Neil V Morgan

List of Publications by Year
in descending order

111 papers	11,060 citations	38720 50 h-index	30058 103 g-index
119 all docs	119 docs citations	119 times ranked	12215 citing authors

#	Article	IF	Citations
1	Heterozygous germline mutations in BMPR2, encoding a TGF-Î² receptor, cause familial primary pulmonary hypertension. Nature Genetics, 2000, 26, 81-84.	9.4	1,388
2	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 2001, 345, 325-334.	13.9	676
3	Sporadic primary pulmonary hypertension is associated with germline mutations of the gene encoding BMPR-II, a receptor member of the TGF-beta family. Journal of Medical Genetics, 2000, 37, 741-745.	1.5	645
4	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102.	2.6	521
5	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	9.4	497
6	HIF activation identifies early lesions in VHL kidneys. Cancer Cell, 2002, 1, 459-468.	7.7	456
7	Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogryposisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404.	9.4	313
8	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	9.4	294
9	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196.	9.4	266
10	The Deubiquitinase OTULIN Is an Essential Negative Regulator of Inflammation and Autoimmunity. Cell, 2016, 166, 1215-1230.e20.	13.5	259
11	STAT2 deficiency and susceptibility to viral illness in humans. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 3053-3058.	3.3	222
12	Phenotypic spectrum of neurodegeneration associated with mutations in the PLA2G6 gene (PLAN). Neurology, 2008, 70, 1623-1629.	1.5	215
13	Isolation of a cDNA Representing the Fanconi Anemia Complementation Group E Gene. American Journal of Human Genetics, 2000, 67, 1306-1308.	2.6	201
14	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	9.4	201
15	Spontaneous functional correction of homozygous Fanconi anaemia alleles reveals novel mechanistic basis for reverse mosaicism. Nature Genetics, 1999, 22, 379-383.	9.4	190
16	Mutations in SLC29A3, Encoding an Equilibrative Nucleoside Transporter ENT3, Cause a Familial Histiocytosis Syndrome (Faisalabad Histiocytosis) and Familial Rosai-Dorfman Disease. PLoS Genetics, 2010, 6, e1000833.	1.5	174
17	Homozygous loss-of-function mutations in the gene encoding the dopamine transporter are associated with infantile parkinsonism-dystonia. Journal of Clinical Investigation, 2009, 119, 1595-603.	3.9	173
18	Whole-Exome-Sequencing-Based Discovery of Human FADD Deficiency. American Journal of Human Genetics, 2010, 87, 873-881.	2.6	171

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19	Inactivation of IL11 Signaling Causes Craniosynostosis, Delayed Tooth Eruption, and Supernumerary Teeth. American Journal of Human Genetics, 2011, 89, 67-81.	2.6	164
20	Mutations in the Embryonal Subunit of the Acetylcholine Receptor (CHRNG) Cause Lethal and Escobar Variants of Multiple Pterygium Syndrome. American Journal of Human Genetics, 2006, 79, 390-395.	2.6	145
21	A Germline Mutation in BLOC1S3/Reduced Pigmentation Causes a Novel Variant of Hermansky-Pudlak Syndrome (HPS8). American Journal of Human Genetics, 2006, 78, 160-166.	2.6	129
22	Mutations in TTC37 Cause Trichohepatoenteric Syndrome (Phenotypic Diarrhea of Infancy). Gastroenterology, 2010, 138, 2388-2398.e2.	0.6	124
23	High Frequency of Large Intragenic Deletions in the Fanconi Anemia Group A Gene. American Journal of Human Genetics, 1999, 65, 1330-1341.	2.6	121
24	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica, 2016, 101, 1170-1179.	1.7	119
25	Complementation Analysis in Fanconi Anemia: Assignment of the Reference FA-H Patient to Group A. American Journal of Human Genetics, 2000, 67, 759-762.	2.6	115
26	Multigene methylation analysis of Wilms' tumour and adult renal cell carcinoma. Oncogene, 2003, 22, 6794-6801.	2.6	112
27	Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. Blood, 2013, 122, 4090-4093.	0.6	108
28	Mutation Analysis of CHRNA1, CHRNB1, CHRND, and RAPSN Genes in Multiple Pterygium Syndrome/Fetal Akinesia Patients. American Journal of Human Genetics, 2008, 82, 222-227.	2.6	104
29	Molecular and genealogical evidence for a founder effect in Fanconi anemia families of the Afrikaner population of South Africa. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 5734-5739.	3.3	100
30	Introducing high-throughput sequencing into mainstream genetic diagnosis practice in inherited platelet disorders. Haematologica, 2018, 103, 148-162.	1.7	96
31	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. Brain, 2010, 133, 2964-2970.	3.7	95
32	Potential genetic causes of miscarriage in euploid pregnancies: a systematic review. Human Reproduction Update, 2019, 25, 452-472.	5.2	95
33	Mutation in Rab3 GTPase-Activating Protein (RAB3GAP) Noncatalytic Subunit in a Kindred with Martsolf Syndrome. American Journal of Human Genetics, 2006, 78, 702-707.	2.6	91
34	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. Nature Genetics, 1995, 11, 338-340.	9.4	89
35	Mutation in the TCRÎ± subunit constant gene (TRAC) leads to a human immunodeficiency disorder characterized by a lack of TCRÎ±Î²+ T cells. Journal of Clinical Investigation, 2011, 121, 695-702.	3.9	86
36	Heterogeneous spectrum of mutations in the Fanconi anaemia group A gene. European Journal of Human Genetics, 1999, 7, 52-59.	1.4	84

