Mirjam van der Burg

List of Publications by Year
in descending order

208	10,475	28274	42399
papers	citations	55 h-index	92 g-index

213	213	213	12322
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Curation and expansion of Human Phenotype Ontology for defined groups of inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2022, 149, 369-378.	2.9	16
2	Hematopoietic stem cell transplantation in a patient with proteasome-associated autoinflammatory syndrome (PRAAS). Journal of Allergy and Clinical Immunology, 2022, 149, 1120-1127.e8.	2.9	11
3	Recommendations for uniform definitions used in newborn screening for severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2022, 149, 1428-1436.	2.9	19
4	Abnormal Results of Newborn Screening for SCID After Azathioprine Exposure In Utero: Benefit of TPMT Genotyping in Both Mother and Child. Journal of Clinical Immunology, 2022, 42, 199-202.	3.8	6
5	Long-Term Follow-Up of Newborns with 22q11 Deletion Syndrome and Low TRECs. Journal of Clinical Immunology, 2022, 42, 618-633.	3.8	9
6	A Pitfall of Whole Exome Sequencing: Variants in the 5â€²UTR Splice Site of BTK Causing XLA. Journal of Clinical Immunology, 2022, , 1.	3.8	0
7	Lessons learned from the diagnostic work-up of a patient with the bare lymphocyte syndrome type II. Clinical Immunology, 2022, 235, 108932.	3.2	2
8	Editorial: New Insights Into B Cell Subsets in Health and Disease. Frontiers in Immunology, 2022, 13, 854889.	4.8	3
9	AKT Hyperphosphorylation and T Cell Exhaustion in Down Syndrome. Frontiers in Immunology, 2022, 13, 724436.	4.8	3
10	Functional and Immune Modulatory Characteristics of Bone Marrow Mesenchymal Stromal Cells in Patients With Aplastic Anemia: A Systematic Review. Frontiers in Immunology, 2022, 13, 859668.	4.8	5
11	A novel digital PCR-based method to quantify (switched) B cells reveals the extent of allelic involvement in different recombination processes in the IGH locus. Molecular Immunology, 2022, 145, 109-123.	2.2	3
12	CD45RB Glycosylation and Ig Isotype Define Maturation of Functionally Distinct B Cell Subsets in Human Peripheral Blood. Frontiers in Immunology, 2022, 13, 891316.	4.8	6
13	Diagnostic Value of a Protocolized In-Depth Evaluation of Pediatric Bone Marrow Failure: A Multi-Center Prospective Cohort Study. Frontiers in Immunology, 2022, 13, 883826.	4.8	4
14	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	3.2	14
15	Protein functionality as a potential bottleneck for somatic revertant variants. Journal of Allergy and Clinical Immunology, 2021, 147, 391-393.e8.	2.9	3
16	Parentsâ€™ Perspectives and Societal Acceptance of Implementation of Newborn Screening for SCID in the Netherlands. Journal of Clinical Immunology, 2021, 41, 99-108.	3.8	25
17	Immunodeficiencies affecting cellular and humoral immunity. , 2021, , 9-39.		1
18	ATM: Translating the DNA Damage Response to Adaptive Immunity. Trends in Immunology, 2021, 42, 350-365.	6.8	22

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19	Modeling Influencing Factors in B-Cell Reconstitution After Hematopoietic Stem Cell Transplantation in Children. Frontiers in Immunology, 2021, 12, 684147.	4.8	7
20	Normal Numbers of Stem Cell Memory T Cells Despite Strongly Reduced Naive T Cells Support Intact Memory T Cell Compartment in Ataxia Telangiectasia. Frontiers in Immunology, 2021, 12, 686333.	4.8	4
21	Need for Uniform Definitions in Newborn Screening for SCID: The Next Challenge for Screeners and Immunologists. International Journal of Neonatal Screening, 2021, 7, 52.	3.2	0
22	Second Tier Testing to Reduce the Number of Non-actionable Secondary Findings and False-Positive Referrals in Newborn Screening for Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2021, 41, 1762-1773.	3.8	10
23	Economic Evaluation of Different Screening Strategies for Severe Combined Immunodeficiency Based on Real-Life Data. International Journal of Neonatal Screening, 2021, 7, 60.	3.2	6
