Anna Simon

List of Publications by Year in Descending Order

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Version: 2024-04-28

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

133
papers

9,088
citations

47
ph-index
g-index

145
ext. papers

10,761
ext. citations

6
avg, IF
L-index

#	Paper	IF	Citations
133	Optimal use of [18F]FDG-PET/CT in patients with fever or inflammation of unknown origin. Quarterly Journal of Nuclear Medicine and Molecular Imaging, 2021 , 65, 51-58	1.4	4
132	Curation and expansion of Human Phenotype Ontology for defined groups of inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2021 ,	11.5	3
131	Phenotypic diversity, disease progression, and pathogenicity of MVK missense variants in mevalonic aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2021 , 44, 1272-1287	5.4	2
130	Immunoglobulin Replacement Therapy Versus Antibiotic Prophylaxis as Treatment for Incomplete Primary Antibody Deficiency. <i>Journal of Clinical Immunology</i> , 2021 , 41, 382-392	5.7	3
129	Canakinumab improves patient-reported outcomes in children and adults with autoinflammatory recurrent fever syndromes: results from the CLUSTER trial. <i>Clinical and Experimental Rheumatology</i> , 2021 , 39 Suppl 132, 51-58	2.2	
128	Canakinumab improves patient-reported outcomes in children and adults with autoinflammatory recurrent fever syndromes: results from the CLUSTER trial. <i>Clinical and Experimental Rheumatology</i> , 2021 , 39, 51-58	2.2	
127	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. <i>Genome Medicine</i> , 2019 , 11, 38	14.4	27
126	Classification criteria for autoinflammatory recurrent fevers. <i>Annals of the Rheumatic Diseases</i> , 2019 , 78, 1025-1032	2.4	159
125	Mevalonate Kinase Deficiency 2019 , 315-327		
124	Complex medical history of a patient with a compound heterozygous mutation in. <i>Lupus</i> , 2019 , 28, 1255	5-1.1860	2
123	Defective Protein Prenylation in a Spectrum of Patients With Mevalonate Kinase Deficiency. <i>Frontiers in Immunology</i> , 2019 , 10, 1900	8.4	10
122	An International Delphi Survey for the Definition of New Classification Criteria for Familial Mediterranean Fever, Mevalonate Kinase Deficiency, TNF Receptor-associated Periodic Fever Syndromes, and Cryopyrin-associated Periodic Syndrome. <i>Journal of Rheumatology</i> , 2019 , 46, 429-436	4.1	12
121	Systemic Autoinflammatory Syndromes 2019 , 825-834.e1		
120	Decreased quality of life and societal impact of cryopyrin-associated periodic syndrome treated with canakinumab: a questionnaire based cohort study. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 59	4.2	5
119	Response to Jolobe: 'Molecular diagnostics in FUO'. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2018 , 111, 211	2.7	1
118	In silico validation of the Autoinflammatory Disease Damage Index. <i>Annals of the Rheumatic Diseases</i> , 2018 , 77, 1599-1605	2.4	17
117	Long-term prognosis, treatment, and outcome of patients with fever of unknown origin in whom no diagnosis was made despite extensive investigation: A questionnaire based study. <i>Medicine</i> (United States), 2018, 97, e11241	1.8	11

(2015-2018)

