Margaret M Deangelis

List of Publications by Year in Descending Order

87 papers	6,168 citations	34 h-index	78 g-index
100 ext. papers	7,117 ext. citations	6.7 avg, IF	5.07 L-index

#	Paper	IF	Citations
87	Genetics of Age-Related Macular Degeneration **2022**, 3509-3563		
86	Artificial Intelligence, Heuristic Biases, and the Optimization of Health Outcomes: Cautionary Optimism. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
85	Nr2e3 is a genetic modifier that rescues retinal degeneration and promotes homeostasis in multiple models of retinitis pigmentosa. *Gene Therapy*, **2021**, 28, 223-241	4	13
84	Genetic predictors of severe intraventricular hemorrhage in extremely low-birthweight infants. *Journal of Perinatology*, **2021**, 41, 286-294	3.1	
83	Age-Related Macular Degeneration: From Epigenetics to Therapeutic Implications. *Advances in Experimental Medicine and Biology*, **2021**, 1256, 221-235	3.6	2
82	Binding of Gtf2i-/2l transcription factors to the ARMS2 gene leads to increased circulating HTRA1 in AMD patients and in vitro. *Journal of Biological Chemistry*, **2021**, 296, 100456	5.4	1
81	Genetics of Age-Related Macular Degeneration **2021**, 1-55		
80	The possible impact of increased physical intimate partner violence during the COVID-19 pandemic on ocular health. *SAGE Open Medicine*, **2021**, 9, 20503121211035263	2.4	1
79	Global Women's Eye Health: A Genetic Epidemiologic Perspective. *Essentials in Ophthalmology*, **2021**, 11-46	0.2	1
78	The Serine Protease HTRA-1 Is a Biomarker for ROP and Mediates Retinal Neovascularization. *Frontiers in Molecular Neuroscience*, **2020**, 13, 605918	6.1	2
77	Bone Morphogenetic Protein (BMP)4 But Not BMP2 Disrupts the Barrier Integrity of Retinal Pigment Epithelia and Induces Their Migration: A Potential Role in Neovascular Age-Related Macular Degeneration. *Journal of Clinical Medicine*, **2020**, 9,	5.1	6
76	Homocysteine: A Potential Biomarker for Diabetic Retinopathy. *Journal of Clinical Medicine*, **2019**, 8,	5.1	32
75	The Utah Protocol for Postmortem Eye Phenotyping and Molecular Biochemical Analysis **2019**, 60, 1204-1212		11
74	Progressive optic nerve changes in cavitary optic disc anomaly: integration of copy number alteration and cis-expression quantitative trait loci to assess disease etiology. *BMC Medical Genetics* , **2019**, 20, 63	2.1	0
73	Generation, transcriptome profiling, and functional validation of cone-rich human retinal organoids. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 10824-10833	11.5	65
72	ALPK1 missense pathogenic variant in five families leads to ROSAH syndrome, an ocular multisystem autosomal dominant disorder. *Genetics in Medicine*, **2019**, 21, 2103-2115	8.1	7
71	ABSENCE OF MACULAR DEGENERATION IN A PATIENT WITH ACERULOPLASMINEMIA. *Retina*, **2019** , 39, 1824-1828	3.6	3

70	Single-nuclei RNA-seq on human retinal tissue provides improved transcriptome profiling. *Nature Communications*, **2019**, 10, 5743	17.4	44
69	Prevalence and correlates of bleeding and emotional harms in a national US sample of patients with venous thromboembolism: A cross-sectional structural equation model. *Thrombosis Research*, **2018**, 172, 181-187	8.2	12
68	Genetic Risk Factors for Radiation Vasculopathy **2018**, 59, 1547-1553		2
67	The nuclear hormone receptor gene Nr2c1 (Tr2) is a critical regulator of early retina cell patterning. *Developmental Biology*, **2017**, 429, 343-355	3.1	9
66	Retinopathy of prematurity: A comprehensive risk analysis for prevention and prediction of disease. *PLoS ONE*, **2017**, 12, e0171467	3.7	16
65	Animal Models of Diabetic Retinopathy. *Current Diabetes Reports*, **2017**, 17, 93	5.6	98
64	ER stress-induced aggresome trafficking of HtrA1 protects against proteotoxicity. *Journal of Molecular Cell Biology*, **2017**, 9, 516-532	6.3	6
63	Factors predicting self-reported medication low adherence in a large sample of adults in the US general population: a cross-sectional study. *BMJ Open*, **2017**, 7, e014435	3	16
62	RNA expression in human retina. *Human Molecular Genetics*, **2017**, 26, R68-R74	5.6	8
61	Genetics of age-related macular degeneration (AMD). *Human Molecular Genetics*, **2017**, 26, R45-R50	5.6	53
60	Meta-analysis of gene-environment-wide association scans accounting for education level identifies additional loci for refractive error. *Nature Communications*, **2016**, 7, 11008	17.4	79
59	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nature Genetics*, **2016**, 48, 134-43	36.3	769
58	Adherence to Glaucoma Medications Over 12 Months in Two US Community Pharmacy Chains. *Journal of Clinical Medicine*, **2016**, 5,	5.1	19
57	Epidemiology of age-related macular degeneration (AMD): associations with cardiovascular disease phenotypes and lipid factors. *Eye and Vision (London, England)*, **2016**, 3, 34	4.9	223
56	Genetic variants in complement pathway and ARMS2/HTRA1 genes and risk of age-related macular degeneration in a homogeneous population from central Greece. *Ophthalmic Genetics*, **2016**, 37, 339-44	1.2	2
55	Clinical Characteristics of Uveal Melanoma in Patients With Germline BAP1 Mutations. *JAMA Ophthalmology*, **2015**, 133, 881-7	3.9	83
54	Epigenetic Mechanisms of the Aging Human Retina. *Journal of Experimental Neuroscience*, **2015**, 9, 51-79	3.6	25
53	Quality of Life with Macular Degeneration Is Not as Dark as It May Seem: Patients' Perceptions of the MacDQoL Questionnaire. *Journal of Clinical Medicine*, **2015**, 4, 1841-52	5.1	3

