

Anne Durandy

List of Publications by Year in descending order

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124
papers

14,386
citations

30070

54
h-index

21540

114
g-index

127
all docs

127
docs citations

127
times ranked

13861
citing authors

#	ARTICLE	IF	CITATIONS
1	Activation-Induced Cytidine Deaminase (AID) Deficiency Causes the Autosomal Recessive Form of the Hyper-IgM Syndrome (HIGM2). <i>Cell</i> , 2000, 102, 565-575.	28.9	1,489
2	Impairment of Mycobacterial Immunity in Human Interleukin-12 Receptor Deficiency. <i>Science</i> , 1998, 280, 1432-1435.	12.6	787
3	X-linked anhidrotic ectodermal dysplasia with immunodeficiency is caused by impaired NF- κ B signaling. <i>Nature Genetics</i> , 2001, 27, 277-285.	21.4	784
4	The RIDDLE Syndrome Protein Mediates a Ubiquitin-Dependent Signaling Cascade at Sites of DNA Damage. <i>Cell</i> , 2009, 136, 420-434.	28.9	673
5	Cernunnos, a Novel Nonhomologous End-Joining Factor, Is Mutated in Human Immunodeficiency with Microcephaly. <i>Cell</i> , 2006, 124, 287-299.	28.9	640
6	Human uracil-DNA glycosylase deficiency associated with profoundly impaired immunoglobulin class-switch recombination. <i>Nature Immunology</i> , 2003, 4, 1023-1028.	14.5	573
7	Efficacy of Gene Therapy for X-Linked Severe Combined Immunodeficiency. <i>New England Journal of Medicine</i> , 2010, 363, 355-364.	27.0	561
8	Phosphoinositide 3-Kinase $\hat{\gamma}$ Gene Mutation Predisposes to Respiratory Infection and Airway Damage. <i>Science</i> , 2013, 342, 866-871.	12.6	541
9	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the Clinical Diagnosis of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1763-1770.	3.8	381
10	Clinical spectrum and features of activated phosphoinositide 3-kinase $\hat{\gamma}$ syndrome: A large patient cohort study. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 597-606.e4.	2.9	377
11	ICOS Deficiency Is Associated with a Severe Reduction of CXCR5+CD4 Germinal Center Th Cells. <i>Journal of Immunology</i> , 2006, 177, 4927-4932.	0.8	349
12	Human TRAF3 Adaptor Molecule Deficiency Leads to Impaired Toll-like Receptor 3 Response and Susceptibility to Herpes Simplex Encephalitis. <i>Immunity</i> , 2010, 33, 400-411.	14.3	304
13	AID mutant analyses indicate requirement for class-switch-specific cofactors. <i>Nature Immunology</i> , 2003, 4, 843-848.	14.5	301
14	Autosomal Dominant STAT3 Deficiency and Hyper-IgE Syndrome. <i>Medicine (United States)</i> , 2012, 91, e1-e19.	1.0	274
15	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. <i>Journal of Experimental Medicine</i> , 2006, 203, 1745-1759.	8.5	264
16	Human ICOS deficiency abrogates the germinal center reaction and provides a monogenic model for common variable immunodeficiency. <i>Blood</i> , 2006, 107, 3045-3052.	1.4	254
17	A human immunodeficiency caused by mutations in the PIK3R1 gene. <i>Journal of Clinical Investigation</i> , 2014, 124, 3923-3928.	8.2	239
18	Clinical, immunologic and genetic analysis of 29 patients with autosomal recessive hyper-IgM syndrome due to Activation-Induced Cytidine Deaminase deficiency. <i>Clinical Immunology</i> , 2004, 110, 22-29.	3.2	224