116	Canakinumab for the Treatment of Autoinflammatory Recurrent Fever Syndromes. <i>New England Journal of Medicine</i> , 2018 , 378, 1908-1919	59.2	214
115	Erythematous nodes, urticarial rash and arthralgias in a large pedigree with NLRC4-related autoinflammatory disease, expansion of the phenotype. <i>British Journal of Dermatology</i> , 2017 , 176, 244-	-248	49
114	Development of the autoinflammatory disease damage index (ADDI). <i>Annals of the Rheumatic Diseases</i> , 2017 , 76, 821-830	2.4	54
113	Referral of patients with fever of unknown origin to an expertise center has high diagnostic and therapeutic value. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2017 , 110, 793-801	2.7	12
112	Familial Autoinflammatory Syndromes 2017 , 1666-1684.e4		2
111	A web-based collection of genotype-phenotype associations in hereditary recurrent fevers from the Eurofever registry. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 167	4.2	42
110	International multi-centre study of pregnancy outcomes with interleukin-1 inhibitors. <i>Rheumatology</i> , 2017 , 56, 2102-2108	3.9	54
109	Defective protein prenylation is a diagnostic biomarker of mevalonate kinase deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2017 , 140, 873-875.e6	11.5	19
108	Peri- and Postoperative Treatment with the Interleukin-1 Receptor Antagonist Anakinra Is Safe in Patients Undergoing Renal Transplantation: Case Series and Review of the Literature. <i>Frontiers in Pharmacology</i> , 2017 , 8, 342	5.6	18
107	The challenge of autoinflammatory syndromes: with an emphasis on hyper-IgD syndrome. <i>Rheumatology</i> , 2016 , 55, ii23-ii29	3.9	9
106	Rheumatologic diseases as the cause of fever of unknown origin. <i>Best Practice and Research in Clinical Rheumatology</i> , 2016 , 30, 789-801	5.3	19
105	THU0569 Pharmacokinetics and Pharmacodynamics of Canakinumab in Patients with Autoinflammatory Periodic Fever Syndromes (Colchicine Resistant FMF, HIDS/MKD and TRAPS). <i>Annals of the Rheumatic Diseases</i> , 2016 , 75, 397.3-398	2.4	1
104	The Phenotype and Genotype of Mevalonate Kinase Deficiency: A Series of 114 Cases From the Eurofever Registry. <i>Arthritis and Rheumatology</i> , 2016 , 68, 2795-2805	9.5	112
103	FRI0489 Canakinumab Improves Patient Reported Outcomes in Patients with Periodic Fever Syndromes. <i>Annals of the Rheumatic Diseases</i> , 2016 , 75, 616.1-616	2.4	2
102	FRI0488 A Phase Iii Pivotal Umbrella Trial of Canakinumab in Patients with Autoinflammatory Periodic Fever Syndromes (Colchicine Resistant FMF, HIDS/MKD and TRAPS). <i>Annals of the Rheumatic Diseases</i> , 2016 , 75, 615.2-616	2.4	3
101	Prognosis of Good syndrome: mortality and morbidity of thymoma associated immunodeficiency in perspective. <i>Clinical Immunology</i> , 2016 , 171, 12-17	9	39
100	Evidence-based recommendations for genetic diagnosis of familial Mediterranean fever. <i>Annals of the Rheumatic Diseases</i> , 2015 , 74, 635-41	2.4	112
99	ATP-Induced IL-1িSpecific Secretion: True Under Stringent Conditions. <i>Frontiers in Immunology</i> , 2015 , 6, 54	8.4	33

98	Marked variability in clinical presentation and outcome of patients with C1q immunodeficiency. Journal of Autoimmunity, 2015 , 62, 39-44	15.5	27
97	TLR2/TLR4-dependent exaggerated cytokine production in hyperimmunoglobulinaemia D and periodic fever syndrome. <i>Rheumatology</i> , 2015 , 54, 363-8	3.9	33
96	Recommendations for the management of autoinflammatory diseases. <i>Annals of the Rheumatic Diseases</i> , 2015 , 74, 1636-44	2.4	179
95	Th17 cytokine deficiency in patients with Aspergillus skull base osteomyelitis. <i>BMC Infectious Diseases</i> , 2015 , 15, 140	4	18
94	Hyper-IgD syndrome/mevalonate kinase deficiency: what is new?. <i>Seminars in Immunopathology</i> , 2015 , 37, 371-6	12	41
93	The role of interleukin-1 beta in the pathophysiology of Schnitzler's syndrome. <i>Arthritis Research and Therapy</i> , 2015 , 17, 187	5.7	32
92	Myeloid lineage-restricted somatic mosaicism of NLRP3 mutations in patients with variant Schnitzler syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2015 , 135, 561-4	11.5	94
91	Anakinra injection site reaction on FDG PET/CT. Clinical Nuclear Medicine, 2015, 40, 492-3	1.7	1
90	Mast-cell interleukin-1 Ineutrophil interleukin-17 and epidermal antimicrobial proteins in the neutrophilic urticarial dermatosis in Schnitzler's syndrome. <i>British Journal of Dermatology</i> , 2015 , 173, 448-56	4	26
89	International experience of pregnancy outcomes in auto-inflammatory syndromes treated with Interleukin-1 inhibitors. <i>Pediatric Rheumatology</i> , 2015 , 13,	3.5	2
88	Successful kidney transplantation during anakinra treatment without complications. <i>Pediatric Rheumatology</i> , 2015 , 13,	3.5	78
87	Genetic and phenotypic characteristics of 114 patients with mevalonate kinase deficiency. <i>Pediatric Rheumatology</i> , 2015 , 13,	3.5	5
86	A novel mutation in NLRC4 in a large pedigree with an anakinra responsive autoinflammatory disease. <i>Pediatric Rheumatology</i> , 2015 , 13, P30	3.5	78
85	Fever of unknown origin. <i>Clinical Medicine</i> , 2015 , 15, 280-4	1.9	56
84	Evidence-based provisional clinical classification criteria for autoinflammatory periodic fevers. <i>Annals of the Rheumatic Diseases</i> , 2015 , 74, 799-805	2.4	170
83	Mevalonate kinase deficiency nomenclature. <i>Rheumatology International</i> , 2014 , 34, 295-6	3.6	2
82	The Concept of Autoinflammatory Diseases 2014 , 39-50		
81	Cholesterol metabolism and immunity. New England Journal of Medicine, 2014, 371, 1933-5	59.2	37

