Neil V Morgan

List of Publications by Year
in descending order

111 papers	11,060 citations	38742 50 h-index	30087 103 g-index
119 all docs	119 docs citations	119 times ranked	12215 citing authors

#	Article	IF	Citations
1	A novel RUNX1 exon 3 - 7 deletion causing a familial platelet disorder. Platelets, 2022, 33, 320-323.	2.3	4
2	Post-translational polymodification of Î²1-tubulin regulates motor protein localization in platelet production and function. Haematologica, 2022, 107, 243-259.	3.5	15
3	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.	3.8	3
4	Sorting nexin 24 is required for Î±-granule biogenesis and cargo delivery in megakaryocytes. Haematologica, 2022, 107, 1902-1913.	3.5	2
5	Prevalence and natural history of variants in the ANKRD26 gene: a short review and update of reported cases. Platelets, 2022, 33, 1107-1112.	2.3	8
6	An adaptable analysis workflow for characterization of platelet spreading and morphology. Platelets, 2021, 32, 54-58.	2.3	18
7	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.	3.8	4
8	Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant in HPDL. Brain Communications, 2021, 3, fcab002.	3.3	8
9	Heterozygous mutation SLFN14 K208N in mice mediates species-specific differences in platelet and erythroid lineage commitment. Blood Advances, 2021, 5, 377-390.	5.2	5
10	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.	3.5	29
11	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.	3.8	21
12	High-throughput platelet spreading analysis: a tool for the diagnosis of platelet-based bleeding disorders. Haematologica, 2020, 105, e124-e128.	3.5	20
13	SLFN14 gene mutations associated with bleeding. Platelets, 2020, 31, 407-410.	2.3	7
14	New insights into glycoprotein Ibî± desialylation-mediated platelet clearance. Platelets, 2020, 31, 621-623.	2.3	1
15	Cell-Free DNA in the Investigation of Miscarriage. Journal of Clinical Medicine, 2020, 9, 3428.	2.4	10
16	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.	2.5	10
17	Flow studies on human GPVI-deficient blood under coagulating and noncoagulating conditions. Blood Advances, 2020, 4, 2953-2961.	5.2	35
18	Germline TET2 loss of function causes childhood immunodeficiency and lymphoma. Blood, 2020, 136, 1055-1066.	1.4	58

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19	Comprehensive Description of Monoallelic GP1BA Variants Associated with Thrombocytopenia. Blood, 2020, 136, 34-35.	1.4	0
20	Inherited Thrombocytopenia: Update on Genes and Genetic Variants Which may be Associated With Bleeding. Frontiers in Cardiovascular Medicine, 2019, 6, 80.	2.4	40
21	Optimised insert design for improved single-molecule imaging and quantification through CRISPR-Cas9 mediated knock-in. Scientific Reports, 2019, 9, 14219.	3.3	19
22	Potential genetic causes of miscarriage in euploid pregnancies: a systematic review. Human Reproduction Update, 2019, 25, 452-472.	10.8	95
23	Evaluation of the Total Thrombus-Formation System (T-TAS): application to human and mouse blood analysis. Platelets, 2019, 30, 893-900.	2.3	19
24	Investigation of the contribution of an underlying platelet defect in women with unexplained heavy menstrual bleeding. Platelets, 2019, 30, 56-65.	2.3	9
25	Role of the novel endoribonuclease SLFN14 and its disease-causing mutations in ribosomal degradation. Rna, 2018, 24, 939-949.	3.5	13
26	Introducing high-throughput sequencing into mainstream genetic diagnosis practice in inherited platelet disorders. Haematologica, 2018, 103, 148-162.	3.5	96
27	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.	2.3	19
28	Phenotype description and response to thrombopoietin receptor agonist in DIAPH1-related disorder. Blood Advances, 2018, 2, 2341-2346.	5.2	33
29	Mutation in GNE is associated with severe congenital thrombocytopenia. Blood, 2018, 132, 1855-1858.	1.4	46
30	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.	1.4	0
31	Inherited platelet disorders: Insight from platelet genomics using next-generation sequencing. Platelets, 2017, 28, 14-19.	2.3	30
32	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.	2.3	8
33	Gene of the issue: RUNX1 mutations and inherited bleeding. Platelets, 2017, 28, 208-210.	2.3	15
34	CRISPR-Cas9 Mediated Labelling Allows for Single Molecule Imaging and Resolution. Scientific Reports, 2017, 7, 8450.	3.3	34
35	Schlafen 14 (SLFN14) is a novel antiviral factor involved in the control of viral replication. Immunobiology, 2017, 222, 979-988.	1.9	35
36	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.	3.8	31

