Isabelle Touitou

List of Publications by Year
in descending order

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Version: 2024-02-01

73 papers	6,979 citations	136950 32 h-index	88630 70 g-index
76 all docs	76 docs citations	76 times ranked	4794 citing authors

#	Article	IF	Citations
1	Twists and turns of the genetic story of mevalonate kinase-associated diseases: A review. Genes and Diseases, 2022, 9, 1000-1007.	3.4	10
2	Biallelic NLRP7 variants in patients with recurrent hydatidiform mole: A review and expert consensus. Human Mutation, 2022, 43, 1732-1744.	2.5	6
3	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.	3.8	16
4	Phenotypic Associations of PSTPIP1 Sequence Variants in PSTPIP1-Associated Autoinflammatory Diseases. Journal of Investigative Dermatology, 2021, 141, 1141-1147.	0.7	16
5	TNFR1-d2 carrying the p.(Thr79Met) pathogenic variant is a potential novel actor of TNFÎ±/TNFR1 signalling regulation in the pathophysiology of TRAPS. Scientific Reports, 2021, 11, 4172.	3.3	1
6	Mevalonate Kinase-Associated Diseases: Hunting for Phenotypeâ€“Genotype Correlation. Journal of Clinical Medicine, 2021, 10, 1552.	2.4	14
7	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.	3.4	14
8	Comment on: Familial Mediterranean fever: breaking all the (genetic) rules. Rheumatology, 2020, 59, 452-452.	1.9	0
9	Correspondance on â€˜Clinical characteristics and genetic analyses of 187 patients with undefined autoinflammatory diseasesâ€™. Annals of the Rheumatic Diseases, 2020, , annrheumdis-2020-219566.	0.9	0
10	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.	3.2	43
11	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.	2.5	10
12	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.	1.2	13
13	Is gene panel sequencing more efficient than clinical-based gene sequencing to diagnose autoinflammatory diseases? A randomized study. Clinical and Experimental Immunology, 2020, 203, 105-114.	2.6	8
14	First report of MEFV gene duplication in a patient with familial Mediterranean fever. Clinical and Experimental Rheumatology, 2020, 38 Suppl 127, 129-130.	0.8	0
15	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, 1729.	2.4	9
16	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.	2.8	10
17	Classification criteria for autoinflammatory recurrent fevers. Annals of the Rheumatic Diseases, 2019, 78, 1025-1032.	0.9	300
18	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.	1.8	14

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19	Genetic Approach to the Diagnosis of Autoinflammatory Diseases. , 2019, , 225-237.		2
20	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.	2.8	65
21	New workflow for classification of genetic variantsâ€™ pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). Journal of Medical Genetics, 2018, 55, 530-537.	3.2	117
22	Reply to SÃ¶nmez et al.. European Journal of Human Genetics, 2018, 26, 1564-1565.	2.8	0
23	Mosaicism in autoinflammatory diseases: Cryopyrin-associated periodic syndromes (CAPS) and beyond. A systematic review. Critical Reviews in Clinical Laboratory Sciences, 2018, 55, 432-442.	6.1	45
24	Consensus proposal for taxonomy and definition of the autoinflammatory diseases (AIDs): a Delphi study. Annals of the Rheumatic Diseases, 2018, 77, 1558-1565.	0.9	114
25	Chronic leg ulcer revealing adenosine deaminase 2 deficiency: an atypical presentation. European Journal of Dermatology, 2018, 28, 847-848.	0.6	7
26	Dominant familial Mediterranean fever. Rheumatology, 2017, 56, 173-175.	1.9	8
27	How to favour efficient networking of teams working in the field of rare diseases? Experience of the CeRÃ©MAI reference centre for auto-inflammatory diseases. Joint Bone Spine, 2017, 84, 125-128.	1.6	2
28	Criteria for CAPS, is it all in the name?. Annals of the Rheumatic Diseases, 2017, 76, e9-e9.	0.9	3
29	A new autoinflammatory and autoimmune syndrome associated with NLRP1 mutations: NAIAD (NLRP1-associated autoinflammation with arthritis and dyskeratosis). Annals of the Rheumatic Diseases, 2017, 76, 1191-1198.	0.9	181
30	Clinical dose effect and functional consequences of R92Q in two families presenting with a TRAPS/PFAPAâ€like phenotype. Molecular Genetics & Genomic Medicine, 2017, 5, 110-116.	1.2	10
31	TMEM187-IRAK1Polymorphisms Associated with Rheumatoid Arthritis Susceptibility in Tunisian and French Female Populations: Influence of Geographic Origin. Journal of Immunology Research, 2017, 2017, 1-12.	2.2	9
32	Mutation Update forCOL2A1Gene Variants Associated with Type II Collagenopathies. Human Mutation, 2016, 37, 7-15.	2.5	107
33	ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. RMD Open, 2016, 2, e000236.	3.8	47
34	Diagnosis of cryopyrin-associated periodic syndrome: challenges, recommendations and emerging concepts. Expert Review of Clinical Immunology, 2015, 11, 827-835.	3.0	24
35	Evidence-based provisional clinical classification criteria for autoinflammatory periodic fevers. Annals of the Rheumatic Diseases, 2015, 74, 799-805.	0.9	215
36	Clinical utility gene card for: Prototypic hereditary recurrent fever syndromes (monogenic) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 62 Td (a	2.8	2