(2013-2014)

80	MEFV mutations affecting pyrin amino acid 577 cause autosomal dominant autoinflammatory disease. <i>Annals of the Rheumatic Diseases</i> , 2014 , 73, 455-61	2.4	75
79	Evidence based recommendations for diagnosis and treatment of tumor necrosis factor receptor-1 associated periodic syndrome (TRAPS). <i>Pediatric Rheumatology</i> , 2014 , 12,	3.5	78
78	Evidence based recommendations for genetic diagnosis of Familial Mediterranean Fever. <i>Pediatric Rheumatology</i> , 2014 , 12,	3.5	5
77	The discriminative capacity of soluble Toll-like receptor (sTLR)2 and sTLR4 in inflammatory diseases. <i>BMC Immunology</i> , 2014 , 15, 55	3.7	40
76	Evidence based recommendations for diagnosis and management of mevalonate kinase defiency (MKD). <i>Pediatric Rheumatology</i> , 2014 , 12,	3.5	78
75	Validation of the auto-inflammatory diseases activity index (AIDAI) for hereditary recurrent fever syndromes. <i>Annals of the Rheumatic Diseases</i> , 2014 , 73, 2168-73	2.4	87
74	Comment on "Power of rare diseases: found in translation". <i>Science Translational Medicine</i> , 2014 , 6, 219le	17 .5	3
73	Cytokine production assays reveal discriminatory immune defects in adults with recurrent infections and noninfectious inflammation. <i>Vaccine Journal</i> , 2014 , 21, 1061-9		4
72	Circulating galectin-3 in infections and non-infectious inflammatory diseases. <i>European Journal of Clinical Microbiology and Infectious Diseases</i> , 2013 , 32, 1605-10	5.3	31
71	Mutations in the mevalonate kinase (MVK) gene cause nonsyndromic retinitis pigmentosa. Ophthalmology, 2013 , 120, 2697-2705	7.3	47
70	Treatment of autoinflammatory diseases: results from the Eurofever Registry and a literature review. <i>Annals of the Rheumatic Diseases</i> , 2013 , 72, 678-85	2-4	292
69	Long chain fatty acid (Lcfa) abnormalities in hyper Igd syndrome (Hids) and Familial Mediterranean Fever (Fmf): new insight into heritable periodic fevers. <i>Molecular Genetics and Metabolism</i> , 2013 , 108, 166-71	3.7	0
68	Unexplained recurrent fever: when is autoinflammation the explanation?. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2013 , 68, 285-96	9.3	30
67	OR5-002 In vitro studies in Schnitzler syndrome. <i>Pediatric Rheumatology</i> , 2013 , 11,	3.5	78
66	OR7-002 Pyrin 577 mutations in dominant autoinflammation. <i>Pediatric Rheumatology</i> , 2013 , 11,	3.5	78
65	PW02-034 - NLRP3 mosaicism detection in CAPS using NGS. <i>Pediatric Rheumatology</i> , 2013 , 11,	3.5	78
64	OR11-002 - Mutations in MVK cause non-syndromic RP. <i>Pediatric Rheumatology</i> , 2013 , 11, A191	3.5	78
63	PW03-006 - IL-1-B inhibition in Schnitzler syndrome. <i>Pediatric Rheumatology</i> , 2013 , 11,	3.5	78

