPISTREC study results. *Clinical Immunology*, **2019**, 202, 33-39	9	21
119	SYK expression endows human ZAP70-deficient CD8 T cells with residual TCR signaling. *Clinical Immunology*, **2015**, 161, 103-9	9	21
118	Mammalian target of rapamycin inhibition counterbalances the inflammatory status of immune cells in patients with chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1641-1649.e6	11.5	21
117	Human plasma cells express granzyme B. *European Journal of Immunology*, **2014**, 44, 275-84	6.1	21
116	Cutaneous granulomas with primary immunodeficiency in children: a report of 17 new patients and a review of the literature. *Journal of the European Academy of Dermatology and Venereology*, **2019**, 33, 1412-1420	4.6	20
115	Heterozygous Mutations in MAP3K7, Encoding TGF-Activated Kinase 1, Cause Cardiospondylocarpofacial Syndrome. *American Journal of Human Genetics*, **2016**, 99, 407-13	11	20
114	Germline genes hypomethylation and expression define a molecular signature in peripheral blood of ICF patients: implications for diagnosis and etiology. *Orphanet Journal of Rare Diseases*, **2014**, 9, 56	4.2	20
113	Blood CD4+CD45RO+CXCR5+ T cells are decreased but partially functional in signal transducer and activator of transcription 3 deficiency. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 1146-56, 1156.e1-5	11.5	20
112	Cutaneous findings in sporadic and familial autosomal dominant hyper-IgE syndrome: a retrospective, single-center study of 21 patients diagnosed using molecular analysis. *Journal of the American Academy of Dermatology*, **2011**, 65, 1167-72	4.5	20
111	Characteristics of HIV-infected children recently diagnosed in Paris, France. *European Journal of Pediatrics*, **2006**, 165, 684-7	4.1	20
110	IRAK4 Deficiency in a Patient with Recurrent Pneumococcal Infections: Case Report and Review of the Literature. *Frontiers in Pediatrics*, **2017**, 5, 83	3.4	19

109	Diagnosis of autoimmune lymphoproliferative syndrome caused by FAS deficiency in adults. *Haematologica*, **2013**, 98, 389-92	6.6	19
108	Occurrence of aortic aneurysms in 5 cases of Wiskott-Aldrich syndrome. *Pediatrics*, **2011**, 127, e498-504	7.4	18
107	Pneumococcal Meningitis Vaccine Breakthroughs and Failures After Routine 7-Valent and 13-Valent Pneumococcal Conjugate Vaccination in Children in France. *Pediatric Infectious Disease Journal*, **2015**, 34, e260-3	3.4	17
106	Analysis of the interleukin-12/interferon-gamma pathway in children with non-tuberculous mycobacterial cervical lymphadenitis. *European Journal of Pediatrics*, **2007**, 166, 835-41	4.1	17
105	Clinical and Genetic Spectrum of a Large Cohort With Total and Sub-total Complement Deficiencies. *Frontiers in Immunology*, **2019**, 10, 1936	8.4	16
104	Severe cutaneous bacillus Calmette-Guŕin infection in immunocompromised children: the relevance of skin biopsy. *Journal of Cutaneous Pathology*, **2013**, 40, 30-7	1.7	16
103	The transcription factor RFX protects MHC class II genes against epigenetic silencing by DNA methylation. *Journal of Immunology*, **2009**, 183, 2545-53	5.3	16
102	Diffuse large B-cell non-Hodgkin's lymphoma in a patient with autoimmune lymphoproliferative syndrome. *British Journal of Haematology*, **2001**, 113, 432-4	4.5	16
101	Specific T cells for the treatment of cytomegalovirus and/or adenovirus in the context of hematopoietic stem cell transplantation. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 920-924.e3	11.5	15
100	Addressing diagnostic challenges in primary immunodeficiencies: laboratory evaluation of Toll-like receptor- and NF-B-mediated immune responses. *Critical Reviews in Clinical Laboratory Sciences*, **2014**, 51, 112-23	9.4	15
99	Intestinal dysbiosis in inflammatory bowel disease associated with primary immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 775-778.e6	11.5	15