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19	Clinical and immunologic phenotype associated with activated phosphoinositide 3-kinase $\hat{\gamma}$ syndrome 2: A cohort study. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 210-218.e9.	2.9	215
20	Primary antibody deficiencies. <i>Nature Reviews Immunology</i> , 2013, 13, 519-533.	22.7	214
21	Naive and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells. <i>Journal of Experimental Medicine</i> , 2013, 210, 2739-2753.	8.5	158
22	Human PMS2 deficiency is associated with impaired immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2008, 205, 2465-2472.	8.5	151
23	Retinoids Regulate Survival and Antigen Presentation by Immature Dendritic Cells. <i>Journal of Experimental Medicine</i> , 2003, 198, 623-634.	8.5	143
24	Severe combined immunodeficiency caused by deficiency in either the $\hat{\gamma}$ or the $\hat{\mu}$ subunit of CD3. <i>Journal of Clinical Investigation</i> , 2004, 114, 1512-1517.	8.2	141
25	The CARD11-BCL10-MALT1 (CBM) signalosome complex: Stepping into the limelight of human primary immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 276-284.	2.9	133
26	Mutational, functional, and expression studies of the <i>TCF4</i> gene in Pitt-Hopkins syndrome. <i>Human Mutation</i> , 2009, 30, 669-676.	2.5	126
27	Mutations in Activation-Induced Cytidine Deaminase in Patients with Hyper IgM Syndrome. <i>Clinical Immunology</i> , 2000, 97, 203-210.	3.2	125
28	IL-21 signalling via STAT3 primes human naïve B cells to respond to IL-2 to enhance their differentiation into plasmablasts. <i>Blood</i> , 2013, 122, 3940-3950.	1.4	121
29	Activation-induced cytidine deaminase (AID) is required for B-cell tolerance in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 11554-11559.	7.1	118
30	Hyper-IgM syndrome type 4 with a B lymphocyte intrinsic selective deficiency in Ig class-switch recombination. <i>Journal of Clinical Investigation</i> , 2003, 112, 136-142.	8.2	114
31	Analysis of class switch recombination and somatic hypermutation in patients affected with autosomal dominant hyper-IgM syndrome type 2. <i>Clinical Immunology</i> , 2005, 115, 277-285.	3.2	111
32	Defects of class-switch recombination. <i>Journal of Allergy and Clinical Immunology</i> , 2006, 117, 855-864.	2.9	107
33	Memory switched B cell percentage and not serum immunoglobulin concentration is associated with clinical complications in children and adults with specific antibody deficiency and common variable immunodeficiency. <i>Clinical Immunology</i> , 2006, 120, 310-318.	3.2	106
34	B cells from hyper-IgM patients carrying UNG mutations lack ability to remove uracil from ssDNA and have elevated genomic uracil. <i>Journal of Experimental Medicine</i> , 2005, 201, 2011-2021.	8.5	103
35	Occurrence of B-cell lymphomas in patients with activated phosphoinositide 3-kinase $\hat{\gamma}$ syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 233-236.e3.	2.9	101
36	Repair of U/G and U/A in DNA by UNG2-associated repair complexes takes place predominantly by short-patch repair both in proliferating and growth-arrested cells. <i>Nucleic Acids Research</i> , 2004, 32, 5486-5498.	14.5	92

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37	The NEMO Mutation Creating the Most-Upstream Premature Stop Codon Is Hypomorphic Because of a Reinitiation of Translation. <i>American Journal of Human Genetics</i> , 2006, 78, 691-701.	6.2	89
38	Induction by anti-CD40 antibody or soluble CD40 ligand and cytokines of IgG, IgA and IgE production by B cells from patients with X-linked hyper IgM syndrome. <i>European Journal of Immunology</i> , 1993, 23, 2294-2299.	2.9	87
39	Pathophysiology of B-Cell Intrinsic Immunoglobulin Class Switch Recombination Deficiencies. <i>Advances in Immunology</i> , 2007, 94, 275-306.	2.2	86
40	The Block in Immunoglobulin Class Switch Recombination Caused by Activation-Induced Cytidine Deaminase Deficiency Occurs Prior to the Generation of DNA Double Strand Breaks in Switch 1/4 Region. <i>Journal of Immunology</i> , 2003, 171, 2504-2509.	0.8	84
41	Polymerase β 1 mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature (â€œFELS syndromeâ€). <i>Journal of Experimental Medicine</i> , 2012, 209, 2323-2330.	8.5	83
42	Defined Blocks in Terminal Plasma Cell Differentiation of Common Variable Immunodeficiency Patients. <i>Journal of Immunology</i> , 2005, 175, 5498-5503.	0.8	81
43	Hyper-IgM syndromes. <i>Current Opinion in Rheumatology</i> , 2006, 18, 369-376.	4.3	79
44	Severe combined immunodeficiency caused by deficiency in either the γ or the μ subunit of CD3. <i>Journal of Clinical Investigation</i> , 2004, 114, 1512-1517.	8.2	78
45	Hyper-immunoglobulin M syndromes caused by intrinsic B-lymphocyte defects. <i>Immunological Reviews</i> , 2005, 203, 67-79.	6.0	76
46	Function of Apollo (SNM1B) at telomere highlighted by a splice variant identified in a patient with Hoyeraalâ€Hreidarsson syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 10097-10102.	7.1	76
47	Mini-review Activation-induced cytidine deaminase: a dual role in class-switch recombination and somatic hypermutation. <i>European Journal of Immunology</i> , 2003, 33, 2069-2073.	2.9	70
48	First Report of the Hyper-IgM Syndrome Registry of the Latin American Society for Immunodeficiencies: Novel Mutations, Unique Infections, and Outcomes. <i>Journal of Clinical Immunology</i> , 2014, 34, 146-156.	3.8	70
49	Activation-Induced Cytidine Deaminase Expression in Human B Cell Precursors Is Essential for Central B Cell Tolerance. <i>Immunity</i> , 2015, 43, 884-895.	14.3	69
50	Human MSH6 Deficiency Is Associated with Impaired Antibody Maturation. <i>Journal of Immunology</i> , 2012, 188, 2023-2029.	0.8	67
51	RANK-dependent autosomal recessive osteopetrosis: Characterization of five new cases with novel mutations. <i>Journal of Bone and Mineral Research</i> , 2012, 27, 342-351.	2.8	66
52	Signal transducer and activator of transcription 3 (STAT3) mutations underlying autosomal dominant hyper-IgE syndrome impair human CD8+ T-cell memory formation and function. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 400-411.e9.	2.9	63
53	Human genetic defects in class-switch recombination (hyper-IgM syndromes). <i>Current Opinion in Immunology</i> , 2001, 13, 543-548.	5.5	60
54	X-linked primary immunodeficiency associated with hemizygous mutations in the moesin (MSN) gene. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 1681-1689.e8.	2.9	60