62	Schnitzler's syndrome: diagnosis, treatment, and follow-up. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2013 , 68, 562-8	9.3	146
61	Sustained efficacy of the monoclonal anti-interleukin-1 beta antibody canakinumab in a 9-month trial in Schnitzler's syndrome. <i>Annals of the Rheumatic Diseases</i> , 2013 , 72, 1634-8	2.4	77
60	THU0377 Efficacy, safety and pharmacokinetics of the anti-interleukin-1 beta antibody canakinumab in patients with schnitzler syndrome. <i>Annals of the Rheumatic Diseases</i> , 2013 , 71, 283.1-28	3 ^{2.4}	
59	OP0175 The eurofever registry for autoinflammatory disease: Update on enrollment after 2 years. <i>Annals of the Rheumatic Diseases</i> , 2013 , 71, 114.1-114	2.4	
58	Familial Autoinflammatory Syndromes 2013 , 1597-1615.e4		
57	Systemic autoinflammatory syndromes 2013 , 728-739		
56	Pattern recognition receptors in infectious skin diseases. <i>Microbes and Infection</i> , 2012 , 14, 881-93	9.3	21
55	How not to miss autoinflammatory diseases masquerading as urticaria. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2012 , 67, 1465-74	9.3	45
54	An international registry on autoinflammatory diseases: the Eurofever experience. <i>Annals of the Rheumatic Diseases</i> , 2012 , 71, 1177-82	2.4	121
53	Strong induction of AIM2 expression in human epidermis in acute and chronic inflammatory skin conditions. <i>Experimental Dermatology</i> , 2012 , 21, 961-4	4	53
52	Pattern recognition receptors in immune disorders affecting the skin. <i>Journal of Innate Immunity</i> , 2012 , 4, 225-40	6.9	11
51	Treating inflammation by blocking interleukin-1 in a broad spectrum of diseases. <i>Nature Reviews Drug Discovery</i> , 2012 , 11, 633-52	64.1	1160
50	Successful canakinumab treatment identifies IL-1[as a pivotal mediator in Schnitzler syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2011 , 128, 1352-4	11.5	42
49	Mitochondrial reactive oxygen species promote production of proinflammatory cytokines and are elevated in TNFR1-associated periodic syndrome (TRAPS). <i>Journal of Experimental Medicine</i> , 2011 , 208, 519-33	16.6	614
48	Variable expression and treatment of PAPA syndrome. <i>Annals of the Rheumatic Diseases</i> , 2011 , 70, 116	3 <i>-3</i> 7.Q	34
47	Audiometric characteristics of a Dutch family with Muckle-Wells syndrome. <i>Hearing Research</i> , 2011 , 282, 243-51	3.9	9
46	Effects of the histone deacetylase inhibitor ITF2357 in autoinflammatory syndromes. <i>Molecular Medicine</i> , 2011 , 17, 363-8	6.2	21
45	Hyper-IgD syndrome or mevalonate kinase deficiency. <i>Current Opinion in Rheumatology</i> , 2011 , 23, 419-2	23 5.3	39

(2008-2011)