98	Mechanisms of genotype-phenotype correlation in autosomal dominant anhidrotic ectodermal dysplasia with immune deficiency. *Journal of Allergy and Clinical Immunology*, **2018**, 141, 1060-1073.e3	11.5	14
97	Massive expansion of maternal T cells in response to EBV infection in a patient with SCID-Xl. *Blood*, **2012**, 120, 1957-9	2.2	14
96	A gene responsible for Ghosal hemato-diaphyseal dysplasia maps to chromosome 7q33-34. *Human Genetics*, **2007**, 121, 269-73	6.3	14
95	Persistence of natural killer cells with expansion of a hypofunctional CD56-CD16+KIR+NKG2C+ subset in a patient with atypical Janus kinase 3-deficient severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 1230-3, 1233.e1-2	11.5	13
94	Imported African histoplasmosis in an immunocompetent patient 40 years after staying in a disease-endemic area. *American Journal of Tropical Medicine and Hygiene*, **2014**, 91, 1011-4	3.2	13
93	Occurrence of myelodysplastic syndrome in 2 patients with reticular dysgenesis. *Journal of Allergy and Clinical Immunology*, **2011**, 128, 230-232.e2	11.5	13
92	Burkholderia pseudomallei infection in chronic granulomatous disease. *European Journal of Pediatrics*, **2006**, 165, 175-7	4.1	13

91	Impaired lymphocyte function and differentiation in CTPS1-deficient patients result from a hypomorphic homozygous mutation. *JCI Insight*, **2020**, 5,	9.9	13
90	Rapid identification and characterization of infected cells in blood during chronic active Epstein-Barr virus infection. *Journal of Experimental Medicine*, **2020**, 217,	16.6	13
89	Spectrum of Pulmonary Aspergillosis in Hyper-IgE Syndrome with Autosomal-Dominant STAT3 Deficiency. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 1986-1995.e3	5.4	12
88	DOCK8 Drives Src-Dependent NK Cell Effector Function. *Journal of Immunology*, **2017**,	5.3	12
87	Physical health conditions and quality of life in adults with primary immunodeficiency diagnosed during childhood: A French Reference Center for PIDs (CEREDIH) study. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1275-1281.e7	11.5	12
86	Cellular and humoral aberrations in a kindred with IL-1 receptor-associated kinase 4 deficiency. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 948-50	11.5	12
85	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. *JCI Insight*, **2019**, 5,	9.9	12
84	Infection in Humans With STAT3-Deficiency Is Associated With Defective Interferon-Gamma and Th17 Responses. *Frontiers in Immunology*, **2020**, 11, 38	8.4	12
83	Somatic reversion of pathogenic DOCK8 variants alters lymphocyte differentiation and function to effectively cure DOCK8 deficiency. *Journal of Clinical Investigation*, **2021**, 131,	15.9	12
82	Lack of interaction between NEMO and SHARPIN impairs linear ubiquitination and NF-B activation and leads to incontinentia pigmenti. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 1671-1682.e2	11.5	11
81	Severe Mycobacterial Diseases in a Patient with GOF IB Mutation Without EDA. *Journal of Clinical Immunology*, **2016**, 36, 12-5	5.7	11
80	A deep intronic splice mutation of underlies hyper IgE syndrome by negative dominance. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 16463-16472	11.5	11
79	Immune constitution monitoring after PBMC transplantation in complete DiGeorge syndrome: an eight-year follow-up. *Clinical Immunology*, **2008**, 128, 164-71	9	11
78	SARS-CoV-2 induces human plasmacytoid pre-dendritic cell diversification via UNC93B and IRAK4 **2021**,		11
77	Progressive Multifocal Leukoencephalopathy in Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2019**, 39, 55-64	5.7	11
76	An atypical case of X-linked lymphoproliferative disease revealed as a late cerebral lymphoma. *Journal of Neuroimmunology*, **2010**, 218, 125-8	3.5	10
75	Risk Factors in Children Older Than 5 Years With Pneumococcal Meningitis: Data From a National Network. *Pediatric Infectious Disease Journal*, **2017**, 36, 457-461	3.4	9