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37	Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. Leukemia, 2004, 18, 420-425.	3.3	78
38	Paraneoplastic erythrocytosis associated with an inactivating point mutation of the von Hippel-Lindau gene in a renal cell carcinoma. Blood, 2002, 99, 3562-3565.	0.6	72
39	SLFN14 mutations underlie thrombocytopenia with excessive bleeding and platelet secretion defects. Journal of Clinical Investigation, 2015, 125, 3600-3605.	3.9	71
40	Germline mutation in DOK7 associated with fetal akinesia deformation sequence. Journal of Medical Genetics, 2009, 46, 338-340.	1.5	70
41	Novel mutations in RASGRP2, which encodes CalDAG-GEFI, abrogate Rap1 activation, causing platelet dysfunction. Blood, 2016, 128, 1282-1289.	0.6	68
42	A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. Journal of Medical Genetics, 2003, 40, 431-435.	1.5	67
43	Use of nextâ€generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. Journal of Thrombosis and Haemostasis, 2015, 13, 643-650.	1.9	63
44	Defective Leukocyte Adhesion and Chemotaxis Contributes to Combined Immunodeficiency in Humans with Autosomal Recessive MST1 Deficiency. Journal of Clinical Immunology, 2016, 36, 117-122.	2.0	63
45	Genotyping and phenotyping of platelet function disorders. Journal of Thrombosis and Haemostasis, 2013, 11, 351-363.	1.9	62
46	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. European Journal of Human Genetics, 2000, 8, 861-868.	1.4	61
47	Mutations in FLVCR2 Are Associated with Proliferative Vasculopathy and Hydranencephaly-Hydrocephaly Syndrome (Fowler Syndrome). American Journal of Human Genetics, 2010, 86, 471-478.	2.6	60
48	Characterization of multiple platelet activation pathways in patients with bleeding as a high-throughput screening option: use of 96-well Optimul assay. Blood, 2014, 123, e11-e22.	0.6	60
49	Germline TET2 loss of function causes childhood immunodeficiency and lymphoma. Blood, 2020, 136, 1055-1066.	0.6	58
50	A novel locus for Meckel-Gruber syndrome, MKS3 , maps to chromosome 8q24. Human Genetics, 2002, 111, 456-461.	1.8	55
51	Dual Proteolytic Pathways Govern Glycolysis and Immune Competence. Cell, 2014, 159, 1578-1590.	13.5	54
52	A common Fanconi anemia mutation in black populations of sub-Saharan Africa. Blood, 2005, 105, 3542-3544.	0.6	53
53	The Fanconi Anemia Group E Gene, FANCE, Maps to Chromosome 6p. American Journal of Human Genetics, 1999, 64, 1400-1405.	2.6	48
54	Mutation in GNE is associated with severe congenital thrombocytopenia. Blood, 2018, 132, 1855-1858.	0.6	46