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55	The mechanisms of immune diversification and their disorders. <i>Nature Reviews Immunology</i> , 2003, 3, 962-972.	22.7	59
56	Immunoglobulin class-switch recombination deficiencies. <i>Arthritis Research and Therapy</i> , 2012, 14, 218.	3.5	59
57	Activation-induced cytidine deaminase: structure–function relationship as based on the study of mutants. <i>Human Mutation</i> , 2006, 27, 1185-1191.	2.5	54
58	CD40 ligand expression deficiency in a female carrier of the X-linked hyper-IgM syndrome as a result of X chromosome lyonization. <i>European Journal of Immunology</i> , 1999, 29, 367-373.	2.9	52
59	Activation induced deaminase C-terminal domain links DNA breaks to end protection and repair during class switch recombination. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E988-97.	7.1	52
60	Hematopoietic stem cell transplantation for progressive combined immunodeficiency and lymphoproliferation in patients with activated phosphatidylinositol-3-OH kinase 1 syndrome type 1. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 266-275.	2.9	49
61	Immunoglobulin Replacement Therapy in Primary Antibody Deficiency Diseases – Maximizing Success. <i>International Archives of Allergy and Immunology</i> , 2005, 136, 217-229.	2.1	48
62	Defective anti-polysaccharide response and splenic marginal zone disorganization in ALPS patients. <i>Blood</i> , 2014, 124, 1597-1609.	1.4	48
63	A primary immunodeficiency characterized by defective immunoglobulin class switch recombination and impaired DNA repair. <i>Journal of Experimental Medicine</i> , 2007, 204, 1207-1216.	8.5	47
64	Protective effect of IgM against colonization of the respiratory tract by nontypeable <i>Haemophilus influenzae</i> in patients with hypogammaglobulinemia. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 129, 770-777.	2.9	47
65	Decreased somatic hypermutation induces an impaired peripheral B cell tolerance checkpoint. <i>Journal of Clinical Investigation</i> , 2016, 126, 4289-4302.	8.2	46
66	Cernunnos influences human immunoglobulin class switch recombination and may be associated with B cell lymphomagenesis. <i>Journal of Experimental Medicine</i> , 2012, 209, 291-305.	8.5	44
67	Mild B-cell lymphocytosis in patients with a CARD11 C49Y mutation. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 819-821.e1.	2.9	44
68	Primary Microcephaly, Impaired DNA Replication, and Genomic Instability Caused by Compound Heterozygous <i>ATR</i> Mutations. <i>Human Mutation</i> , 2013, 34, 374-384.	2.5	43
69	Human X-linked variable immunodeficiency caused by a hypomorphic mutation in XIAP in association with a rare polymorphism in CD40LG. <i>Blood</i> , 2011, 118, 252-261.	1.4	41
70	Restoration of Human B-cell Differentiation Into NOD-SCID Mice Engrafted With Gene-corrected CD34+ Cells Isolated From Artemis or RAG1-deficient Patients. <i>Molecular Therapy</i> , 2008, 16, 396-403.	8.2	39
71	Human Models of Inherited Immunoglobulin Class Switch Recombination and Somatic Hypermutation Defects (Hyper-IgM Syndromes). <i>Advances in Immunology</i> , 2004, 82, 295-330.	2.2	37
72	Potential roles of activation-induced cytidine deaminase in promotion or prevention of autoimmunity in humans. <i>Autoimmunity</i> , 2013, 46, 148-156.	2.6	37