74	CD21 deficiency in 2 siblings with recurrent respiratory infections and hypogammaglobulinemia. *Journal of Allergy and Clinical Immunology: in Practice*, **2017**, 5, 1765-1767.e3	5.4	9

73	Hodgkin lymphoma in 2 children with chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2011**, 127, 543-544.e1-3	11.5	9
72	Genetic diagnosis of primary immunodeficiencies: A´survey of the French national registry. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 1646-1649.e10	11.5	9
71	Successful Haploidentical Stem Cell Transplantation with Post-Transplant Cyclophosphamide in a Severe Combined Immune Deficiency Patient: a First Report. *Journal of Clinical Immunology*, **2016**, 36, 437-40	5.7	9
70	DEF6 deficiency, a mendelian susceptibility to EBV infection, lymphoma, and autoimmunity. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 740-743.e9	11.5	9
69	A RAB27A duplication in several cases of Griscelli syndrome type 2: An explanation for cases lacking a genetic diagnosis. *Human Mutation*, **2017**, 38, 1355-1359	4.7	8
68	Recurrent Respiratory Infections Revealing CD8 Deficiency. *Journal of Clinical Immunology*, **2015**, 35, 692-5	5.7	8
67	Value of allohaemagglutinins in the diagnosis of a polysaccharide antibody deficiency. *Clinical and Experimental Immunology*, **2015**, 180, 271-9	6.2	8
66	BCG Moreau Vaccine Safety Profile and NK Cells-Double Protection Against Disseminated BCG Infection in Retrospective Study of BCG Vaccination in 52 Polish Children with Severe Combined Immunodeficiency. *Journal of Clinical Immunology*, **2020**, 40, 138-146	5.7	8
65	Improving the diagnostic efficiency of primary immunodeficiencies with targeted next-generation sequencing. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 734-737	11.5	8
64	OL-EDA-ID Syndrome: a Novel Hypomorphic NEMO Mutation Associated with a Severe Clinical Presentation and Transient HLH. *Journal of Clinical Immunology*, **2017**, 37, 7-11	5.7	7
63	Detection of 28 novel mutations in the Wiskott-Aldrich syndrome and X-linked thrombocytopenia based on multiplex PCR. *Blood Cells, Molecules, and Diseases*, **2007**, 39, 102-6	2.1	7
62	A gain-of-function RAC2 mutation is associated with bone-marrow hypoplasia and an autosomal dominant form of severe combined immunodeficiency. *Haematologica*, **2021**, 106, 404-411	6.6	7
61	Topoisomerase 2 mutation impairs early B-cell development. *Blood*, **2020**, 135, 1497-1501	2.2	6
60	Autoimmune Lymphoproliferative Syndrome-FAS Patients Have an Abnormal Regulatory T Cell (Treg) Phenotype but Display Normal Natural Treg-Suppressive Function on T Cell Proliferation. *Frontiers in Immunology*, **2018**, 9, 718	8.4	6
59	Deficiency of interleukin-1 receptor-associated kinase 4 presenting as fatal Pseudomonas aeruginosa bacteremia in two siblings. *Pediatric Infectious Disease Journal*, **2015**, 34, 299-300	3.4	6
58	Human STAT3 variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. *Journal of Experimental Medicine*, **2021**, 218,	16.6	6
57	Proinflammatory cytokine response toward fungi but not bacteria in chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 928-930.e4	11.5	6
56	Copy number variations and founder effect underlying complete IL-10R deficiency in Portuguese kindreds. *PLoS ONE*, **2018**, 13, e0205826	3.7	6

55	Temporal interferon-gamma release response to Mycobacterium kansasii infection in an anorexia nervosa patient. *Journal of Medical Microbiology*, **2012**, 61, 1617-1620	3.2	5
54	Long-term safety and efficacy of lentiviral hematopoietic stem/progenitor cell gene therapy for Wiskott-Aldrich syndrome.. *Nature Medicine*, **2022**,	50.5	5
53	Alemtuzumab as First Line Treatment in Children with Familial Lymphohistiocytosis. *Blood*, **2019**, 134, 80-80	2.2	5
