C N Hahn

List of Publications by Year in Descending Order

82 papers	3,370 citations	33 h-index	57 g-index
87 ext. papers	3,902 ext. citations	7.5 avg, IF	4.47 L-index

#	Paper	IF	Citations
82	Pathogenic variants in cause recessive central conducting lymphatic anomaly with lymphedema.. *Science Translational Medicine*, **2022**, 14, eabm4869	17.5	0
81	Targeted gene panels identify a high frequency of pathogenic germline variants in patients diagnosed with a hematological malignancy and at least one other independent cancer. *Leukemia*, **2021**, 35, 3245-3256	10.7	10
80	The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. *Haematologica* , **2021**, 106, 3004-3007	6.6	3
79	GATA2 deficiency syndrome: A decade of discovery. *Human Mutation*, **2021**, 42, 1399-1421	4.7	4
78	Childhood acute myeloid leukemia shows a high level of germline predisposition. *Blood*, **2021**, 138, 2293-2298	22	0
77	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. *Blood Advances*, **2020**, 4, 1131-1144	7.8	37
76	Secondary leukemia in patients with germline transcription factor mutations (RUNX1, GATA2, CEBPA). *Blood*, **2020**, 136, 24-35	2.2	35
75	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. *British Journal of Haematology*, **2020**, 190, e297-e301	4.5	10
74	Two monogenic disorders masquerading as one: severe congenital neutropenia with monocytosis and non-syndromic sensorineural hearing loss. *BMC Medical Genetics*, **2020**, 21, 35	2.1	1
73	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. *Leukemia*, **2019**, 33, 2842-2853	10.7	19
72	Genomic subtyping and therapeutic targeting of acute erythroleukemia. *Nature Genetics*, **2019**, 51, 694-704	30.3	54
71	A novel germline mutation in a family with ataxia-pancytopenia syndrome and pediatric acute lymphoblastic leukemia. *Haematologica*, **2019**, 104, e318-e321	6.6	11
70	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. *Blood*, **2019**, 134, 1439-1439	2.2	2
69	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. *Blood*, **2019**, 134, 3794-3794	2.2	
68	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. *Npj Genomic Medicine*, **2019**, 4, 28	6.2	5
67	A four-gene LincRNA expression signature predicts risk in multiple cohorts of acute myeloid leukemia patients. *Leukemia*, **2018**, 32, 263-272	10.7	25
66	Differential effects on gene transcription and hematopoietic differentiation correlate with GATA2 mutant disease phenotypes. *Leukemia*, **2018**, 32, 194-202	10.7	32

65	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. *Leukemia*, **2018**, 32, 2502-2507	10.7	33
64	Development of a Data Portal for Aggregation and Analysis of Genomics Data in Familial Platelet Disorder with Predisposition to Myeloid Malignancy - the RUNX1.DB. *Blood*, **2018**, 132, 5241-5241	2.2	
63	Myeloid neoplasms with germline DDX41 mutation. *International Journal of Hematology*, **2017**, 106, 163-174	2.3	44
62	A novel, somatic, transforming mutation in the extracellular domain of Epidermal Growth Factor Receptor identified in myeloproliferative neoplasm. *Scientific Reports*, **2017**, 7, 2467	4.9	4
61	T cell receptor assessment in autoimmune disease requires access to the most adjacent immunologically active organ. *Journal of Autoimmunity*, **2017**, 81, 24-33	15.5	5
60	Self-reverting mutations partially correct the blood phenotype in a Diamond Blackfan anemia patient. *Haematologica*, **2017**, 102, e506-e509	6.6	17
59	ARMC5 is not implicated in familial hyperaldosteronism type II (FH-II). *Journal of Human Hypertension*, **2017**, 31, 857-859	2.6	3
58	A Method for Next-Generation Sequencing of Paired Diagnostic and Remission Samples to Detect Mitochondrial DNA Mutations Associated with Leukemia. *Journal of Molecular Diagnostics*, **2017**, 19, 711-721	5.1	7
57	Clinical implications of transient myeloproliferative disorder in a neonate without Down syndrome features. *Journal of Paediatrics and Child Health*, **2017**, 53, 1018-1020	1.3	2
56	Apparent ЈAK2-negativeTpolycythaemia vera due to compound mutations in exon 14. *British Journal of Haematology*, **2017**, 178, 333-336	4.5	7
55	In depth analysis of the Sox4 gene locus that consists of sense and natural antisense transcripts. *Data in Brief*, **2016**, 7, 282-90	1.2	4
54	Derivation of an endogenous small RNA from double-stranded Sox4 sense and natural antisense transcripts in the mouse brain. *Genomics*, **2016**, 107, 88-99	4.3	14
53	Delayed diagnosis leading to accelerated-phase chronic eosinophilic leukemia due to a cytogenetically cryptic, imatinib-responsive TNIP1-PDFGRB fusion gene. *Leukemia*, **2016**, 30, 1402-5	10.7	6
52	Expanded Phenotypic and Genetic Heterogeneity in the Clinical Spectrum of FPD-AML: Lymphoid Malignancies and Skin Disorders Are Common Features in Carriers of Germline RUNX1 Mutations. *Blood*, **2016**, 128, 1212-1212	2.2	2
51	Ectrodactyly and Lethal Pulmonary Acinar Dysplasia Associated with Homozygous FGFR2 Mutations Identified by Exome Sequencing. *Human Mutation*, **2016**, 37, 955-63	4.7	28
50	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. *Blood*, **2016**, 127, 1017-23	2.2	117
49	Revealing Missing Human Protein Isoforms Based on Ab Initio Prediction, RNA-seq and Proteomics. *Scientific Reports*, **2015**, 5, 10940	4.9	38
48	Allan-Herndon-Dudley syndrome with unusual profound sensorineural hearing loss. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1872-6	2.5	7