24	Considerations for radiotherapy in Bloom Syndrome: A case series. European Journal of Medical Genetics, 2021, 64, 104293.	1.3	1
25	The expansion of human T-bet ^{high} CD21 ^{low} B cells is T cell dependent. Science Immunology, 2021, 6, eabh0891.	11.9	82
26	Future Perspectives of Newborn Screening for Inborn Errors of Immunity. International Journal of Neonatal Screening, 2021, 7, 74.	3.2	8
27	Primary Ovarian Failure in Addition to Classical Clinical Features of Coats Plus Syndrome in a Female Carrying 2 Truncating Variants of CTC1. Hormone Research in Paediatrics, 2021, 94, 448-455.	1.8	3
28	Early diagnosis of ataxia telangiectasia in the neonatal phase: a parentsâ€™ perspective. European Journal of Pediatrics, 2020, 179, 251-256.	2.7	11
29	The Phenotypic Spectrum of PNKP-Associated Disease and the Absence of Immunodeficiency and Cancer Predisposition in a Dutch Cohort. Pediatric Neurology, 2020, 113, 26-32.	2.1	6
30	Loss of ZBTB24 impairs nonhomologous end-joining and class-switch recombination in patients with ICF syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	27
31	Development of adaptive immune cells and receptor repertoires from infancy to adulthood. Current Opinion in Systems Biology, 2020, 24, 51-55.	2.6	3
32	The Clinical and Genetic Spectrum of 82 Patients With RAG Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries. Frontiers in Immunology, 2020, 11, 900.	4.8	16
33	iPSC-Based Modeling of RAG2 Severe Combined Immunodeficiency Reveals Multiple T Cell Developmental Arrests. Stem Cell Reports, 2020, 14, 300-311.	4.8	18
34	Defective formation of IgA memory B cells, Th1 and Th17 cells in symptomatic patients with selective IgA deficiency. Clinical and Translational Immunology, 2020, 9, e1130.	3.8	17
35	Rapid Low-Cost Microarray-Based Genotyping for Genetic Screening in Primary Immunodeficiency. Frontiers in Immunology, 2020, 11, 614.	4.8	21
36	Successful Preclinical Development of Gene Therapy for Recombinase-Activating Gene-1-Deficient SCID. Molecular Therapy - Methods and Clinical Development, 2020, 17, 666-682.	4.1	37

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37	EuroFlow Standardized Approach to Diagnostic Immunopheneotyping of Severe PID in Newborns and Young Children. Frontiers in Immunology, 2020, 11, 371.	4.8	17
38	Editorial: Application of Cytometry in Primary Immunodeficiencies. Frontiers in Immunology, 2020, 11, 463.	4.8	4
39	Dissection of the Pre-Germinal Center B-Cell Maturation Pathway in Common Variable Immunodeficiency Based on Standardized Flow Cytometric EuroFlow Tools. Frontiers in Immunology, 2020, 11, 603972.	4.8	13
40	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. Journal of Allergy and Clinical Immunology, 2019, 143, 726-735.	2.9	39
41	The presence of CLL-associated stereotypic B cell receptors in the normal BCR repertoire from healthy individuals increases with age. Immunity and Ageing, 2019, 16, 22.	4.2	17
42	Universal Newborn Screening for Severe Combined Immunodeficiency (SCID). Frontiers in Pediatrics, 2019, 7, 373.	1.9	82
43	Repertoire Sequencing of B Cells Elucidates the Role of UNG and Mismatch Repair Proteins in Somatic Hypermutation in Humans. Frontiers in Immunology, 2019, 10, 1913.	4.8	9
44	Precursor B-cell development in bone marrow of Good syndrome patients. Clinical Immunology, 2019, 200, 39-42.	3.2	14
45	An essential role for the Zn2+ transporter ZIP7 in B cell development. Nature Immunology, 2019, 20, 350-361.	14.5	92
46	EuroFlow-Based Flowcytometric Diagnostic Screening and Classification of Primary Immunodeficiencies of the Lymphoid System. Frontiers in Immunology, 2019, 10, 1271.	4.8	43
47	Impaired CpG Demethylation in Common Variable Immunodeficiency Associates With B Cell Phenotype and Proliferation Rate. Frontiers in Immunology, 2019, 10, 878.	4.8	19