52	Burden of Poor Health Conditions and Quality of Life in 656 Children with Primary Immunodeficiency. *Journal of Pediatrics*, **2018**, 194, 211-217.e5	3.6	5
51	Successful in utero stem cell transplantation in X-linked severe combined immunodeficiency. *Blood Advances*, **2019**, 3, 237-241	7.8	5
50	Chronic Intestinal Pseudo-Obstruction and Lymphoproliferative Syndrome as a Novel Phenotype Associated With Tetratricopeptide Repeat Domain 7A Deficiency. *Frontiers in Immunology*, **2019**, 10, 2592	8.4	5
49	Comprehensive molecular diagnosis of Epstein-Barr virus-associated lymphoproliferative diseases using next-generation sequencing. *International Journal of Hematology*, **2018**, 108, 319-328	2.3	5
48	Early-onset hypogammaglobulinemia: A survey of 44 patients. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1097-9.e2	11.5	4
47	A neuropathological study of cerebrovascular abnormalities in a signal transducer and activator of transcription 3-deficient patient. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1418-21.e1-5	11.5	4
46	Diffuse lymphoplasmacytic bronchiolitis in cartilage-hair hypoplasia. *Journal of Pediatrics*, **2008**, 152, 429-33	3.6	4
45	Novel primary immunodeficiencies. *Advances in Experimental Medicine and Biology*, **2005**, 568, 89-99	3.6	4
44	Fulminant arterial vasculitis as an unusual complication of disseminated staphylococcal disease due to the emerging CC1 methicillin-susceptible Staphylococcus aureus clone: a case report. *BMC Infectious Diseases*, **2019**, 19, 302	4	3
43	Increased proportions of □T lymphocytes in atypical SCID associate with disease manifestations. *Clinical Immunology*, **2019**, 201, 30-34	9	3
42	The Genetic and Molecular Basis of Severe Combined Immunodeficiency. *Current Pediatrics Reports*, **2015**, 3, 22-33	0.7	3
41	Severe combined immunodeficiency caused by a new homozygous RAG1 mutation with progressive encephalopathy. *Hematology/ Oncology and Stem Cell Therapy*, **2014**, 7, 44-9	2.7	3
40	Pachymeningitis after meningococcal infection. *Lancet, The*, **2013**, 381, 1596	40	3
39	Type □hyper IgM syndrome with novel mutation from India. *Indian Journal of Pediatrics*, **2014**, 81, 620-2	3	3
38	Current Spectrum of Infections in Patients with X-Linked Agammaglobulinemia. *Journal of Clinical Immunology*, **2021**, 41, 1266-1271	5.7	3

37	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation. *Journal of Clinical Immunology*, **2021**, 41, 125-135	5.7	3
36	Different Immunological Pathways Underlie the Immune Response to Pneumococcal Polysaccharides. *Journal of Clinical Immunology*, **2017**, 37, 277-278	5.7	2
35	Long-term consequences of Hodgkin lymphoma therapy on T-cell lymphopoiesis. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 818-20.e4	11.5	2
34	Evolution of the Definition of Primary Immunodeficiencies **2014**, 29-40		2
33	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. *Frontiers in Immunology*, **2021**, 12, 669943	8.4	2
32	Life-Saving, Dose-Adjusted, Targeted Therapy in a Patient with a STAT3 Gain-of-Function Mutation. *Journal of Clinical Immunology*, **2021**, 41, 807-810	5.7	2
31	Combined immune deficiencies (CIDs) **2020**, 207-268		1
30	A 1-Year Prospective French Nationwide Study of Emergency Hospital Admissions in Children and Adults with Primary Immunodeficiency. *Journal of Clinical Immunology*, **2019**, 39, 702-712	5.7	1
29	Prédisposition génétique aux maladies infectieuses chez l'Homme. *Immuno-Analyse Et Biologie Specialisee*, **2013**, 28, 208-215		1
28	Human monogenic disorders that confer predisposition to specific infections. *Novartis Foundation Symposium*, **2007**, 281, 65-73; discussion 73-8, 208-9		1
27	Abatacept is useful in auto-immune cytopenia with immunopathologic manifestations caused by CTLA-4 defects. *Blood*, **2021**,	2.2	1
26	Inherited disorders of IFN-γ, IFN-γ/IL-12 and NF-κB-mediated immunity **2013**, 454-464		1