47	Characterisation of a compound in-cis GATA2 germline mutation in a pedigree presenting with myelodysplastic syndrome/acute myeloid leukemia with concurrent thrombocytopenia. *Leukemia*, **2015**, 29, 1795-7	10.7	9
46	A tale of two siblings: two cases of AML arising from a single pre-leukemic DNMT3A mutant clone. *Leukemia*, **2015**, 29, 2101-4	10.7	26
45	Splice factor mutations and alternative splicing as drivers of hematopoietic malignancy. *Immunological Reviews*, **2015**, 263, 257-78	11.3	29
44	GATA2 is required for lymphatic vessel valve development and maintenance. *Journal of Clinical Investigation*, **2015**, 125, 2979-94	15.9	136
43	Rare and Common Germline Variants Contribute to Occurrence of Myelodysplastic Syndrome. *Blood*, **2015**, 126, 1644-1644	2.2	1
42	High Incidence of Mutated Cancer-Associated Genes at Diagnosis in CML Patients with Early Transformation to Blast Crisis. *Blood*, **2015**, 126, 600-600	2.2	3
41	ARMC5 mutations are common in familial bilateral macronodular adrenal hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1784-92	5.6	73
40	Aberrant Activation of Epidermal Growth Factor Receptor in MPN May Respond to the Kinase Inhibitor Gefitinib. *Blood*, **2014**, 124, 1882-1882	2.2	
39	Clonal Diversity of Recurrently Mutated Genes in Myelodysplastic Syndromes. *Blood*, **2014**, 124, 4634-4634	2.2	
38	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. *Nature Genetics*, **2013**, 45, 1226-1231	36.3	205
37	Regulation of vascular leak and recovery from ischemic injury by general and VE-cadherin-restricted miRNA antagonists of miR-27. *Blood*, **2013**, 122, 2911-9	2.2	48
36	Loss-of-function germline GATA2 mutations in patients with MDS/AML or MonoMAC syndrome and primary lymphedema reveal a key role for GATA2 in the lymphatic vasculature. *Blood*, **2012**, 119, 1283-91	2.2	216
35	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. *Molecular Psychiatry*, **2012**, 17, 1103-15	15.1	71
34	Genome-wide gene expression profiling identifies overlap with malignant adrenocortical tumours and novel mechanisms of inefficient steroidogenesis in familial ACTH-independent macronodular adrenal hyperplasia. *Endocrine-Related Cancer*, **2012**, 19, L19-23	5.7	4
33	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. *Nature Genetics*, **2011**, 43, 1012-7	36.3	424
32	Deep sequencing analysis of the developing mouse brain reveals a novel microRNA. *BMC Genomics*, **2011**, 12, 176	4.5	46
31	Spatiotemporal regulation of multiple overlapping sense and novel natural antisense transcripts at the Nrgn and Camk2n1 gene loci during mouse cerebral corticogenesis. *Cerebral Cortex*, **2011**, 21, 683-97	5.1	26
30	Spliceosome mutations in hematopoietic malignancies. *Nature Genetics*, **2011**, 44, 9-10	36.3	46