44	A preliminary score for the assessment of disease activity in hereditary recurrent fevers: results from the AIDAI (Auto-Inflammatory Diseases Activity Index) Consensus Conference. <i>Annals of the Rheumatic Diseases</i> , 2011 , 70, 309-14	2.4	54
43	On-demand anakinra treatment is effective in mevalonate kinase deficiency. <i>Annals of the Rheumatic Diseases</i> , 2011 , 70, 2155-8	2.4	115
42	Concerted action of wild-type and mutant TNF receptors enhances inflammation in TNF receptor 1-associated periodic fever syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010 , 107, 9801-6	11.5	143
41	Blocking IL-1beta to slow down progression of ALS?. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010 , 107, 12741-2	11.5	8
40	IL-1beta processing in host defense: beyond the inflammasomes. <i>PLoS Pathogens</i> , 2010 , 6, e1000661	7.6	354
39	A clinical criterion to exclude the hyperimmunoglobulin D syndrome (mild mevalonate kinase deficiency) in patients with recurrent fever. <i>Journal of Rheumatology</i> , 2009 , 36, 1677-81	4.1	41
38	Abnormal IgD and IgA1 O-glycosylation in hyperimmunoglobulinaemia D and periodic fever syndrome. <i>Clinical and Experimental Medicine</i> , 2009 , 9, 291-6	4.9	7
37	Dysregulation of innate immunity: hereditary periodic fever syndromes. <i>British Journal of Haematology</i> , 2009 , 144, 279-302	4.5	30
36	Cathepsin D activity protects against development of type AA amyloid fibrils. <i>European Journal of Clinical Investigation</i> , 2009 , 39, 412-6	4.6	11
35	Horror autoinflammaticus: the molecular pathophysiology of autoinflammatory disease (*). <i>Annual Review of Immunology</i> , 2009 , 27, 621-68	34.7	808
34	Recurrent febrile syndromes: what a rheumatologist needs to know. <i>Nature Reviews Rheumatology</i> , 2009 , 5, 249-56	8.1	33
33	Complete remission of severe idiopathic cold urticaria on interleukin-1 receptor antagonist (anakinra). <i>Netherlands Journal of Medicine</i> , 2009 , 67, 302-5	0.5	31
32	Response to B chnitzler's Syndrome: A True Auto-Inflammatory Disorder? D Seminars in Arthritis and Rheumatism, 2008 , 38, 164	5.3	
31	IL-1 blockade in Schnitzler syndrome: ex vivo findings correlate with clinical remission. <i>Journal of Allergy and Clinical Immunology</i> , 2008 , 121, 260-2	11.5	72
30	Drosomycin-like defensin, a human homologue of Drosophila melanogaster drosomycin with antifungal activity. <i>Antimicrobial Agents and Chemotherapy</i> , 2008 , 52, 1407-12	5.9	27
29	Increased susceptibility of serum amyloid A 1.1 to degradation by MMP-1: potential explanation for higher risk of type AA amyloidosis. <i>Rheumatology</i> , 2008 , 47, 1651-4	3.9	27
28	Lovastatin inhibits formation of AA amyloid. Journal of Leukocyte Biology, 2008, 83, 1295-9	6.5	11
27	Long-term follow-up, clinical features, and quality of life in a series of 103 patients with hyperimmunoglobulinemia D syndrome. <i>Medicine (United States)</i> , 2008 , 87, 301-310	1.8	263