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73	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 998-1007.e6.	2.9	37
74	Somatic Hypermutation Shapes the Antibody Repertoire of Memory B Cells in Humans. <i>Journal of Experimental Medicine</i> , 2001, 194, 375-378.	8.5	36
75	Insights into the B cell specific process of immunoglobulin class switch recombination. <i>Immunology Letters</i> , 2011, 138, 97-103.	2.5	36
76	Mutations in the adaptor-binding domain and associated linker region of p110 β cause Activated PI3K- δ Syndrome 1 (APDS1). <i>Haematologica</i> , 2017, 102, e278-e281.	3.5	36
77	Genetically acquired class-switch recombination defects: the multi-faced hyper-IgM syndrome. <i>Immunology Letters</i> , 2005, 97, 1-6.	2.5	35
78	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2013, 210, 2503-2513.	8.5	33
79	Late-onset combined immune deficiency associated to skin granuloma due to heterozygous compound mutations in RAG1 gene in a 14years old male. <i>Human Immunology</i> , 2013, 74, 18-22.	2.4	32
80	Loss of ARHGEF1 causes a human primary antibody deficiency. <i>Journal of Clinical Investigation</i> , 2019, 129, 1047-1060.	8.2	32
81	A homozygousPMS2founder mutation with an attenuated constitutional mismatch repair deficiency phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 348-352.	3.2	30
82	Increased activation of PI3 kinase- δ predisposes to B-cell lymphoma. <i>Blood</i> , 2020, 135, 638-643.	1.4	29
83	Characterization of Ig Gene Somatic Hypermutation in the Absence of Activation-Induced Cytidine Deaminase. <i>Journal of Immunology</i> , 2008, 181, 1299-1306.	0.8	27
84	Normal CD40-mediated activation of monocytes and dendritic cells from patients with hyper-IgM syndrome due to a CD40 pathway defect in B cells. <i>European Journal of Immunology</i> , 1998, 28, 3648-3654.	2.9	25
85	A Syndrome Involving Intrauterine Growth Retardation, Microcephaly, Cerebellar Hypoplasia, B Lymphocyte Deficiency, and Progressive Pancytopenia. <i>Pediatrics</i> , 2000, 105, e39-e39.	2.1	25
86	Genomic Spectrum and Phenotypic Heterogeneity of Human IL-21 Receptor Deficiency. <i>Journal of Clinical Immunology</i> , 2021, 41, 1272-1290.	3.8	25
87	Ataxia-telangiectasia in twins presenting as autosomal recessive hyper-immunoglobulin M syndrome. <i>Israel Medical Association Journal</i> , 2007, 9, 406-7.	0.1	25
88	A severe form of abetalipoproteinemia caused by new splicing mutations of microsomal triglyceride transfer protein (MTTP). <i>Human Mutation</i> , 2011, 32, 751-759.	2.5	23
89	Class Switch Recombination Process in Ataxia Telangiectasia Patients with Elevated Serum Levels of IgM. <i>Journal of Immunoassay and Immunochemistry</i> , 2015, 36, 16-26.	1.1	21
90	The UNG2 Arg88Cys variant abrogates RPA-mediated recruitment of UNG2 to single-stranded DNA. <i>DNA Repair</i> , 2012, 11, 559-569.	2.8	20