52	Ancestry of the Timorese: age-related macular degeneration associated genotype and allele sharing among human populations from throughout the world. *Frontiers in Genetics*, **2015**, 6, 238	4.5	4
51	Analysis of Genetic and Environmental Risk Factors and Their Interactions in Korean Patients with Age-Related Macular Degeneration. *PLoS ONE*, **2015**, 10, e0132771	3.7	29
50	Genetic variants associated with severe retinopathy of prematurity in extremely low birth weight infants **2014**, 55, 6194-203		35
49	Search for age-related macular degeneration risk variants in Alzheimer disease genes and pathways. *Neurobiology of Aging*, **2014**, 35, 1510.e7-18	5.6	34
48	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. *Human Molecular Genetics*, **2014**, 23, 5827-37	5.6	34
47	Differential Gene Expression in Age-Related Macular Degeneration. *Cold Spring Harbor Perspectives in Medicine*, **2014**, 5, a017210	5.4	14
46	Plasma homocysteine and genetic variants of homocysteine metabolism enzymes in patients from central Greece with primary open-angle glaucoma and pseudoexfoliation glaucoma. *Clinical Ophthalmology*, **2014**, 8, 1819-25	2.5	10
45	FLT1 genetic variation predisposes to neovascular AMD in ethnically diverse populations and alters systemic FLT1 expression **2014**, 55, 3543-54		14
44	Prospective study of common variants in CX3CR1 and risk of macular degeneration: pooled analysis from 5 long-term studies. *JAMA Ophthalmology*, **2014**, 132, 84-95	3.9	22
43	Modifier genes as therapeutics: the nuclear hormone receptor Rev Erb alpha (Nr1d1) rescues Nr2e3 associated retinal disease. *PLoS ONE*, **2014**, 9, e87942	3.7	25
42	Seven new loci associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 433-9, 439e1-2	36.3	577
41	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 1375-9	36.3	130
40	Age-related macular degeneration-associated silent polymorphisms in HtrA1 impair its ability to antagonize insulin-like growth factor 1. *Molecular and Cellular Biology*, **2013**, 33, 1976-90	4.8	17
39	An inherited disorder with splenomegaly, cytopenias, and vision loss. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 475-81	2.5	3
38	Germline BAP1 inactivation is preferentially associated with metastatic ocular melanoma and cutaneous-ocular melanoma families. *PLoS ONE*, **2012**, 7, e35295	3.7	190
37	High throughput mass spectrometry-based mutation profiling of primary uveal melanoma **2012**, 53, 6991-6		38
36	Unilateral sporadic retinal dysplasia: results of histopathologic, immunohistochemical, chromosomal, genetic, and VEGF-A analyses. *Journal of AAPOS*, **2011**, 15, 579-86	1.3	4
35	Clinical validation of a genetic model to estimate the risk of developing choroidal neovascular age-related macular degeneration. *Human Genomics*, **2011**, 5, 420-40	6.8	43