25	Two Monogenetic Disorders, Activated PI3-Kinase-δ Syndrome 2 and Smith-Magenis Syndrome, in One Patient: Case Report and a Literature Review of Neurodevelopmental Impact in Primary Immunodeficiencies Associated With Disturbed PI3K Signaling. *Frontiers in Pediatrics*, **2021**, 9, 688022	3.4	1
24	NLRC4 GOF Mutations, a Challenging Diagnosis from Neonatal Age to Adulthood. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
23	Long term follow-up of pediatric-onset Evans syndrome: broad immunopathological manifestations and high treatment burden. *Haematologica*, **2021**,	6.6	1
22	Variant of X-Linked Chronic Granulomatous Disease Revealed by a Severe Burkholderia cepacia Invasive Infection in an Infant. *Case Reports in Immunology*, **2013**, 2013, 323614	1.9	0
21	Identification of Germline Non-coding Deletions in XIAP Gene Causing XIAP Deficiency Reveals a Key Promoter Sequence.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
20	Gain-of-function variants in humans cause immune dysregulation associated with abnormal T/B cell late differentiation.. *Science Immunology*, **2022**, 7, eabi7160	28	0

19	Activated PI3-kinase Syndrome: Long-term Follow-up after Cord Blood Transplantation. *Journal of Clinical Immunology*, **2016**, 36, 544-6	5.7
18	Long-term follow-up of an activated PI3K-syndrome 2 in patient presenting with an agammaglobulinemia phenotype. *Annals of Allergy, Asthma and Immunology*, **2018**, 121, 739-740.e1	3.2
17	Immunodeficiencies at the Interface of Innate and Adaptive Immunity **2019**, 509-522.e1	
16	Life-threatening pulmonary interstitial lung disease complicating pediatric nonhumoral immunodeficiencies. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 2456-2458.e4	5.4
15	Le contrle gntique des maladies infectieuses : des lois de Mendel au squenage de lexome. *Bulletin De LAcademie Nationale De Medecine*, **2013**, 197, 157-171	0.1
14	Syndromes dactivation lymphohistiocytaire constitutionnels. *Revue DOncologie Hmatologie Pdiatrique*, **2013**, 1, 104-110	
13	Immunit inne Dficits immunitaires et infections pneumocoques. *Reanimation: Journal De La Societe De Reanimation De Langue Francaise*, **2011**, 20, 464-465	
12	Infections Streptococcus pneumoniae et Staphylococcus aureus : quels sont les dficits immunitaires hrditaires rechercher ?. *Revue Francophone Des Laboratoires*, **2008**, 2008, 91-95	0
11	Rapid and Safe T Cell Immune Reconstitution By T Cell Progenitor Injection Following Haploidentical Transplantation for Severe Combined Immunodeficiency (SCID). *Blood*, **2021**, 138, 1752-1752	2.2
10	Inherited disorders of IFN- IFN- and NF-B-mediated immunity **2008**, 553-560	
9	Peculiar hyper-IgM syndrome. Case report / Sindrom hiper-IgM atipic. Prezentare de caz. *Romanian Journal of Laboratory Medicine*, **2015**, 23, 341-345	0.3
8	Defects in Intrinsic and Innate Immunity: Receptors and Signaling Components **2017**, 339-392	
7	Complementation of a pathogenicIFNGR2misfolding mutation with modifiers of N-glycosylation. *Journal of Cell Biology*, **2008**, 182, i6-i6	7.3
6	Major Histocompatibility Complex Class II Deficiency **2016**, 378-390	
5	Mendelian Susceptibility to Infections with Viruses, Mycobacteria, Bacteria, and Candida **2016**, 407-415	
4	A New Missense Mutation in CD79B Leads to Autosomal Recessive Agammaglobulinemia in Two Siblings. *Journal of Clinical Immunology*, **2021**, 41, 1356-1360	5.7
3	Investigation of primary immune deficiency after severe bacterial infection in children: A population-based study in western France. *Archives De Pediatrie*, **2021**, 28, 398-404	1.8
2	A very uncommon cause of acute kidney injury in infancy. *Kidney International*, **2021**, 100, 948-950	9.9

1 Bayesian Modeling Immune Reconstitution Apply to CD34+ Selected Stem Cell Transplantation for Severe Combined Immunodeficiency.. *Frontiers in Pediatrics*, **2021**, 9, 804912 3.4