48	B Cell Reconstitution and Influencing Factors After Hematopoietic Stem Cell Transplantation in Children. Frontiers in Immunology, 2019, 10, 782.	4.8	36
49	Defects in memory B-cell and plasma cell subsets expressing different immunoglobulin-subclasses in patients with CVID and immunoglobulin subclass deficiencies. Journal of Allergy and Clinical Immunology, 2019, 144, 809-824.	2.9	55
50	Cost-effectiveness of newborn screening for severe combined immunodeficiency. European Journal of Pediatrics, 2019, 178, 721-729.	2.7	19
51	Delineating Human B Cell Precursor Development With Genetically Identified PID Cases as a Model. Frontiers in Immunology, 2019, 10, 2680.	4.8	14
52	Dilemma of Reporting Incidental Findings in Newborn Screening Programs for SCID: Parentsâ€™ Perspective on Ataxia Telangiectasia. Frontiers in Immunology, 2019, 10, 2438.	4.8	19
53	<scp>IVI</scp>gâ€induced plasmablasts in patients with GuillainâBarrÃ© syndrome. Annals of Clinical and Translational Neurology, 2019, 6, 129-143.	3.7	12
54	A 3-Year-Old Girl With a Mediastinal Mass. Chest, 2019, 155, e13-e16.	0.8	0

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55	Phenotypical heterogeneity in RAG-deficient patients from a highly consanguineous population. Clinical and Experimental Immunology, 2019, 195, 202-212.	2.6	22
56	Selection and validation of antibody clones against IgG and IgA subclasses in switched memory B-cells and plasma cells. Journal of Immunological Methods, 2019, 475, 112372.	1.4	17
57	Optimization and testing of dried antibody tube: The EuroFlow LST and PIDOT tubes as examples. Journal of Immunological Methods, 2019, 475, 112287.	1.4	29
58	Polymerase Î´ deficiency causes syndromic immunodeficiency with replicative stress. Journal of Clinical Investigation, 2019, 129, 4194-4206.	8.2	41
59	The EuroFlow PID Orientation Tube for Flow Cytometric Diagnostic Screening of Primary Immunodeficiencies of the Lymphoid System. Frontiers in Immunology, 2019, 10, 246.	4.8	100
60	Class-Switch Recombination Defects. Rare Diseases of the Immune System, 2019, , 179-199.	0.1	0
61	IgM Augments Complement Bactericidal Activity with Serum from a Patient with a Novel CD79a Mutation. Journal of Clinical Immunology, 2018, 38, 185-192.	3.8	16
62	Adaptive antibody diversification through N -linked glycosylation of the immunoglobulin variable region. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1901-1906.	7.1	98
63	A kindred with mutant IKAROS and autoimmunity. Journal of Allergy and Clinical Immunology, 2018, 142, 699-702.e12.	2.9	39
64	The TH1 phenotype of follicular helper T cells indicates an IFN-Î³â€“associated immune dysregulation in patients with CD21low common variable immunodeficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 730-740.	2.9	109
65	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. Journal of Allergy and Clinical Immunology, 2018, 141, 408-411.e8.	2.9	6
66	Identification of CVID Patients With Defects in Immune Repertoire Formation or Specification. Frontiers in Immunology, 2018, 9, 2545.	4.8	38
67	Introducing Newborn Screening for Severe Combined Immunodeficiency (SCID) in the Dutch Neonatal Screening Program. International Journal of Neonatal Screening, 2018, 4, 40.	3.2	30
68	No Overt Clinical Immunodeficiency Despite Immune Biological Abnormalities in Patients With Constitutional Mismatch Repair Deficiency. Frontiers in Immunology, 2018, 9, 1506.	4.8	24
69	Deficiencies in the CD19 complex. Clinical Immunology, 2018, 195, 82-87.	3.2	17
70	Exhaustion of the CD8+ T Cell Compartment in Patients with Mutations in Phosphoinositide 3-Kinase Delta. Frontiers in Immunology, 2018, 9, 446.	4.8	52
71	Mutations affecting the actin regulator WD repeatâ€“containing protein 1 lead to aberrant lymphoid immunity. Journal of Allergy and Clinical Immunology, 2018, 142, 1589-1604.e11.	2.9	64