34	Systems biology-based analysis implicates a novel role for vitamin D metabolism in the pathogenesis of age-related macular degeneration. *Human Genomics*, **2011**, 5, 538-68	6.8	50
33	Identifying subtypes of patients with neovascular age-related macular degeneration by genotypic and cardiovascular risk characteristics. *BMC Medical Genetics*, **2011**, 12, 83	2.1	11
32	Genetics of age-related macular degeneration: current concepts, future directions. *Seminars in Ophthalmology*, **2011**, 26, 77-93	2.4	65
31	Genetic variations strongly influence phenotypic outcome in the mouse retina. *PLoS ONE*, **2011**, 6, e21858	3.7	25
30	Influence of ROBO1 and RORA on risk of age-related macular degeneration reveals genetically distinct phenotypes in disease pathophysiology. *PLoS ONE*, **2011**, 6, e25775	3.7	22
29	Association between assisted reproductive technology and advanced retinopathy of prematurity. *Clinical Ophthalmology*, **2010**, 4, 1385-90	2.5	9
28	Genetic variants near TIMP3 and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 7401-6	11.5	417
27	Prospective study of common variants in the retinoic acid receptor-related orphan receptor gene and risk of neovascular age-related macular degeneration. *JAMA Ophthalmology*, **2010**, 128, 1462-71		22
26	Convergence of linkage, gene expression and association data demonstrates the influence of the RAR-related orphan receptor alpha (RORA) gene on neovascular AMD: a systems biology based approach. *Vision Research*, **2010**, 50, 698-715	2.1	37
25	Genotypic association analysis using discordant-relative-pairs. *Annals of Human Genetics*, **2009**, 73, 84-94	2.2	5
24	Comprehensive analysis of complement factor H and LOC387715/ARMS2/HTRA1 variants with respect to phenotype in advanced age-related macular degeneration. *American Journal of Ophthalmology*, **2009**, 148, 869-74	4.9	48
23	Alleles in the HtrA serine peptidase 1 gene alter the risk of neovascular age-related macular degeneration. *Ophthalmology*, **2008**, 115, 1209-1215.e7	7.3	80
22	Increased choroidal neovascularization following laser induction in mice lacking lysyl oxidase-like 1. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 2599-605		49
21	DNA sequence variants in the LOXL1 gene are associated with pseudoexfoliation glaucoma in a U.S. clinic-based population with broad ethnic diversity. *BMC Medical Genetics*, **2008**, 9, 5	2.1	91
20	The NEI/NCBI dbGAP database: genotypes and haplotypes that may specifically predispose to risk of neovascular age-related macular degeneration. *BMC Medical Genetics*, **2008**, 9, 51	2.1	49
19	Comprehensive analysis of CRP, CFH Y402H and environmental risk factors on risk of neovascular age-related macular degeneration. *Molecular Vision*, **2008**, 14, 1487-95	2.3	29
18	Genetics of Age-related Macular Degeneration **2008**, 1881-1900		1
17	Cigarette smoking, CFH, APOE, ELOVL4, and risk of neovascular age-related macular degeneration. *JAMA Ophthalmology*, **2007**, 125, 49-54		103

16	Retinitis pigmentosa and common variable immunodeficiency disease: associated or separate?. *Southern Medical Journal*, **2006**, 99, 914	0.6	
15	Extremely discordant sib-pair study design to determine risk factors for neovascular age-related macular degeneration. *JAMA Ophthalmology*, **2004**, 122, 575-80		38
14	Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. *Human Molecular Genetics*, **2002**, 11, 1219-27	5.6	203
13	Novel mutations in the NRL gene and associated clinical findings in patients with dominant retinitis pigmentosa. *JAMA Ophthalmology*, **2002**, 120, 369-75		48
12	Two families from New England with usher syndrome type IC with distinct haplotypes. *American Journal of Ophthalmology*, **2001**, 131, 355-8	4.9	4
11	Identification and mutation analysis of a cochlear-expressed, zinc finger protein gene at the DFNB7/11 and dn hearing-loss loci on human chromosome 9q and mouse chromosome 19. *Gene*, **1998**, 215, 461-9	3.8	29
10	Assembly of a high-resolution map of the Acadian Usher syndrome region and localization of the nuclear EF-hand acidic gene. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **1998**, 1407, 84-91	6.9	9
9	The mouse deafness locus (dn) is associated with an inversion on chromosome 19. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **1998**, 1407, 257-62	6.9	4
8	Construction of a 780-kb PAC, BAC, and cosmid contig encompassing the minimal critical deletion involved in B cell chronic lymphocytic leukemia at 13q14.3. *Genomics*, **1997**, 46, 183-90	4.3	38
7	Identification and characterization of two polymorphic Ya5 Alu repeats. *Mutation Research - Mutation Research Genomics*, **1997**, 382, 5-11		10
6	A comprehensive genetic map of the mouse genome. *Nature*, **1996**, 380, 149-52	50.4	760
5	Solid-phase reversible immobilization for the isolation of PCR products. *Nucleic Acids Research*, **1995**, 23, 4742-3	20.1	255
4	An STS-based map of the human genome. *Science*, **1995**, 270, 1945-54	33.3	718
3	Traumatic brain injury causes a decrease in M2 muscarinic cholinergic receptor binding in the rat brain. *Brain Research*, **1994**, 653, 39-44	3.7	45
2	D-aspartate uptake and release in the guinea pig spinal cord after partial ablation of the cerebral cortex. *Journal of Neurochemistry*, **1988**, 50, 103-11	6	30
1	Single-nuclei RNA-seq on human retinal tissue provides improved transcriptome profiling		4