Christine BellannÃ©-Chantelot

List of Publications by Year
in descending order

55 papers	2,823 citations	218677 26 h-index	182427 51 g-index
56 all docs	56 docs citations	56 times ranked	4085 citing authors

#	ARTICLE	IF	CITATIONS
1	A Syndrome with Congenital Neutropenia and Mutations inG6PC3. New England Journal of Medicine, 2009, 360, 32-43.	27.0	331
2	Uncoupling of GTP hydrolysis from eIF6 release on the ribosome causes Shwachman-Diamond syndrome. Genes and Development, 2011, 25, 917-929.	5.9	247
3	Genetic and clinical implications of the Val617Phe JAK2 mutation in 72 families with myeloproliferative disorders. Blood, 2006, 108, 346-352.	1.4	221
4	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. Haematologica, 2018, 103, 1278-1287.	3.5	129
5	Classification of and risk factors for hematologic complications in a French national cohort of 102 patients with Shwachman-Diamond syndrome. Haematologica, 2012, 97, 1312-1319.	3.5	120
6	JAGN1 deficiency causes aberrant myeloid cell homeostasis and congenital neutropenia. Nature Genetics, 2014, 46, 1021-1027.	21.4	119
7	Diabetes, Associated Clinical Spectrum, Long-term Prognosis, and Genotype/Phenotype Correlations in 201 Adult Patients With Hepatocyte Nuclear Factor 1B (HNF1B) Molecular Defects. Diabetes Care, 2017, 40, 1436-1443.	8.6	117
8	Congenital neutropenia in the era of genomics: classification, diagnosis, and natural history. British Journal of Haematology, 2017, 179, 557-574.	2.5	115
9	Germline duplication of ATG2B and GSKIP predisposes to familial myeloid malignancies. Nature Genetics, 2015, 47, 1131-1140.	21.4	107
10	Congenital Hyperinsulinism: Pancreatic [¹⁸F]Fluoro-<scp>l</scp>-Dihydroxyphenylalanine (DOPA) Positron Emission Tomography and Immunohistochemistry Study of DOPA Decarboxylase and Insulin Secretion. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 933-940.	3.6	97
11	Analysis of the Ten-Eleven Translocation 2 (TET2) gene in familial myeloproliferative neoplasms. Blood, 2009, 114, 1628-1632.	1.4	96
12	Description and outcome of a cohort of 8 patients with WHIM syndrome from the French Severe Chronic Neutropenia Registry. Orphanet Journal of Rare Diseases, 2012, 7, 71.	2.7	96
13	Update of variants identified in the pancreatic Î²â€cell K _{ATP} channel genes KCNJ11 and ABCC8 in individuals with congenital hyperinsulinism and diabetes. Human Mutation, 2020, 41, 884-905.	2.5	90
14	Distinct patterns of mutations occurring in de novo AML versus AML arising in the setting of severe congenital neutropenia. Blood, 2007, 110, 1648-1655.	1.4	88
15	Mutations in the SRP54 gene cause severe congenital neutropenia as well as Shwachman-Diamondâ€“like syndrome. Blood, 2018, 132, 1318-1331.	1.4	85
16	The added value of [18F]fluoro-L-DOPA PET in the diagnosis of hyperinsulinism of infancy: a retrospective study involving 49 children. European Journal of Nuclear Medicine and Molecular Imaging, 2007, 34, 2120-2128.	6.4	71
17	Clinical Characteristics and Diagnostic Criteria of Maturity-Onset Diabetes Of The Young (MODY) due to Molecular Anomalies of the HNF1A Gene. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1346-E1351.	3.6	70
18	Epidemiology of Congenital Neutropenia. Hematology/Oncology Clinics of North America, 2013, 27, 1-17.	2.2	55

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19	EFL1 mutations impair eIF6 release to cause Shwachman-Diamond syndrome. Blood, 2019, 134, 277-290.	1.4	48
20	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	12.8	44
21	Next-generation sequencing identifies monogenic diabetes in 16% of patients with late adolescence/adult-onset diabetes selected on a clinical basis: a cross-sectional analysis. BMC Medicine, 2019, 17, 132.	5.5	41
22	Searching for Maturity-Onset Diabetes of the Young (MODY): When and What for?. Canadian Journal of Diabetes, 2016, 40, 455-461.	0.8	40
23	New somatic BRAF splicing mutation in Langerhans cell histiocytosis. Molecular Cancer, 2017, 16, 115.	19.2	37
24	Lymphoid differentiation of hematopoietic stem cells requires efficient Cxcr4 desensitization. Journal of Experimental Medicine, 2017, 214, 2023-2040.	8.5	36
25	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	3.8	30
26	Tubular proteinuria in patients with HNF1Î± mutations: HNF1Î± drives endocytosis in the proximal tubule. Kidney International, 2016, 89, 1075-1089.	5.2	29
27	Clinical utility gene card for: Maturity-onset diabetes of the young. European Journal of Human Genetics, 2014, 22, 1153-1153.	2.8	26
28	Heterozygous variants of CLPB are a cause of severe congenital neutropenia. Blood, 2022, 139, 779-791.	1.4	25
29	Application of wholeâ€exome sequencing to unravel the molecular basis of undiagnosed syndromic congenital neutropenia with intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 62-71.	1.2	23
30	Gene Panel Sequencing of Patients With Monogenic Diabetes Brings to Light Genes Typically Associated With Syndromic Presentations. Diabetes, 2022, 71, 578-584.	0.6	23
31	New pathogenic mechanisms induced by germline erythropoietin receptor mutations in primary erythrocytosis. Haematologica, 2018, 103, 575-586.	3.5	17
32	Germline genetic factors in the pathogenesis of myeloproliferative neoplasms. Blood Reviews, 2020, 42, 100710.	5.7	16
33	Liver adenomatosis in patients with hepatocyte nuclear factorâ€1 alpha maturity onset diabetes of the young (HNF1Aâ€MODY): Clinical, radiological and pathological characteristics in a French series. Journal of Diabetes, 2020, 12, 48-57.	1.8	14
34	The Common HNF1A Variant I27L Is a Modifier of Age at Diabetes Diagnosis in Individuals With HNF1A-MODY. Diabetes, 2018, 67, 1903-1907.	0.6	12
35	Biological behaviors of mutant proinsulin contribute to the phenotypic spectrum of diabetes associated with insulin gene mutations. Molecular and Cellular Endocrinology, 2020, 518, 111025.	3.2	11
36	ATG2B and GSKIP: 2 new genes predisposing to myeloid malignancies. Molecular and Cellular Oncology, 2016, 3, e1094564.	0.7	10