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55	Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutation, 1996, 8, 140-148.	1.1	42
56	CHRNGgenotypeâ€“phenotype correlations in the multiple pterygium syndromes. Journal of Medical Genetics, 2012, 49, 21-26.	1.5	41
57	Inherited Thrombocytopenia: Update on Genes and Genetic Variants Which may be Associated With Bleeding. Frontiers in Cardiovascular Medicine, 2019, 6, 80.	1.1	40
58	VACTERL with hydrocephalus in twins due to Fanconi anemia (FA): Mutation in the FAC gene. American Journal of Medical Genetics Part A, 1997, 68, 86-90.	2.4	39
59	Histidinemia in mice: A metabolic defect treated using a novel approach to hepatocellular transplantation. Hepatology, 1995, 21, 1405-1412.	3.6	38
60	A patient-derived mutant form of the Fanconi anemia protein, FANCA, is defective in nuclear accumulation. Experimental Hematology, 1999, 27, 587-593.	0.2	35
61	Inherited thrombocytopenia: novel insights into megakaryocyte maturation, proplatelet formation and platelet lifespan. Platelets, 2016, 27, 519-525.	1.1	35
62	Schlafen 14 (SLFN14) is a novel antiviral factor involved in the control of viral replication. Immunobiology, 2017, 222, 979-988.	0.8	35
63	Flow studies on human GPVI-deficient blood under coagulating and noncoagulating conditions. Blood Advances, 2020, 4, 2953-2961.	2.5	35
64	Variable presentation of primary immune deficiency: Two cases with CD3 gamma deficiency presenting with only autoimmunity. Pediatric Allergy and Immunology, 2013, 24, 257-262.	1.1	34
65	CRISPR-Cas9 Mediated Labelling Allows for Single Molecule Imaging and Resolution. Scientific Reports, 2017, 7, 8450.	1.6	34
66	Phenotype description and response to thrombopoietin receptor agonist in DIAPH1-related disorder. Blood Advances, 2018, 2, 2341-2346.	2.5	33
67	Comparison of multiple electrode aggregometry with lumiâ€aggregometry for the diagnosis of patients with mild bleeding disorders. Journal of Thrombosis and Haemostasis, 2017, 15, 2045-2052.	1.9	31
68	Astute Clinician Report: A Novel 10Âbp Frameshift Deletion in Exon 2 of ICOS Causes a Combined Immunodeficiency Associated with an Enteritis and Hepatitis. Journal of Clinical Immunology, 2015, 35, 598-603.	2.0	30
69	Inherited platelet disorders: Insight from platelet genomics using next-generation sequencing. Platelets, 2017, 28, 14-19.	1.1	30
70	Multiplex ligation-dependent probe amplification (MLPA) analysis is an effective tool for the detection of novel intragenic PLA2G6 mutations: Implications for molecular diagnosis. Molecular Genetics and Metabolism, 2010, 100, 207-212.	0.5	29
71	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.	1.7	29
72	Novel TSHR mutations in consanguineous families with congenital nongoitrous hypothyroidism. Clinical Endocrinology, 2010, 73, 671-677.	1.2	28

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73	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. Human Molecular Genetics, 2016, 25, 1836-1845.	1.4	28
74	Positive diepoxybutane test in only one of two brothers found to be compound heterozygotes for Fanconi's anaemia complementation group C mutations. British Journal of Haematology, 1996, 93, 813-816.	1.2	25
75	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. Acta Neuropathologica Communications, 2014, 2, 148.	2.4	23
76	Initiation codon mutation in betaB1-crystallin (CRYBB1) associated with autosomal recessive nuclear pulverulent cataract. Molecular Vision, 2009, 15, 1014-9.	1.1	23
77	Microsatellite markers as a rapid approach for autozygosity mapping in Hermansky-Pudlak syndrome: Identification of the second HPS7 mutation in a patient presenting late in life. Thrombosis and Haemostasis, 2013, 109, 766-768.	1.8	22
78	Acrocallosal syndrome: Identification of a novel KIF7 mutation and evidence for oligogenic inheritance. European Journal of Medical Genetics, 2013, 56, 39-42.	0.7	21
79	GoldVariants, a resource for sharing rare genetic variants detected in bleeding, thrombotic, and platelet disorders: Communication from the ISTH SSC Subcommittee on Genomics in Thrombosis and Hemostasis. Journal of Thrombosis and Haemostasis, 2021, 19, 2612-2617.	1.9	21
80	<scp>CD3G</scp> Gene Defects in Familial Autoimmune Thyroiditis. Scandinavian Journal of Immunology, 2014, 80, 354-361.	1.3	20
81	High-throughput platelet spreading analysis: a tool for the diagnosis of platelet-based bleeding disorders. Haematologica, 2020, 105, e124-e128.	1.7	20
82	Promoter mutation is a common variant in GJC2-associated Pelizaeusâ€“Merzbacher-like disease. Molecular Genetics and Metabolism, 2011, 104, 637-643.	0.5	19
83	A comprehensive targeted nextâ€generation sequencing panel for genetic diagnosis of patients with suspected inherited thrombocytopenia. Research and Practice in Thrombosis and Haemostasis, 2018, 2, 640-652.	1.0	19
84	Optimised insert design for improved single-molecule imaging and quantification through CRISPR-Cas9 mediated knock-in. Scientific Reports, 2019, 9, 14219.	1.6	19
85	Evaluation of the Total Thrombus-Formation System (T-TAS): application to human and mouse blood analysis. Platelets, 2019, 30, 893-900.	1.1	19
86	An adaptable analysis workflow for characterization of platelet spreading and morphology. Platelets, 2021, 32, 54-58.	1.1	18
87	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. Orphanet Journal of Rare Diseases, 2013, 8, 74.	1.2	17
88	What is the role of genetic testing in the investigation of patients with suspected platelet function disorders?. British Journal of Haematology, 2014, 165, 193-203.	1.2	16
89	Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors. Thrombosis and Haemostasis, 2015, 113, 826-837.	1.8	15
90	Gene of the issue: RUNX1 mutations and inherited bleeding. Platelets, 2017, 28, 208-210.	1.1	15