72	Genetic defects in PI3KÎ´ affect B-cell differentiation and maturation leading to hypogammaglobulineamia and recurrent infections. Clinical Immunology, 2017, 176, 77-86.	3.2	80

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73	A novel mutation in TAP1 gene leading to MHC class I deficiency: Report of two cases and review of the literature. Clinical Immunology, 2017, 178, 74-78.	3.2	31
74	Antigen Receptor Galaxy: A User-Friendly, Web-Based Tool for Analysis and Visualization of T and B Cell Receptor Repertoire Data. Journal of Immunology, 2017, 198, 4156-4165.	0.8	52
75	An evaluation of the TREC assay with regard to the integration of SCID screening into the Dutch newborn screening program. Clinical Immunology, 2017, 180, 106-110.	3.2	41
76	T and B Cell Markers in Dried Blood Spots of Neonates with Congenital Cytomegalovirus Infection: B Cell Numbers at Birth Are Associated with Long-Term Outcomes. Journal of Immunology, 2017, 198, 102-109.	0.8	9
77	Human IgG2â€•and IgG4â€expressing memory B cells display enhanced molecular and phenotypic signs of maturity and accumulate with age. Immunology and Cell Biology, 2017, 95, 744-752.	2.3	49
78	Circulating T Cells of Patients with Nijmegen Breakage Syndrome Show Signs of Senescence. Journal of Clinical Immunology, 2017, 37, 133-142.	3.8	13
79	Low T Cell Numbers Resembling Tâ˜B+ SCID in a Patient with Wiskottâ€“Aldrich Syndrome and the Outcome of Two Hematopoietic Stem Cell Transplantations. Journal of Clinical Immunology, 2017, 37, 18-21.	3.8	0
80	T-cell receptor sequencing reveals decreased diversity 18Âyears after early thymectomy. Journal of Allergy and Clinical Immunology, 2017, 140, 1743-1746.e7.	2.9	6
81	Public Clonotypes and Convergent Recombination Characterize the NaÃ¯ve CD8+ T-Cell Receptor Repertoire of Extremely Preterm Neonates. Frontiers in Immunology, 2017, 8, 1859.	4.8	25
82	Combined immunodeficiencies: twenty years experience from a single center in Turkey. Central-European Journal of Immunology, 2016, 1, 107-115.	1.2	20
83	Evaluation of the Antigen-Experienced B-Cell Receptor Repertoire in Healthy Children and Adults. Frontiers in Immunology, 2016, 7, 410.	4.8	53
84	Changes in Healthy Human IgG Fc-Glycosylation after Birth and during Early Childhood. Journal of Proteome Research, 2016, 15, 1853-1861.	3.7	91
85	XLF deficiency results in reduced N-nucleotide addition during V(D)J recombination. Blood, 2016, 128, 650-659.	1.4	33
86	Increased PI3K/Akt activity and deregulated humoral immune response in human PTEN deficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 1744-1747.e5.	2.9	52
87	Disturbed B-lymphocyte selection in autoimmune lymphoproliferative syndrome. Blood, 2016, 127, 2193-2202.	1.4	25
88	Decreased IL7RÎ± and TdT expression underlie the skewed immunoglobulin repertoire of human B-cell precursors from fetal origin. Scientific Reports, 2016, 6, 33924.	3.3	20
89	Adaptive immune defects in a patient with leukocyte adhesion deficiency type <scp>III</scp> with a novel mutation in <scp>FERMT</scp>3. Pediatric Allergy and Immunology, 2016, 27, 214-217.	2.6	18
90	An infant with ZAP-70 deficiency with disseminated mycobacterial disease. Journal of Clinical Immunology, 2016, 36, 103-106.	3.8	11

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91	Identification of checkpoints in human T-cell development using severe combined immunodeficiency stem cells. Journal of Allergy and Clinical Immunology, 2016, 137, 517-526.e3.	2.9	26
92	Decreased somatic hypermutation induces an impaired peripheral B cell tolerance checkpoint. Journal of Clinical Investigation, 2016, 126, 4289-4302.	8.2	46
93	Three faces of recombination activating gene 1 (RAG1) mutations. Acta Microbiologica Et Immunologica Hungarica, 2015, 62, 393-401.	0.8	6
94	Autosomal recessive hyper IgM syndrome associated with activation-induced cytidine deaminase gene in three Turkish siblings presented with tuberculosis lymphadenitis â€" Case report. Acta Microbiologica Et Immunologica Hungarica, 2015, 62, 267-274.	0.8	3