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37	Meeting abstracts from the 64th British Thyroid Association Annual Meeting. Thyroid Research, 2017, 10, .	1.5	2
38	Inherited thrombocytopenia: novel insights into megakaryocyte maturation, proplatelet formation and platelet lifespan. Platelets, 2016, 27, 519-525.	2.3	35
39	Novel mutations in RASGRP2, which encodes CalDAG-GEFI, abrogate Rap1 activation, causing platelet dysfunction. Blood, 2016, 128, 1282-1289.	1.4	68
40	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica, 2016, 101, 1170-1179.	3.5	119
41	The Deubiquitinase OTULIN Is an Essential Negative Regulator of Inflammation and Autoimmunity. Cell, 2016, 166, 1215-1230.e20.	28.9	259
42	Defective Leukocyte Adhesion and Chemotaxis Contributes to Combined Immunodeficiency in Humans with Autosomal Recessive MST1 Deficiency. Journal of Clinical Immunology, 2016, 36, 117-122.	3.8	63
43	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. Human Molecular Genetics, 2016, 25, 1836-1845.	2.9	28
44	Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors. Thrombosis and Haemostasis, 2015, 113, 826-837.	3.4	15
45	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.	3.8	63
46	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.	3.8	30
47	SLFN14 mutations underlie thrombocytopenia with excessive bleeding and platelet secretion defects. Journal of Clinical Investigation, 2015, 125, 3600-3605.	8.2	71
48	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. Acta Neuropathologica Communications, 2014, 2, 148.	5.2	23
49	Dual Proteolytic Pathways Govern Glycolysis and Immune Competence. Cell, 2014, 159, 1578-1590.	28.9	54
50	<scp>CD3G</scp> Gene Defects in Familial Autoimmune Thyroiditis. Scandinavian Journal of Immunology, 2014, 80, 354-361.	2.7	20
51	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.	2.5	16
52	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.	1.4	60
53	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. Orphanet Journal of Rare Diseases, 2013, 8, 74.	2.7	17
54	Acrocallosal syndrome: Identification of a novel KIF7 mutation and evidence for oligogenic inheritance. European Journal of Medical Genetics, 2013, 56, 39-42.	1.3	21

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55	Genotyping and phenotyping of platelet function disorders. Journal of Thrombosis and Haemostasis, 2013, 11, 351-363.	3.8	62
56	Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. Blood, 2013, 122, 4090-4093.	1.4	108
57	Variable presentation of primary immune deficiency: Two cases with CD3 gamma deficiency presenting with only autoimmunity. Pediatric Allergy and Immunology, 2013, 24, 257-262.	2.6	34
58	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.	7.1	222
59	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.	3.4	22
60	Functional Variations In Genes Encoding Platelet G-Protein Coupled Receptors In Unselected and Platelet Function Disorder Populations. Blood, 2013, 122, 3511-3511.	1.4	3
61	A NovelABCA12Mutation in Two Families with Congenital Ichthyosis. Scientifica, 2012, 2012, 1-6.	1.7	6
62	CHRNGgenotypeâ€“phenotype correlations in the multiple pterygium syndromes. Journal of Medical Genetics, 2012, 49, 21-26.	3.2	41
63	Promoter mutation is a common variant in GJC2-associated Pelizaeusâ€“Merzbacher-like disease. Molecular Genetics and Metabolism, 2011, 104, 637-643.	1.1	19
64	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.	8.2	86
65	Inactivation of IL11 Signaling Causes Craniosynostosis, Delayed Tooth Eruption, and Supernumerary Teeth. American Journal of Human Genetics, 2011, 89, 67-81.	6.2	164
66	Mutations in FLVCR2 Are Associated with Proliferative Vasculopathy and Hydranencephaly-Hydrocephaly Syndrome (Fowler Syndrome). American Journal of Human Genetics, 2010, 86, 471-478.	6.2	60
67	Whole-Exome-Sequencing-Based Discovery of Human FADD Deficiency. American Journal of Human Genetics, 2010, 87, 873-881.	6.2	171
68	Novel TSHR mutations in consanguineous families with congenital nongoitrous hypothyroidism. Clinical Endocrinology, 2010, 73, 671-677.	2.4	28
69	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.	3.5	174
70	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. Brain, 2010, 133, 2964-2970.	7.6	95
71	Mutations in TTC37 Cause Trichohepatoenteric Syndrome (Phenotypic Diarrhea of Infancy). Gastroenterology, 2010, 138, 2388-2398.e2.	1.3	124
72	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.	1.1	29

