Isabelle Touitou

List of Publications by Year in Descending Order

70 papers	5,331 citations	30 h-index	73 g-index
76 ext. papers	6,307 ext. citations	4.9 avg, IF	5.19 L-index

#	Paper	IF	Citations
70	Mevalonate Kinase-Associated Diseases: Hunting for Phenotype-Genotype Correlation. *Journal of Clinical Medicine*, **2021**, 10,	5.1	3
69	Is gene panel sequencing more efficient than clinical-based gene sequencing to diagnose autoinflammatory diseases? A randomized study. *Clinical and Experimental Immunology*, **2021**, 203, 105-114	6.2	2
68	INSAID Variant Classification and Eurofever Criteria Guide Optimal Treatment Strategy in Patients with TRAPS: Data from the Eurofever Registry. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 783-791.e4	5.4	5
67	Phenotypic Associations of PSTPIP1 Sequence Variants in PSTPIP1-Associated Autoinflammatory Diseases. *Journal of Investigative Dermatology*, **2021**, 141, 1141-1147	4.3	3
66	TNFR1-d2 carrying the p.(Thr79Met) pathogenic variant is a potential novel actor of TNF/TNFR1 signalling regulation in the pathophysiology of TRAPS. *Scientific Reports*, **2021**, 11, 4172	4.9	
65	DADA2 diagnosed in adulthood versus childhood: A comparative study on 306 patients including a systematic literature review and 12 French cases. *Seminars in Arthritis and Rheumatism*, **2021**, 51, 1170-1179	5.3	2
64	ISSAID/EMQN Best Practice Guidelines for the Genetic Diagnosis of Monogenic Autoinflammatory Diseases in the Next-Generation Sequencing Era. *Clinical Chemistry*, **2020**, 66, 525-536	5.5	17
63	Pregnancy after oocyte donation in a patient with NLRP7 gene mutations and recurrent molar hydatidiform pregnancies. *Journal of Assisted Reproduction and Genetics*, **2020**, 37, 2273-2277	3.4	4
62	Clinical and pathological dermatological features of deficiency of adenosine deaminase 2: A multicenter, retrospective, observational study. *Journal of the American Academy of Dermatology*, **2020**, 83, 1794-1798	4.5	6
61	Fièvre méditerranéenne familiale et autres maladies auto-inflammatoires : de la génétique à la pratique médicale. *Bulletin De L'Academie Nationale De Medecine*, **2020**, 204, 517-523	0.1	
60	Correspondance on "Clinical characteristics and genetic analyses of 187 patients with undefined autoinflammatory diseases". *Annals of the Rheumatic Diseases*, **2020**,	2.4	
59	Comment on: Familial Mediterranean fever: breaking all the (genetic) rules. *Rheumatology*, **2020**, 59, 452	3.9	
58	First report of MEFV gene duplication in a patient with familial Mediterranean fever. *Clinical and Experimental Rheumatology*, **2020**, 38 Suppl 127, 129-130	2.2	
57	Current practices for the genetic diagnosis of autoinflammatory diseases: results of a European Molecular Genetics Quality Network Survey. *European Journal of Human Genetics*, **2019**, 27, 1502-1508	5.3	9
56	Classification criteria for autoinflammatory recurrent fevers. *Annals of the Rheumatic Diseases*, **2019**, 78, 1025-1032	2.4	159
55	The Changing Concepts Regarding the Mediterranean Fever Gene: Toward a Spectrum of Pyrin-Associated Autoinflammatory Diseases with Variable Heredity. *Journal of Pediatrics*, **2019**, 209, 12-16.e1	3.6	5
54	Genetic Approach to the Diagnosis of Autoinflammatory Diseases **2019**, 225-237		0