26	AL amyloidosis enhances development of amyloid A amyloidosis. <i>British Journal of Dermatology</i> , 2007 , 156, 748-9	4	11
25	Schnitzler syndrome: beyond the case reports: review and follow-up of 94 patients with an emphasis on prognosis and treatment. <i>Seminars in Arthritis and Rheumatism</i> , 2007 , 37, 137-48	5.3	176
24	Pseudonormalisation of the T wave: old wine?: A fresh look at a 25-year-old observation. <i>Netherlands Heart Journal</i> , 2007 , 15, 257-9	2.2	6
23	Pathogenesis of familial periodic fever syndromes or hereditary autoinflammatory syndromes. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2007, 292, R86-98	3.2	106
22	Comment on: Schnitzlers syndromeexacerbation after anti-TNF treatment. <i>Rheumatology</i> , 2007 , 46, 1741 author reply 1741	3.9	4
21	Defective apoptosis of peripheral-blood lymphocytes in hyper-IgD and periodic fever syndrome. <i>Blood</i> , 2007 , 109, 2416-8	2.2	32
20	Approach to the diagnosis of hereditary autoinflammatory syndromes. <i>Future Rheumatology</i> , 2007 , 2, 5-8		1
19	Approach to genetic analysis in the diagnosis of hereditary autoinflammatory syndromes. <i>Rheumatology</i> , 2006 , 45, 269-73	3.9	68
18	Beneficial response to anakinra and thalidomide in Schnitzler's syndrome. <i>Annals of the Rheumatic Diseases</i> , 2006 , 65, 542-4	2.4	104
17	Hot and hobbling with hives: Schnitzler syndrome. Clinical Immunology, 2006, 119, 131-4	9	16
16	Familial Mediterranean fevera not so unusual cause of abdominal pain. <i>Baillierels Best Practice and Research in Clinical Gastroenterology</i> , 2005 , 19, 199-213	2.5	30
16 15		2.5 4.9	30 91
	Research in Clinical Gastroenterology, 2005 , 19, 199-213		
15	Research in Clinical Gastroenterology, 2005, 19, 199-213 Hereditary periodic fever and reactive amyloidosis. Clinical and Experimental Medicine, 2005, 5, 87-98 Serum amyloid A serum concentrations and genotype do not explain low incidence of amyloidosis in Hyper-IgD syndrome. Amyloid: the International Journal of Experimental and Clinical Investigation:	4.9	91
15 14	Research in Clinical Gastroenterology, 2005, 19, 199-213 Hereditary periodic fever and reactive amyloidosis. Clinical and Experimental Medicine, 2005, 5, 87-98 Serum amyloid A serum concentrations and genotype do not explain low incidence of amyloidosis in Hyper-IgD syndrome. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2005, 12, 115-9 Effect of etanercept and anakinra on inflammatory attacks in the hyper-IgD syndrome: introducing	4.9	91
15 14 13	Research in Clinical Gastroenterology, 2005, 19, 199-213 Hereditary periodic fever and reactive amyloidosis. Clinical and Experimental Medicine, 2005, 5, 87-98 Serum amyloid A serum concentrations and genotype do not explain low incidence of amyloidosis in Hyper-IgD syndrome. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2005, 12, 115-9 Effect of etanercept and anakinra on inflammatory attacks in the hyper-IgD syndrome: introducing a vaccination provocation model. Netherlands Journal of Medicine, 2005, 63, 260-4	4.9 2.7 0.5	91 17 115
15 14 13	Hereditary periodic fever and reactive amyloidosis. <i>Clinical and Experimental Medicine</i> , 2005 , 5, 87-98 Serum amyloid A serum concentrations and genotype do not explain low incidence of amyloidosis in Hyper-IgD syndrome. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2005 , 12, 115-9 Effect of etanercept and anakinra on inflammatory attacks in the hyper-IgD syndrome: introducing a vaccination provocation model. <i>Netherlands Journal of Medicine</i> , 2005 , 63, 260-4 Mevalonate kinase deficiency: Evidence for a phenotypic continuum. <i>Neurology</i> , 2004 , 62, 994-7	4.9 2.7 0.5	91 17 115 111

LIST OF PUBLICATIONS

8	A founder effect in the hyperimmunoglobulinemia D and periodic fever syndrome. <i>American Journal of Medicine</i> , 2003 , 114, 148-52	2.4	46
7	Pseudothrombocytopenia: a report of a new method to count platelets in a patient with EDTA- and temperature-independent antibodies of the IgM type. <i>European Journal of Haematology</i> , 2002 , 69, 243-	7 ^{3.8}	17
6	Molecular analysis of the mevalonate kinase gene in a cohort of patients with the hyper-igd and periodic fever syndrome: its application as a diagnostic tool. <i>Annals of Internal Medicine</i> , 2001 , 135, 338-	.8 43	69
5	Molecular analysis of MVK mutations and enzymatic activity in hyper-IgD and periodic fever syndrome. <i>European Journal of Human Genetics</i> , 2001 , 9, 260-6	5.3	138
4	Genetic analysis as a valuable key to diagnosis and treatment of periodic Fever. <i>Archives of Internal Medicine</i> , 2001 , 161, 2491-3		19
3	Familial periodic fever and amyloidosis due to a new mutation in the TNFRSF1A gene. <i>American Journal of Medicine</i> , 2001 , 110, 313-6	2.4	32
2	Limited efficacy of thalidomide in the treatment of febrile attacks of the hyper-IgD and periodic fever syndrome: a randomized, double-blind, placebo-controlled trial. <i>Journal of Pharmacology and Experimental Therapeutics</i> , 2001 , 298, 1221-6	4.7	39
1	Pseudohypoparathyroidism type Ia. Albright hereditary osteodystrophy: a model for research on G protein-coupled receptors and genomic imprinting. <i>Netherlands Journal of Medicine</i> , 2000 , 56, 100-9	0.5	8