95	Persistent subclinical immune defects in HIV-1-infected children treated with antiretroviral therapy. Aids, 2015, 29, 1745-1756.	2.2	9
96	Strategies for B-Cell Receptor Repertoire Analysis in Primary Immunodeficiencies: From Severe Combined Immunodeficiency to Common Variable Immunodeficiency. Frontiers in Immunology, 2015, 6, 157.	4.8	20
97	Immune Dysfunction in Children with CHARGE Syndrome: A Cross-Sectional Study. PLoS ONE, 2015, 10, e0142350.	2.5	27
98	Immunodeficiency in a Child with Rapadilino Syndrome: A Case Report and Review of the Literature. Case Reports in Immunology, 2015, 2015, 1-4.	0.4	3
99	CD21 and CD19 deficiency: Two defects in the same complex leading to different disease modalities. Clinical Immunology, 2015, 161, 120-127.	3.2	42
100	Mutations in Brutonâ€™s tyrosine kinase impair IgA responses. International Journal of Hematology, 2015, 101, 305-313.	1.6	19
101	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71
102	A case of XLF deficiency presented with diffuse large B cell lymphoma in the brain. Clinical Immunology, 2015, 161, 394-395.	3.2	7
103	Key stages of bone marrow B-cell maturation are defective in patients with common variable immunodeficiency disorders. Journal of Allergy and Clinical Immunology, 2015, 136, 487-490.e2.	2.9	20
104	Silent brain infarcts in two patients with zeta chain-associated protein 70kDa (ZAP70) deficiency. Clinical Immunology, 2015, 158, 88-91.	3.2	16
105	Novel mutations in TNFRSF7/CD27: Clinical, immunologic, and genetic characterization of human CD27 deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 703-712.e10.	2.9	109
106	B-cell reconstitution after lentiviral vectorâ€"mediated gene therapy in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2015, 136, 692-702.e2.	2.9	41
107	Overview of 15-year severe combined immunodeficiency in the Netherlands: towards newborn blood spot screening. European Journal of Pediatrics, 2015, 174, 1183-1188.	2.7	16
108	TREC Based Newborn Screening for Severe Combined Immunodeficiency Disease: A Systematic Review. Journal of Clinical Immunology, 2015, 35, 416-430.	3.8	140

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109	Functional analysis of naturally occurring DCLRE1C mutations and correlation with the clinical phenotype of ARTEMIS deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 140-150.e7.	2.9	63
110	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulatorâ€“dependent autoimmunity. Journal of Allergy and Clinical Immunology, 2015, 135, 1578-1588.e5.	2.9	84
111	DNA-PKcs Is Involved in Ig Class Switch Recombination in Human B Cells. Journal of Immunology, 2015, 195, 5608-5615.	0.8	30
112	The 11q Terminal Deletion Disorder Jacobsen Syndrome is a Syndromic Primary Immunodeficiency. Journal of Clinical Immunology, 2015, 35, 761-768.	3.8	25
113	Clinical Spectrum of SCID: The Key is in the Thymus?. Frontiers in Immunology, 2014, 5, 111.	4.8	1
114	Unraveling the Repertoire in WiskottÃ¢â‚¬â€œAldrich Syndrome. Frontiers in Immunology, 2014, 5, 539.	4.8	7
115	<scp>CD3G</scp> Gene Defects in Familial Autoimmune Thyroiditis. Scandinavian Journal of Immunology, 2014, 80, 354-361.	2.7	20
116	Defective B-cell memory in patients with Down syndrome. Journal of Allergy and Clinical Immunology, 2014, 134, 1346-1353.e9.	2.9	53
117	ImmunoGlobulin galaxy (IGGalaxy) for simple determination and quantitation of immunoglobulin heavy chain rearrangements from NGS. BMC Immunology, 2014, 15, 59.	2.2	30
118	Human CD19 and CD40L deficiencies impair antibody selection and differentially affect somatic hypermutation. Journal of Allergy and Clinical Immunology, 2014, 134, 135-144.e7.	2.9	71
119	B-cell development and functions and therapeutic options in adenosine deaminaseâ€“deficient patients. Journal of Allergy and Clinical Immunology, 2014, 133, 799-806.e10.	2.9	30