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91	Post-translational polymodification of Î²1-tubulin regulates motor protein localization in platelet production and function. Haematologica, 2022, 107, 243-259.	1.7	15
92	Role of the novel endoribonuclease SLFN14 and its disease-causing mutations in ribosomal degradation. Rna, 2018, 24, 939-949.	1.6	13
93	Cell-Free DNA in the Investigation of Miscarriage. Journal of Clinical Medicine, 2020, 9, 3428.	1.0	10
94	A comprehensive bioinformatic analysis of 126 patients with an inherited platelet disorder to identify both sequence and copy number genetic variants. Human Mutation, 2020, 41, 1848-1865.	1.1	10
95	Investigation of the contribution of an underlying platelet defect in women with unexplained heavy menstrual bleeding. Platelets, 2019, 30, 56-65.	1.1	9
96	Whole exome sequencing identifies a mutation in thrombomodulin as the genetic cause of a suspected platelet disorder in a family with normal platelet function. Platelets, 2017, 28, 611-613.	1.1	8
97	Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant in HPDL. Brain Communications, 2021, 3, fcab002.	1.5	8
98	Prevalence and natural history of variants in the ANKRD26 gene: a short review and update of reported cases. Platelets, 2022, 33, 1107-1112.	1.1	8
99	SLFN14 gene mutations associated with bleeding. Platelets, 2020, 31, 407-410.	1.1	7
100	A NovelABCA12Mutation in Two Families with Congenital Ichthyosis. Scientifica, 2012, 2012, 1-6.	0.6	6
101	Heterozygous mutation SLFN14 K208N in mice mediates species-specific differences in platelet and erythroid lineage commitment. Blood Advances, 2021, 5, 377-390.	2.5	5
102	Novel gene variants in patients with plateletâ€based bleeding using combined exome sequencing and RNAseq murine expression data. Journal of Thrombosis and Haemostasis, 2021, 19, 262-268.	1.9	4
103	A novel RUNX1 exon 3 - 7 deletion causing a familial platelet disorder. Platelets, 2022, 33, 320-323.	1.1	4
104	Functional Variations In Genes Encoding Platelet G-Protein Coupled Receptors In Unselected and Platelet Function Disorder Populations. Blood, 2013, 122, 3511-3511.	0.6	3
105	Rare missense variants in Tropomyosinâ€4 (TPM4) are associated with platelet dysfunction, cytoskeletal defects, and excessive bleeding. Journal of Thrombosis and Haemostasis, 2022, 20, 478-485.	1.9	3
106	Meeting abstracts from the 64th British Thyroid Association Annual Meeting. Thyroid Research, 2017, 10, .	0.7	2
107	Sorting nexin 24 is required for Î±-granule biogenesis and cargo delivery in megakaryocytes. Haematologica, 2022, 107, 1902-1913.	1.7	2
108	New insights into glycoprotein IbÎ± desialylation-mediated platelet clearance. Platelets, 2020, 31, 621-623.	1.1	1

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109	Molecular Diagnosis of Fanconi Anemia and Dyskeratosis Congenita. , 2004, 91, 3-18.		0
110	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.	0.6	0
111	Comprehensive Description of Monoallelic GP1BA Variants Associated with Thrombocytopenia. Blood, 2020, 136, 34-35.	0.6	0