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91	Blood CD4+CD45RO+CXCR5+ T cells are decreased but partially functional in signal transducer and activator of transcription 3 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1146-1156.e5.	2.9	20
92	Activated PI3Kinase Delta Syndrome—A Multifaceted Disease. <i>Frontiers in Pediatrics</i> , 2021, 9, 652405.	1.9	19
93	Immunoglobulin class switch recombination: study through human natural mutants. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2009, 364, 577-582.	4.0	18
94	Locus suicide recombination actively occurs on the functionally rearranged IgH allele in B-cells from inflamed human lymphoid tissues. <i>PLoS Genetics</i> , 2019, 15, e1007721.	3.5	18
95	Topoisomerase 2 β mutation impairs early B-cell development. <i>Blood</i> , 2020, 135, 1497-1501.	1.4	18
96	Predominantly Antibody Deficiencies. , 2008, , 97-130.		17
97	Hyper-immunoglobulin-M syndromes caused by an intrinsic B cell defect. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2003, 3, 421-425.	2.3	14
98	Molecular genetic analysis of Hungarian patients with the hyper-immunoglobulin M syndrome. <i>Molecular Immunology</i> , 2008, 45, 278-282.	2.2	13
99	Genotoxic stress increases cytoplasmic mitochondrial DNA editing by human APOBEC3 mutator enzymes at a single cell level. <i>Scientific Reports</i> , 2019, 9, 3109.	3.3	13
100	Known and potential molecules associated with altered B cell development leading to predominantly antibody deficiencies. <i>Pediatric Allergy and Immunology</i> , 2021, 32, 1601-1615.	2.6	10
101	Inherited Defects of Immunoglobulin Class Switch Recombination. <i>Advances in Experimental Medicine and Biology</i> , 2010, 685, 166-174.	1.6	9
102	Study of patients with Hyper-IgM type IV phenotype who recovered spontaneously during late childhood and review of the literature. <i>European Journal of Pediatrics</i> , 2011, 170, 1039-1047.	2.7	9
103	Primary Immunodeficiencies: Genotype-Phenotype Correlations. , 2006, , 443-460.		7
104	De novo 13q12.3–q14.11 deletion involving <i>BRCA2</i> gene in a patient with developmental delay, elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an Aicardi like phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2571-2576.	1.2	6
105	Class Switch Recombination Defects: impact on B cell maturation and antibody responses. <i>Clinical Immunology</i> , 2021, 222, 108638.	3.2	6
106	Terminal defects of B lymphocyte differentiation. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2001, 1, 519-524.	2.3	5
107	Analysis of somatic hypermutations in the IgM switch region in human B cells. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 411-419.e1.	2.9	5
108	Early-onset hypogammaglobulinemia: A survey of 44 patients. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1097-1099.e2.	2.9	5

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109	Activation-induced deaminase is critical for the establishment of DNA methylation patterns prior to the germinal center reaction. <i>Nucleic Acids Research</i> , 2021, 49, 5057-5073.	14.5	5
110	From Dysgammaglobulinemia to Autosomal-Dominant Activation-Induced Cytidine Deaminase Deficiency: Unraveling an Inherited Immunodeficiency After 50 Years. <i>Journal of Pediatrics</i> , 2020, 223, 207-211.e1.	1.8	4
111	Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. <i>Journal of Clinical Medicine</i> , 2020, 9, 818.	2.4	4
112	UnAIDed Class Switching in Activated B-Cells Reveals Intrinsic Features of a Self-Cleaving IgH Locus. <i>Frontiers in Immunology</i> , 2021, 12, 737427.	4.8	4
113	A novel form of non-X-linked hyperigm associated with growth and pubertal disturbances and with lymphoma development. <i>Journal of Pediatrics</i> , 2006, 148, 404-406.	1.8	3
114	Class-Switch Recombination Defects. , 2014, , 367-387.		2
115	Predominantly Antibody Deficiencies. , 2017, , 183-244.		2
116	Two Monogenetic Disorders, Activated PI3-Kinase-Î Syndrome 2 and Smithâ€Magenis Syndrome, in One Patient: Case Report and a Literature Review of Neurodevelopmental Impact in Primary Immunodeficiencies Associated With Disturbed PI3K Signaling. <i>Frontiers in Pediatrics</i> , 2021, 9, 688022.	1.9	2
117	Connection between induction of DNA lesions and DNA recombination/repair during Ig class switch recombination. <i>Cell Cycle</i> , 2011, 10, 1335-1336.	2.6	1
118	The Hyper IgM Syndromes â€ a Long List of Genes and Years of Discovery. , 2014, , 197-216.		1
119	Immune Deficiencies Caused by B Cell Defects. , 2015, , 463-479.		1
120	Predominantly antibody deficiencies. , 2021, , 93-123.		1
121	Characterization of immunoglobulin mutations in humans with activation-induced cytidine deaminase deficiency. <i>Arthritis Research and Therapy</i> , 2007, 9, P14.	3.5	0
122	AID in Immunodeficiency and Cancer. <i>Molecular Medicine and Medicinal</i> , 2010, , 152-186.	0.4	0
123	Predominantly Antibody Deficiency. , 2012, , 113-192.		0
124	Activated PI3-kinase Î Syndrome: Long-term Follow-up after Cord Blood Transplantation. <i>Journal of Clinical Immunology</i> , 2016, 36, 544-546.	3.8	0