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73	Germline mutation in DOK7 associated with fetal akinesia deformation sequence. Journal of Medical Genetics, 2009, 46, 338-340.	3.2	70
74	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.	8.2	173
75	Initiation codon mutation in betaB1-crystallin (CRYBB1) associated with autosomal recessive nuclear pulverulent cataract. Molecular Vision, 2009, 15, 1014-9.	1.1	23
76	Phenotypic spectrum of neurodegeneration associated with mutations in the PLA2G6 gene (PLAN). Neurology, 2008, 70, 1623-1629.	1.1	215
77	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.	6.2	104
78	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.	6.2	129
79	Mutation in Rab3 GTPase-Activating Protein (RAB3GAP) Noncatalytic Subunit in a Kindred with Martsolf Syndrome. American Journal of Human Genetics, 2006, 78, 702-707.	6.2	91
80	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.	6.2	145
81	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196.	21.4	266
82	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	21.4	497
83	A common Fanconi anemia mutation in black populations of sub-Saharan Africa. Blood, 2005, 105, 3542-3544.	1.4	53
84	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
85	Molecular Diagnosis of Fanconi Anemia and Dyskeratosis Congenita. , 2004, 91, 3-18.		0
86	Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogryposisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404.	21.4	313
87	Deletion and reduced expression of the Fanconi anemia FANCA gene in sporadic acute myeloid leukemia. Leukemia, 2004, 18, 420-425.	7.2	78
88	Multigene methylation analysis of Wilms' tumour and adult renal cell carcinoma. Oncogene, 2003, 22, 6794-6801.	5.9	112
89	A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. Journal of Medical Genetics, 2003, 40, 431-435.	3.2	67
90	Paraneoplastic erythrocytosis associated with an inactivating point mutation of the von Hippel-Lindau gene in a renal cell carcinoma. Blood, 2002, 99, 3562-3565.	1.4	72

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91	HIF activation identifies early lesions in VHL kidneys. Cancer Cell, 2002, 1, 459-468.	16.8	456
92	A novel locus for Meckel-Gruber syndrome, MKS3 , maps to chromosome 8q24. Human Genetics, 2002, 111, 456-461.	3.8	55
93	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102.	6.2	521
94	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 2001, 345, 325-334.	27.0	676
95	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.	7.1	100
96	Heterozygous germline mutations in BMPR2, encoding a TGF-Î² receptor, cause familial primary pulmonary hypertension. Nature Genetics, 2000, 26, 81-84.	21.4	1,388
97	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. European Journal of Human Genetics, 2000, 8, 861-868.	2.8	61
98	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.	3.2	645
99	Isolation of a cDNA Representing the Fanconi Anemia Complementation Group E Gene. American Journal of Human Genetics, 2000, 67, 1306-1308.	6.2	201
100	Complementation Analysis in Fanconi Anemia: Assignment of the Reference FA-H Patient to Group A. American Journal of Human Genetics, 2000, 67, 759-762.	6.2	115
101	Spontaneous functional correction of homozygous Fanconi anaemia alleles reveals novel mechanistic basis for reverse mosaicism. Nature Genetics, 1999, 22, 379-383.	21.4	190
102	Heterogeneous spectrum of mutations in the Fanconi anaemia group A gene. European Journal of Human Genetics, 1999, 7, 52-59.	2.8	84
103	A patient-derived mutant form of the Fanconi anemia protein, FANCA, is defective in nuclear accumulation. Experimental Hematology, 1999, 27, 587-593.	0.4	35
104	The Fanconi Anemia Group E Gene, FANCE, Maps to Chromosome 6p. American Journal of Human Genetics, 1999, 64, 1400-1405.	6.2	48
105	High Frequency of Large Intragenic Deletions in the Fanconi Anemia Group A Gene. American Journal of Human Genetics, 1999, 65, 1330-1341.	6.2	121
106	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
107	Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutation, 1996, 8, 140-148.	2.5	42
108	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.	2.5	25

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109	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	21.4	294
110	Histidinemia in mice: A metabolic defect treated using a novel approach to hepatocellular transplantation. Hepatology, 1995, 21, 1405-1412.	7.3	38
111	Localisation of the Fanconi anaemia complementation group A gene to chromosome 16q24.3. Nature Genetics, 1995, 11, 338-340.	21.4	89