53	Positive Impact of Expert Reference Center Validation on Performance of Next-Generation Sequencing for Genetic Diagnosis of Autoinflammatory Diseases. *Journal of Clinical Medicine*, **2019**, 8,	5.1	4
52	A decision tree for the genetic diagnosis of deficiency of adenosine deaminase 2 (DADA2): a French reference centres experience. *European Journal of Human Genetics*, **2018**, 26, 960-971	5.3	45
51	New workflow for classification of genetic variants'pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). *Journal of Medical Genetics*, **2018**, 55, 530-537	5.8	73
50	Mosaicism in autoinflammatory diseases: Cryopyrin-associated periodic syndromes (CAPS) and beyond. A systematic review. *Critical Reviews in Clinical Laboratory Sciences*, **2018**, 55, 432-442	9.4	28
49	Consensus proposal for taxonomy and definition of the autoinflammatory diseases (AIDs): a Delphi study. *Annals of the Rheumatic Diseases*, **2018**, 77, 1558-1565	2.4	61
48	Chronic leg ulcer revealing adenosine deaminase 2 deficiency: an atypical presentation. *European Journal of Dermatology*, **2018**, 28, 847-848	0.8	5
47	Reply to S□mez et al. *European Journal of Human Genetics*, **2018**, 26, 1564-1565	5.3	
46	Criteria for CAPS, is it all in the name?. *Annals of the Rheumatic Diseases*, **2017**, 76, e9	2.4	2
45	A new autoinflammatory and autoimmune syndrome associated with NLRP1 mutations: NAIAD (associated autoinflammation with arthritis and dyskeratosis). *Annals of the Rheumatic Diseases*, **2017**, 76, 1191-1198	2.4	138
44	Polymorphisms Associated with Rheumatoid Arthritis Susceptibility in Tunisian and French Female Populations: Influence of Geographic Origin. *Journal of Immunology Research*, **2017**, 2017, 4915950	4.5	7
43	Clinical dose effect and functional consequences of R92Q in two families presenting with a TRAPS/PFAPA-like phenotype. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 110-116	2.3	8
42	ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. *RMD Open*, **2016**, 2, e000236	5.9	38
41	Mutation Update for COL2A1 Gene Variants Associated with Type II Collagenopathies. *Human Mutation*, **2016**, 37, 7-15	4.7	75
40	The autoinflammatory diseases: a fashion with blurred boundaries!. *Seminars in Immunopathology*, **2015**, 37, 359-62	12	3
39	Tolerance and efficacy of off-label anti-interleukin-1 treatments in France: a nationwide survey. *Orphanet Journal of Rare Diseases*, **2015**, 10, 19	4.2	86
38	Diagnosis of cryopyrin-associated periodic syndrome: challenges, recommendations and emerging concepts. *Expert Review of Clinical Immunology*, **2015**, 11, 827-35	5.1	20
37	Evidence-based provisional clinical classification criteria for autoinflammatory periodic fevers. *Annals of the Rheumatic Diseases*, **2015**, 74, 799-805	2.4	170
36	Clinical utility gene card for: prototypic hereditary recurrent fever syndromes (monogenic autoinflammatory syndromes). *European Journal of Human Genetics*, **2015**, 23,	5.3	2

35	International periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis syndrome cohort: description of distinct phenotypes in 301 patients. *Rheumatology*, **2014**, 53, 1125-9	3.9	115
34	Identification of a new exon 2-skipped TNFR1 transcript: regulation by three functional polymorphisms of the TNFR-associated periodic syndrome (TRAPS) gene. *Annals of the Rheumatic Diseases*, **2014**, 73, 290-7	2.4	8
33	The phenotype of TNF receptor-associated autoinflammatory syndrome (TRAPS) at presentation: a series of 158 cases from the Eurofever/EUROTRAPS international registry. *Annals of the Rheumatic Diseases*, **2014**, 73, 2160-7	2.4	179
32	The expanding spectrum of rare monogenic autoinflammatory diseases. *Orphanet Journal of Rare Diseases*, **2013**, 8, 162	4.2	20
31	Treatment of autoinflammatory diseases: results from the Eurofever Registry and a literature review. *Annals of the Rheumatic Diseases*, **2013**, 72, 678-85	2.4	292
30	Inheritance of autoinflammatory diseases: shifting paradigms and nomenclature. *Journal of Medical Genetics*, **2013**, 50, 349-59	5.8	35
29	New genetic interpretation of old diseases. *Autoimmunity Reviews*, **2012**, 12, 5-9	13.6	17
28	An international registry on autoinflammatory diseases: the Eurofever experience. *Annals of the Rheumatic Diseases*, **2012**, 71, 1177-82	2.4	121
27	Guidelines for the genetic diagnosis of hereditary recurrent fevers. *Annals of the Rheumatic Diseases*, **2012**, 71, 1599-605	2.4	132
26	The spectrum of NLRP7 mutations in French patients with recurrent hydatidiform mole. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2011**, 157, 197-9	2.4	18
25	Mutations causing familial biparental hydatidiform mole implicate c6orf221 as a possible regulator of genomic imprinting in the human oocyte. *American Journal of Human Genetics*, **2011**, 89, 451-8	11	180
24	Mevalonate kinase deficiency: a survey of 50 patients. *Pediatrics*, **2011**, 128, e152-9	7.4	160
23	Mutations in the autoinflammatory cryopyrin-associated periodic syndrome gene: epidemiological study and lessons from eight years of genetic analysis in France. *Annals of the Rheumatic Diseases*, **2011**, 70, 495-9	2.4	118
22	Combined mutation and rearrangement screening by quantitative PCR high-resolution melting: is it relevant for hereditary recurrent Fever genes?. *PLoS ONE*, **2010**, 5, e14096	3.7	5
21	Web resources for rare auto-inflammatory diseases: towards a common patient registry. *Rheumatology*, **2009**, 48, 665-9	3.9	10
20	The clinical spectrum of 94 patients carrying a single mutated MEFV allele. *Rheumatology*, **2009**, 48, 840-3	3.9	48
19	Expression of the familial Mediterranean fever gene is regulated by nonsense-mediated decay. *Human Molecular Genetics*, **2009**, 18, 4746-55	5.6	15
18	Familial mediterranean Fever in the world. *Arthritis and Rheumatism*, **2009**, 61, 1447-53		233