29	Mechanisms of Co-Operation of DNMT3A Mutations with JAK2 V617F Through Histone H4 Arginine 3 Provides New Insights in MPN Disease Pathogenesis. *Blood*, **2011**, 118, 2823-2823	2.2	
28	Poor prognosis in familial acute myeloid leukaemia with combined biallelic CEBPA mutations and downstream events affecting the ATM, FLT3 and CDX2 genes. *British Journal of Haematology*, **2010**, 150, 382-5	4.5	13
27	Novel RUNX1 mutations in familial platelet disorder with enhanced risk for acute myeloid leukemia: clues for improved identification of the FPD/AML syndrome. *Leukemia*, **2010**, 24, 242-6	10.7	73
26	Stress-induced premature senescence mediated by a novel gene, SENEX, results in an anti-inflammatory phenotype in endothelial cells. *Blood*, **2010**, 116, 4016-24	2.2	36
25	GATA2 is a New Predisposition Gene for Familial Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia (AML). *Blood*, **2010**, 116, LBA-3-LBA-3	2.2	8
24	JAM-C induces endothelial cell permeability through its association and regulation of {beta}3 integrins. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2009**, 29, 1200-6	9.4	35
23	Chronic increases in sphingosine kinase-1 activity induce a pro-inflammatory, pro-angiogenic phenotype in endothelial cells. *Cellular and Molecular Biology Letters*, **2009**, 14, 424-41	8.1	23
22	Molecular networks involved in mouse cerebral corticogenesis and spatio-temporal regulation of Sox4 and Sox11 novel antisense transcripts revealed by transcriptome profiling. *Genome Biology*, **2009**, 10, R104	18.3	31
21	Sphingosine kinase-1 associates with integrin {alpha}V{beta}3 to mediate endothelial cell survival. *American Journal of Pathology*, **2009**, 175, 2217-25	5.8	17
20	Basal and angiopoietin-1-mediated endothelial permeability is regulated by sphingosine kinase-1. *Blood*, **2008**, 111, 3489-97	2.2	78
19	Phenoxodiol, an experimental anticancer drug, shows potent antiangiogenic properties in addition to its antitumour effects. *International Journal of Cancer*, **2006**, 118, 2412-20	7.5	67
18	Effect of disrupted SOX18 transcription factor function on tumor growth, vascularization, and endothelial development. *Journal of the National Cancer Institute*, **2006**, 98, 1060-7	9.7	65
17	Expression profiling reveals functionally important genes and coordinately regulated signaling pathway genes during in vitro angiogenesis. *Physiological Genomics*, **2005**, 22, 57-69	3.6	16
16	Sphingosine kinase-1 enhances endothelial cell survival through a PECAM-1-dependent activation of PI-3K/Akt and regulation of Bcl-2 family members. *Blood*, **2005**, 105, 3169-77	2.2	147
15	PPARgamma agonists ameliorate endothelial cell activation via inhibition of diacylglycerol-protein kinase C signaling pathway: role of diacylglycerol kinase. *Circulation Research*, **2004**, 94, 1515-22	15.7	89
14	A vascular cell-restricted RhoGAP, p73RhoGAP, is a key regulator of angiogenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 12212-7	11.5	36
13	Apolipoprotein E enhances hepatic lipase-mediated hydrolysis of reconstituted high-density lipoprotein phospholipid and triacylglycerol in an isoform-dependent manner. *Biochemistry*, **2004**, 43, 12306-14	3.2	19
12	Ozz-E3, a muscle-specific ubiquitin ligase, regulates beta-catenin degradation during myogenesis. *Developmental Cell*, **2004**, 6, 269-82	10.2	76

11	Role of protein kinase Czeta in thrombin-induced endothelial permeability changes: inhibition by angiopoietin-1. *Blood*, **2004**, 104, 1716-24	2.2	59
10	Systemic and neurologic abnormalities distinguish the lysosomal disorders sialidosis and galactosialidosis in mice. *Human Molecular Genetics*, **2002**, 11, 1455-64	5.6	78
9	Osteoblast gene expression in rat long bones: effects of ovariectomy and dihydrotestosterone on mRNA levels. *Calcified Tissue International*, **2000**, 67, 75-9	3.9	35
8	Lack of PPCA expression only partially coincides with lysosomal storage in galactosialidosis mice: indirect evidence for spatial requirement of the catalytic rather than the protective function of PPCA. *Human Molecular Genetics*, **1998**, 7, 1787-94	5.6	19
7	Correction of murine galactosialidosis by bone marrow-derived macrophages overexpressing human protective protein/cathepsin A under control of the colony-stimulating factor-1 receptor promoter. *Proceedings of the National Academy of Sciences of the United States of America*, **1998**, 95, 14880-5	11.5	42
6	Generalized CNS disease and massive GM1-ganglioside accumulation in mice defective in lysosomal acid beta-galactosidase. *Human Molecular Genetics*, **1997**, 6, 205-11	5.6	135
5	Transcriptional synergism between vitamin D-responsive elements in the rat 25-hydroxyvitamin D3 24-hydroxylase (CYP24) promoter. *Journal of Biological Chemistry*, **1996**, 271, 29715-21	5.4	102
4	Identification of a vitamin D responsive element in the promoter of the rat cytochrome P450(24) gene. *Nucleic Acids Research*, **1994**, 22, 2410-6	20.1	53
3	Superinduction by cycloheximide of cytochrome P4502H1 and 5-aminolevulinate synthase gene transcription in chick embryo liver. *Archives of Biochemistry and Biophysics*, **1993**, 300, 531-4	4.1	19
2	Localization of the human vitamin D 24-hydroxylase gene (CYP24) to chromosome 20q13.2-->q13.3. *Cytogenetic and Genome Research*, **1993**, 62, 192-3	1.9	22
1	Transcriptional regulation of the chicken CYP2H1 gene. Localization of a phenobarbital-responsive enhancer domain. *Journal of Biological Chemistry*, **1991**, 266, 17031-9	5.4	28