120	Targeted next-generation sequencing: AÂnovel diagnostic tool for primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2014, 133, 529-534.e1.	2.9	143
121	Targeted genome editing in human repopulating haematopoietic stem cells. Nature, 2014, 510, 235-240.	27.8	517
122	Primary Immune Deficiency Treatment Consortium (PIDTC) report. Journal of Allergy and Clinical Immunology, 2014, 133, 335-347.e11.	2.9	65
123	Similar recombination-activating gene (RAG) mutations result in similar immunobiological effects but in different clinical phenotypes. Journal of Allergy and Clinical Immunology, 2014, 133, 1124-1133.e1.	2.9	71
124	Human syndromes of immunodeficiency and dysregulation are characterized by distinct defects in T-cell receptor repertoire development. Journal of Allergy and Clinical Immunology, 2014, 133, 1109-1115.e14.	2.9	62
125	Quantifying independent risk factors for failing to rescreen in a breast cancer screening program in Flanders, Belgium. Preventive Medicine, 2014, 69, 280-286.	3.4	12
126	The value of DNA storage and pedigree analysis in rare diseases: a 17-year-old boy with X-linked lymphoproliferative disease (XLP) caused by a de novo SH2D1A mutation. European Journal of Pediatrics, 2014, 173, 1695-1698.	2.7	6

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127	Levels of somatic hypermutations in B cell receptors increase during childhood. Clinical and Experimental Immunology, 2014, 178, 394-398.	2.6	17
128	Human IgE+ B cells are derived from T cellâ€“dependent and T cellâ€“independent pathways. Journal of Allergy and Clinical Immunology, 2014, 134, 688-697.e6.	2.9	79
129	Wiskottâ€“Aldrich Syndrome protein deficiency perturbs the homeostasis of B-cell compartment in humans. Journal of Autoimmunity, 2014, 50, 42-50.	6.5	72
130	Differential role of nonhomologous end joining factors in the generation, DNA damage response, and myeloid differentiation of human induced pluripotent stem cells. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 8889-8894.	7.1	34
131	Antibody deficiency in patients with ataxia telangiectasia is caused by disturbed B- and T-cell homeostasis and reduced immune repertoire diversity. Journal of Allergy and Clinical Immunology, 2013, 131, 1367-1375.e9.	2.9	107
132	No significant prognostic value of normal precursor <scp>B</scp>â€cell regeneration in paediatric acute myeloid leukaemia after induction treatment. British Journal of Haematology, 2013, 161, 861-864.	2.5	6
133	Categorizing B-cell defects using an in-vitro B-cell differentiation assay. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 61-61.	0.0	0
134	Applicability of a reproducible flow cytometry scoring system in the diagnosis of refractory cytopenia of childhood. Leukemia, 2013, 27, 1923-1925.	7.2	20
135	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	2.8	115
136	Clinical Spectrum of LIG 4 Deficiency Is Broadened with Severe Dysmaturity, Primordial Dwarfism, and Neurological Abnormalities. Human Mutation, 2013, 34, 1611-1614.	2.5	34
137	Wiskott-Aldrich syndrome proteinâ€“mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. Journal of Experimental Medicine, 2013, 210, 355-374.	8.5	49
138	A reversion of an IL2RG mutation in combined immunodeficiency providing competitive advantage to the majority of CD8+ T cells. Haematologica, 2013, 98, 1030-1038.	3.5	48
139	Anti-TNF treatment blocks the induction of T cell-dependent humoral responses. Annals of the Rheumatic Diseases, 2013, 72, 1037-1043.	0.9	94
140	Common variable immunodeficiency and idiopathic primary hypogammaglobulinemia: two different conditions within the same disease spectrum. Haematologica, 2013, 98, 1617-1623.	3.5	67
141	A Girl with Autoimmune Cytopenias, Nonmalignant Lymphadenopathy, and Recurrent Infections. Case Reports in Immunology, 2012, 2012, 1-6.	0.4	0