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37	The autoinflammatory diseases: a fashion with blurred boundaries!. Seminars in Immunopathology, 2015, 37, 359-362.	6.1	3
38	Tolerance and efficacy of off-label anti-interleukin-1 treatments in France: a nationwide survey. Orphanet Journal of Rare Diseases, 2015, 10, 19.	2.7	117
39	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, 2160-2167.	0.9	256
40	International periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis syndrome cohort: description of distinct phenotypes in 301 patients. Rheumatology, 2014, 53, 1125-1129.	1.9	155
41	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, 290-297.	0.9	10
42	The expanding spectrum of rare monogenic autoinflammatory diseases. Orphanet Journal of Rare Diseases, 2013, 8, 162.	2.7	26
43	Treatment of autoinflammatory diseases: results from the Eurofever Registry and a literature review. Annals of the Rheumatic Diseases, 2013, 72, 678-685.	0.9	350
44	Inheritance of autoinflammatory diseases: shifting paradigms and nomenclature. Journal of Medical Genetics, 2013, 50, 349-359.	3.2	46
45	Guidelines for the genetic diagnosis of hereditary recurrent fevers. Annals of the Rheumatic Diseases, 2012, 71, 1599-1605.	0.9	160
46	New genetic interpretation of old diseases. Autoimmunity Reviews, 2012, 12, 5-9.	5.8	21
47	An International registry on Autoinflammatory diseases: the Eurofever experience. Annals of the Rheumatic Diseases, 2012, 71, 1177-1182.	0.9	158
48	The spectrum of NLRP7 mutations in French patients with recurrent hydatidiform mole. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2011, 157, 197-199.	1.1	19
49	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, 451-458.	6.2	207
50	Mevalonate Kinase Deficiency: A Survey of 50 Patients. Pediatrics, 2011, 128, e152-e159.	2.1	195
51	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, 495-499.	0.9	146
52	Combined Mutation And Rearrangement Screening by Quantitative PCR High-Resolution Melting: Is It Relevant for Hereditary Recurrent Fever Genes?. PLoS ONE, 2010, 5, e14096.	2.5	5
53	Web resources for rare auto-inflammatory diseases: towards a common patient registry. Rheumatology, 2009, 48, 665-669.	1.9	13
54	The clinical spectrum of 94 patients carrying a single mutated MEFV allele. Rheumatology, 2009, 48, 840-842.	1.9	64

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55	Expression of the familial Mediterranean fever gene is regulated by nonsense-mediated decayâ€. Human Molecular Genetics, 2009, 18, 4746-4755.	2.9	17
56	Familial Mediterranean Fever in the World. Arthritis and Rheumatism, 2009, 61, 1447-1453.	6.7	323
57	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, 890-896.	2.8	15
58	Characterization of new mutations in the 5â€²-flanking region of the familial Mediterranean fever gene. Genes and Immunity, 2009, 10, 273-279.	4.1	8
59	The infevers autoinflammatory mutation online registry: update with new genes and functions. Human Mutation, 2008, 29, 803-808.	2.5	239
60	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, 1404-1406.	2.8	17
61	Autoinflammatory gene mutations in Behcet's disease. Annals of the Rheumatic Diseases, 2007, 66, 832-834.	0.9	63
62	Country as the primary risk factor for renal amyloidosis in familial mediterranean fever. Arthritis and Rheumatism, 2007, 56, 1706-1712.	6.7	243
63	Transmission of familial Mediterranean fever mutations following bone marrow transplantation. Clinical Genetics, 2007, 72, 162-163.	2.0	5
64	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, 1530-1531.	0.9	36
65	A decision tree for genetic diagnosis of hereditary periodic fever in unselected patients. Annals of the Rheumatic Diseases, 2006, 65, 1427-1432.	0.9	74
66	Infevers: An evolving mutation database for auto-inflammatory syndromes. Human Mutation, 2004, 24, 194-198.	2.5	277
67	Identifying Mutations in Autoinflammatory Diseases. Molecular Diagnosis and Therapy, 2004, 4, 109-118.	3.3	26
68	INFEVERS: the Registry for FMF and hereditary inflammatory disorders mutations. Nucleic Acids Research, 2003, 31, 282-285.	14.5	171
69	The spectrum of Familial Mediterranean Fever (FMF) mutations. European Journal of Human Genetics, 2001, 9, 473-483.	2.8	480
70	MEFV mutations in Behï¿½et's disease. Human Mutation, 2000, 16, 271-272.	2.5	144
71	Familial Mediterranean fever clinical and genetic features in Druzes and in Iraqi Jews: a preliminary study. Journal of Rheumatology, 1998, 25, 916-9.	2.0	19
72	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, 375-381.	3.2	4

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73	A candidate gene for familial Mediterranean fever. Nature Genetics, 1997, 17, 25-31.	21.4	1,402