MariÃ«lle E Van Gijn

List of Publications by Year
in descending order

57 papers	6,011 citations	218677 26 h-index	155660 55 g-index
61 all docs	61 docs citations	61 times ranked	9158 citing authors

#	Article	IF	Citations
1	Adult-onset autoinflammation caused by somatic mutations in UBA1: AÂDutch case series of patients with VEXAS. Journal of Allergy and Clinical Immunology, 2022, 149, 432-439.e4.	2.9	105
2	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
3	Enhanced hepatic clearance of hyposialylated platelets explains thrombocytopenia in GNE-related macrothrombocytopenia. Blood Advances, 2022, 6, 3347-3351.	5.2	5
4	FAIR Genomes metadata schema promoting Next Generation Sequencing data reuse in Dutch healthcare and research. Scientific Data, 2022, 9, 169.	5.3	8
5	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
6	National external quality assessment for next-generation sequencing-based diagnostics of primary immunodeficiencies. European Journal of Human Genetics, 2021, 29, 20-28.	2.8	5
7	Collodion babies: A 15-year retrospective multicenter study in The Netherlandsâ€"Evaluation of severity scores to predict the underlying disease. Journal of the American Academy of Dermatology, 2021, 84, 1111-1113.	1.2	4
8	Diagnostic next generation sequencing in neonatal erythroderma. JDDG - Journal of the German Society of Dermatology, 2021, 19, 612-614.	0.8	5
9	A Minimal Parameter Set Facilitating Early Decision-making in the Diagnosis of Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2021, 41, 1219-1228.	3.8	8
10	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
11	Implementation of Early Next-Generation Sequencing for Inborn Errors of Immunity: A Prospective Observational Cohort Study of Diagnostic Yield and Clinical Implications in Dutch Genome Diagnostic Centers. Frontiers in Immunology, 2021, 12, 780134.	4.8	12
12	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	1.9	36
13	CAPICE: a computational method for Consequence-Agnostic Pathogenicity Interpretation of Clinical Exome variations. Genome Medicine, 2020, 12, 75.	8.2	30
14	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
15	Cost and impact of early diagnosis in primary immunodeficiency disease: A literature review. Clinical Immunology, 2020, 213, 108359.	3.2	25
16	Dutch genome diagnostic laboratories accelerated and improved variant interpretation and increased accuracy by sharing data. Human Mutation, 2019, 40, 2230-2238.	2.5	32
17	Clinical characteristics and genetic analyses of 187 patients with undefined autoinflammatory diseases. Annals of the Rheumatic Diseases, 2019, 78, 1405-1411.	0.9	44
18	Gene Mosaicism Screening Using Single-Molecule Molecular Inversion Probes in Routine Diagnostics for Systemic Autoinflammatory Diseases. Journal of Molecular Diagnostics, 2019, 21, 943-950.	2.8	5

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19	Diagnostic Yield of Next Generation Sequencing in Genetically Undiagnosed Patients with Primary Immunodeficiencies: a Systematic Review. Journal of Clinical Immunology, 2019, 39, 577-591.	3.8	58
20	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
21	Classification criteria for autoinflammatory recurrent fevers. Annals of the Rheumatic Diseases, 2019, 78, 1025-1032.	0.9	300
22	FRI0568â€…THE USE OF NEXT GENERATION SEQUENCING PANEL IN UNDIFFERENTIATED AUTOINFLAMMATORY DISEASES IDENTIFY A SEPARATE SUBSET OF COLCHICINE-RESPONDER RECURRENT FEVERS DISTINCT FROM PFAPA SYNDROME. , 2019, , .		3
23	Phenotypic variability including BehÃ§et's disease-like manifestations in DADA2 patients due to a homozygous c.973-2A>G splice site mutation. Clinical and Experimental Rheumatology, 2019, 37 Suppl 121, 142-146.	0.8	7
24	Lethal neonatal bone marrow failure syndrome with multiple congenital abnormalities, including limb defects, due to a constitutional deletion of 3â€² MECOM. Haematologica, 2018, 103, e173-e176.	3.5	13
25	Proline-serine-threonine phosphatase interacting protein 1 (PSTPIP1) controls immune synapse stability in human T cells. Journal of Allergy and Clinical Immunology, 2018, 142, 1947-1955.	2.9	17
26	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
27	Somatic alterations compromised molecular diagnosis of DOCK8 hyper-IgE syndrome caused by a novel intronic splice site mutation. Scientific Reports, 2018, 8, 16719.	3.3	5
28	A novel LPS-responsive beige-like anchor protein (LRBA) mutation presents with normal cytotoxic T lymphocyte-associated protein 4 (CTLA-4) and overactive TH17 immunity. Journal of Allergy and Clinical Immunology, 2018, 142, 1968-1971.	2.9	13
29	Congenital erythroderma should be considered as an urgent warning sign of immunodeficiency: a case of Omenn syndrome. European Journal of Dermatology, 2017, 27, 313-314.	0.6	8
30	X-linked primary immunodeficiency associated with hemizygous mutations in the moesin (MSN) gene. Journal of Allergy and Clinical Immunology, 2016, 138, 1681-1689.e8.	2.9	60
31	Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. Rheumatology, 2016, 55, 902-910.	1.9	116
32	Reduced serpinB9-mediated caspase-1 inhibition can contribute to autoinflammatory disease. Oncotarget, 2016, 7, 19265-19271.	1.8	15
33	A novel human STAT3 mutation presents with autoimmunity involving Th17 hyperactivation. Oncotarget, 2015, 6, 20037-20042.	1.8	30
34	Diagnostic Value of Urinary Mevalonic Acid Excretion in Patients with a Clinical Suspicion of Mevalonate Kinase Deficiency (MKD). JIMD Reports, 2015, 27, 33-38.	1.5	40
35	Myeloid lineageâ€“restricted somatic mosaicism of NLRP3 mutations in patients with variant Schnitzler syndrome. Journal of Allergy and Clinical Immunology, 2015, 135, 561-564.e4.	2.9	115
36	Dysfunctional BLK in common variable immunodeficiency perturbs B-cell proliferation and ability to elicit antigen-specific CD4+ T-cell help. Oncotarget, 2015, 6, 10759-10771.	1.8	20