142	The defect in humoral immunity in patients with Nijmegen breakage syndrome is explained by defects in peripheral B lymphocyte maturation. Cytometry Part A: the Journal of the International Society for Analytical Cytology, 2012, 81A, 835-842.	1.5	26
143	The EuroChimerism concept for a standardized approach to chimerism analysis after allogeneic stem cell transplantation. Leukemia, 2012, 26, 1821-1828.	7.2	83
144	Two SCID cases with Cernunnosâ€XLF deficiency successfully treated by hematopoietic stem cell transplantation. Pediatric Transplantation, 2012, 16, E167-71.	1.0	22

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145	New frontiers of primary antibody deficiencies. Cellular and Molecular Life Sciences, 2012, 69, 59-73.	5.4	22
146	B-cell replication history and somatic hypermutation status identify distinct pathophysiologic backgrounds in common variable immunodeficiency. Blood, 2011, 118, 6814-6823.	1.4	112
147	Standardization of DNA isolation from low cell numbers for chimerism analysis by PCR of short tandem repeats. Leukemia, 2011, 25, 1467-1470.	7.2	40
148	Artemis splice defects cause atypical SCID and can be restored in vitro by an antisense oligonucleotide. Genes and Immunity, 2011, 12, 434-444.	4.1	27
149	Idiopathic CD4+ T lymphopenia without autoimmunity or granulomatous disease in the slipstream of RAG mutations. Blood, 2011, 117, 5892-5896.	1.4	107
150	Ageâ€matched Reference Values for Bâ€lymphocyte Subpopulations and CVID Classifications in Children. Scandinavian Journal of Immunology, 2011, 74, 502-510.	2.7	72
151	Checkpoints of B cell differentiation: visualizing Igâ€centric processes. Annals of the New York Academy of Sciences, 2011, 1246, 11-25.	3.8	23
152	Genetic characteristics of eighty-seven patients with the Wiskottâ€“Aldrich syndrome. Molecular Immunology, 2011, 48, 788-792.	2.2	35
153	Dissection of B-Cell Development to Unravel Defects in Patients with a Primary Antibody Deficiency. Advances in Experimental Medicine and Biology, 2011, 697, 183-196.	1.6	10
154	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. American Journal of Human Genetics, 2011, 88, 796-804.	6.2	158
155	Educational paper. European Journal of Pediatrics, 2011, 170, 561-571.	2.7	125
156	Educational paper. European Journal of Pediatrics, 2011, 170, 693-702.	2.7	52
157	PID Comes Full Circle: Applications of V(D)J Recombination Excision Circles in Research, Diagnostics and Newborn Screening of Primary Immunodeficiency Disorders. Frontiers in Immunology, 2011, 2, 12.	4.8	62
158	Antibody deficiency due to a missense mutation in CD19 demonstrates the importance of the conserved tryptophan 41 in immunoglobulin superfamily domain formation. Human Molecular Genetics, 2011, 20, 1854-1863.	2.9	31
159	The human syndrome of dendritic cell, monocyte, B and NK lymphoid deficiency. Journal of Experimental Medicine, 2011, 208, 227-234.	8.5	277
160	Loss of juxtaposition of RAG-induced immunoglobulin DNA ends is implicated in the precursor B-cell differentiation defect in NBS patients. Blood, 2010, 115, 4770-4777.	1.4	37
161	Late-onset adenosine deaminase deficiency presenting with Heckâ€™s disease. European Journal of Pediatrics, 2010, 169, 1033-1036.	2.7	14
162	A novel radiosensitive SCID patient with a pronounced G2/M sensitivity. DNA Repair, 2010, 9, 365-373.	2.8	3

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163	B-cell maturation and antibody responses in individuals carrying a mutated CD19 allele. Genes and Immunity, 2010, 11, 523-530.	4.1	34
164	Homeostatic expansion of autoreactive immunoglobulin-secreting cells in the Rag2 mouse model of Omenn syndrome. Journal of Experimental Medicine, 2010, 207, 1525-1540.	8.5	66
165	Decrease of Skin Infiltrating and Circulating CCR10+ T Cells Coincides with Clinical Improvement after Topical Tacrolimus in Omenn Syndrome. Journal of Investigative Dermatology, 2010, 130, 308-311.	0.7	1
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