17	An international external quality assessment for molecular diagnosis of hereditary recurrent fevers: a 3-year scheme demonstrates the need for improvement. *European Journal of Human Genetics*, **2009**, 17, 890-6	5.3	13
16	Characterization of new mutations in the 5Rflanking region of the familial Mediterranean fever gene. *Genes and Immunity*, **2009**, 10, 273-9	4.4	7
15	Search for copy number alterations in the MEFV gene using multiplex ligation probe amplification, experience from three diagnostic centres. *European Journal of Human Genetics*, **2008**, 16, 1404-6	5.3	16
14	The infevers autoinflammatory mutation online registry: update with new genes and functions. *Human Mutation*, **2008**, 29, 803-8	4.7	189
13	Country as the primary risk factor for renal amyloidosis in familial Mediterranean fever. *Arthritis and Rheumatism*, **2007**, 56, 1706-12		204
12	Transmission of familial Mediterranean fever mutations following bone marrow transplantation. *Clinical Genetics*, **2007**, 72, 162-3	4	4
11	Autoinflammatory gene mutations in Behet disease. *Annals of the Rheumatic Diseases*, **2007**, 66, 832-4	2.4	51
10	Refractory auto-inflammatory syndrome associated with digenic transmission of low-penetrance tumour necrosis factor receptor-associated periodic syndrome and cryopyrin-associated periodic syndrome mutations. *Annals of the Rheumatic Diseases*, **2006**, 65, 1530-1	2.4	33
9	A decision tree for genetic diagnosis of hereditary periodic fever in unselected patients. *Annals of the Rheumatic Diseases*, **2006**, 65, 1427-32	2.4	58
8	Infevers: an evolving mutation database for auto-inflammatory syndromes. *Human Mutation*, **2004**, 24, 194-8	4.7	211
7	Identifying mutations in autoinflammatory diseases: towards novel genetic tests and therapies?. *Molecular Diagnosis and Therapy*, **2004**, 4, 109-18		20
6	INFEVERS: the Registry for FMF and hereditary inflammatory disorders mutations. *Nucleic Acids Research*, **2003**, 31, 282-5	20.1	132
5	The spectrum of Familial Mediterranean Fever (FMF) mutations. *European Journal of Human Genetics*, **2001**, 9, 473-83	5.3	394
4	MEFV mutations in Behet disease. *Human Mutation*, **2000**, 16, 271-2	4.7	110
3	Familial Mediterranean fever clinical and genetic features in Druzes and in Iraqi Jews: a preliminary study. *Journal of Rheumatology*, **1998**, 25, 916-9	4.1	16
2	Genotypic diagnosis of familial Mediterranean fever (FMF) using new microsatellite markers: example of two extensive non-Ashkenazi Jewish pedigrees. *Journal of Medical Genetics*, **1997**, 34, 375-81	5.8	3
1	A candidate gene for familial Mediterranean fever. *Nature Genetics*, **1997**, 17, 25-31	36.3	1169