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37	Diagnostic value of urinary mevalonic acid excretion in mevalonate kinase deficiency (MKD). Pediatric Rheumatology, 2014, 12, .	2.1	0
38	Defects in Mitochondrial Clearance Predispose Human Monocytes to Interleukin-1Î² Hypersecretion. Journal of Biological Chemistry, 2014, 289, 5000-5012.	3.4	90
39	Targeted next-generation sequencing: AÂnovel diagnostic tool for primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2014, 133, 529-534.e1.	2.9	143
40	MEFVmutations affecting pyrin amino acid 577 cause autosomal dominant autoinflammatory disease. Annals of the Rheumatic Diseases, 2014, 73, 455-461.	0.9	101
41	A novel FcÎ³RIIa Q27W gene variant is associated with common variable immune deficiency through defective FcÎ³RIIa downstream signaling. Clinical Immunology, 2014, 155, 108-117.	3.2	15
42	Acceptance of Genetic Counseling and Testing in a Hospitalâ€Based Series of Patients with Gynecological Cancer. Journal of Genetic Counseling, 2013, 22, 345-357.	1.6	7
43	Defective calcium signaling and disrupted CD20â€“B-cell receptor dissociation in patients with common variable immunodeficiency disorders. Journal of Allergy and Clinical Immunology, 2012, 129, 755-761.e7.	2.9	27
44	CD27 deficiency is associated with combined immunodeficiency and persistent symptomatic EBV viremia. Journal of Allergy and Clinical Immunology, 2012, 129, 787-793.e6.	2.9	198
45	A novel Dutch mutation in UNC13D reveals an essential role of the C2B domain in munc13â€4 function. Pediatric Blood and Cancer, 2012, 58, 598-605.	1.5	7
46	Risk of colorectal and endometrial cancers in EPCAM deletion-positive Lynch syndrome: a cohort study. Lancet Oncology, The, 2011, 12, 49-55.	10.7	232
47	Recurrence and variability of germline EPCAM deletions in Lynch syndrome. Human Mutation, 2011, 32, 407-414.	2.5	137
48	High incidence of NLRP3 somatic mosaicism in patients with chronic infantile neurologic, cutaneous, articular syndrome: Results of an international multicenter collaborative study. Arthritis and Rheumatism, 2011, 63, 3625-3632.	6.7	247
49	A novel pathogenic MLH1 missense mutation, c.112A > C, p.Asn38His, in six families with Lynch syndrome. Hereditary Cancer in Clinical Practice, 2010, 8, 7.	1.5	21
50	A New Pathogenomic Mechanism In FPD/AML by a Constitutional T(16;21) Involving 16p13 ATF7IP2 and 21q22 RUNX1 Loci. Blood, 2010, 116, 4851-4851.	1.4	0
51	A duplication including GATA4 does not coâ€segregate with congenital heart defects. American Journal of Medical Genetics, Part A, 2009, 149A, 1062-1066.	1.2	12
52	Transcription Factor Achaete Scute-Like 2 Controls Intestinal Stem Cell Fate. Cell, 2009, 136, 903-912.	28.9	615
53	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
54	Loss of intestinal crypt progenitor cells owing to inactivation of both Notch1 and Notch2 is accompanied by derepression of CDK inhibitors p27^{Kip1} and p57^{Kip2}. EMBO Reports, 2008, 9, 377-383.	4.5	362

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55	The Intestinal Wnt/TCF Signature. Gastroenterology, 2007, 132, 628-632.	1.3	439
56	Wnt signalling induces maturation of Paneth cells in intestinal crypts. Nature Cell Biology, 2005, 7, 381-386.	10.3	555
57	Notch/Î³-secretase inhibition turns proliferative cells in intestinal crypts and adenomas into goblet cells. Nature, 2005, 435, 959-963.	27.8	1,382