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37	Identification of biallelic germline variants of SRP68 in a sporadic case with severe congenital neutropenia. Haematologica, 2021, 106, 1216-1219.	3.5	10
38	Germline ATG2B/GSKIP-containing 14q32 duplication predisposes to early clonal hematopoiesis leading to myeloid neoplasms. Leukemia, 2022, 36, 126-137.	7.2	10
39	Postâ€COVIDâ€19 severe neutropenia. Pediatric Blood and Cancer, 2021, 68, e28866.	1.5	9
40	An update on congenital hyperinsulinism: advances in diagnosis and management. Expert Opinion on Orphan Drugs, 2014, 2, 779-795.	0.8	8
41	Pregnancy in Women With Monogenic Diabetes due to Pathogenic Variants of the Glucokinase Gene: Lessons and Challenges. Frontiers in Endocrinology, 2021, 12, 802423.	3.5	7
42	Functional characterization of ABCC8 variants of unknown significance based on bioinformatics predictions, splicing assays, and protein analyses: Benefits for the accurate diagnosis of congenital hyperinsulinism. Human Mutation, 2021, 42, 408-420.	2.5	6
43	Chronic neutropenia: how best to assess severity and approach management?. Expert Review of Hematology, 2021, 14, 945-960.	2.2	6
44	An inherited gainâ€ofâ€function risk allele in <scp>EPOR</scp> predisposes to familial <scp>JAK2^{V617F}</scp> myeloproliferative neoplasms. British Journal of Haematology, 2022, 198, 131-136.	2.5	6
45	ATG2B/GSKIP in de novo acute myeloid leukemia (AML): high prevalence of germline predisposition in French West Indies. Leukemia and Lymphoma, 2021, 62, 1770-1773.	1.3	5
46	Biallelic CXCR2 loss-of-function mutations define a distinct congenital neutropenia entity. Haematologica, 2022, 107, 765-769.	3.5	5
47	Shwachmanâ€Diamond syndrome and solid tumors: Three new patients from the French Registry for Severe Chronic Neutropenia and literature review. Pediatric Blood and Cancer, 2021, 68, e29071.	1.5	4
48	Response to Comment on Dubois-Laforgue et al. Diabetes, Associated Clinical Spectrum, Long-term Prognosis, and Genotype/Phenotype Correlations in 201 Adult Patients With Hepatocyte Nuclear Factor 1B (HNF1B) Molecular Defects. Diabetes Care 2017;40:1436â€“1443. Diabetes Care, 2018, 41, e8-e9.	8.6	3
49	A Novel Clinical Syndrome Associating Severe Congenital Neutropenia and Complex Developmental Aberrations Caused by Deficiency of G6PC3. Blood, 2008, 112, 5-5.	1.4	3
50	Deficiency Of JAGN1 Causes Severe Congenital Neutropenia Associated With Defective Secretory Pathway and Aberrant Myeloid Cell Homeostasis. Blood, 2013, 122, 439-439.	1.4	2
51	Risk of Leukemia in Genetic Subgroups of Congenital Neutropenia (CN): Comparison of Patients with Mutations in HAX1 or ELA2. Blood, 2008, 112, 2532-2532.	1.4	1
52	Update On the Risk of Leukemia in Genetic Subgroups of Congenital Neutropenia (CN): Comparison of Patients with Known Gene Mutations (ELA2, HAX1, WASP, G6PC3, p14).. Blood, 2009, 114, 3597-3597.	1.4	1
53	TET2 Mutations in Polycythemia Vera (PV) in Some Cases Follow Rather Than Precede JAK2 V617F Mutation, Are Not a Disease-Initiating Event, Affect Mainly Erythropoiesis, and Contribute to Increased Aggressivity of PV Clone.. Blood, 2009, 114, 3913-3913.	1.4	1
54	Study of V617F JAK2-Positive and JAK2-Negative Patients within 18 Families with Myeloproliferative Disorders (MPD): Disease Characteristics and Evolution, EPOR and MPL Analysis.. Blood, 2006, 108, 672-672.	1.4	0

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55	Long-Term Follow-up of 94 Families with Myeloproliferative Neoplasms: Implications of the V617F JAK2 mutation, Life Expectancy and Complication Rates. Blood, 2010, 116, 1978-1978.	1.4	0