

# Aideen M Mcinerney-Leo

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

76  
papers

2,127  
citations

24  
h-index

45  
g-index

84  
ext. papers

2,587  
ext. citations

5.6  
avg, IF

4.26  
L-index

#	Paper	IF	Citations
76	The Future of Precision Prevention for Advanced Melanoma.. <i>Frontiers in Medicine</i> , <b>2021</b> , 8, 818096	4.9	0
75	Evaluation of a Genetics Education Program for Health Interpreters: A Pilot Study.. <i>Frontiers in Genetics</i> , <b>2021</b> , 12, 771892	4.5	0
74	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, 1163-1182	5.6	13
73	The impact of Marfan syndrome on an Aboriginal Australian family: P donR like it as much as I donR like cancerP. <i>Journal of Genetic Counseling</i> , <b>2021</b> ,	2.5	1
72	Communicating polygenic risk scores in the familial breast cancer clinic. <i>Patient Education and Counseling</i> , <b>2021</b> , 104, 2512-2521	3.1	2
71	Genomic Risk Score for Melanoma in a Prospective Study of Older Individuals. <i>Journal of the National Cancer Institute</i> , <b>2021</b> , 113, 1379-1385	9.7	4
70	A family with partially penetrant multicentric carpotarsal osteolysis due to gonadal mosaicism: First reported case. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 2477-2481	2.5	0
69	Study protocol: the Australian genetics and life insurance moratorium-monitoring the effectiveness and response (A-GLIMMER) project. <i>BMC Medical Ethics</i> , <b>2021</b> , 22, 63	2.9	1
68	Anatomic Distribution of Cherry Angiomas in the General Population. <i>Dermatology</i> , <b>2021</b> , 1-9	4.4	0
67	Waiting for a diagnosis in Rubinstein-Taybi: The journey from "ignorance is bliss" to the value of "a label". <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 105-111	2.5	0
66	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma I: Primary and Secondary Preventative Behaviours. <i>Dermatology</i> , <b>2021</b> , 237, 806-815	4.4	1
65	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma II: Psychosocial Outcomes and Attitudes. <i>Dermatology</i> , <b>2021</b> , 237, 816-826	4.4	1
64	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbBB role in oncogenesis. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 2393-2401	5.6	0
63	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. <i>JCI Insight</i> , <b>2021</b> , 6,	9.9	3
62	Australian human research ethics committee membersPconfidence in reviewing genomic research applications. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 1811-1818	5.3	0
61	Queensland ConsumersPAwareness and Understanding of Clinical Genetics Services. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 537743	4.5	3
60	Causal Attributions in an Australian Aboriginal Family With Marfan Syndrome: A Qualitative Study. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 461	4.5	1

59	CDKN2A testing threshold in a high-risk Australian melanoma cohort: number of primaries, family history and young age of onset impact risk. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2020</b> , 34, e797-e798	4.6	
58	The emerging field of polygenic risk scores and perspective for use in clinical care. <i>Human Molecular Genetics</i> , <b>2020</b> , 29, R165-R176	5.6	16
57	Compound heterozygous mutations in FBN1 in a large family with Marfan syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1116	2.3	4
56	Use of the arm-span to height ratio as a criterion for Marfan syndrome in Aboriginal Australians: Diagnostically challenging. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 829-830	2.5	
55	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. <i>PLoS ONE</i> , <b>2020</b> , 15, e0238529	3.7	5
54	The interplay of sun damage and genetic risk in Australian multiple and single primary melanoma cases and controls. <i>British Journal of Dermatology</i> , <b>2020</b> , 183, 357-366	4	7
53	Factors influencing cancer genetic somatic mutation test ordering by cancer physician. <i>Journal of Translational Medicine</i> , <b>2020</b> , 18, 431	8.5	4
52	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. <i>Journal of Bone and Mineral Research</i> , <b>2020</b> , 35, 92-105	6.3	19
51	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , <b>2020</b> , 29, 3662-3678	5.6	3
50	Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. <i>Frontiers in Endocrinology</i> , <b>2020</b> , 11, 628946	5.7	4
49	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants <b>2020</b> , 15, e0238529		
48	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants <b>2020</b> , 15, e0238529		
47	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants <b>2020</b> , 15, e0238529		
46	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants <b>2020</b> , 15, e0238529		
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43	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants <b>2020</b> , 15, e0238529		
42	The personal touch: does the communication method affect response to melanoma genetic risk?. <i>British Journal of Dermatology</i> , <b>2019</b> , 180, 1288-1289	4	

41	OR28-3 A Mutation in KCNK16 Segregating with Autosomal Dominant Non-Ketotic Diabetes Drastically Increases TALK-1 Membrane Current: A Novel Gene for MODY?. <i>Journal of the Endocrine Society</i> , <b>2019</b> , 3,	0.4	78
40	Evaluation of the efficacy of 3D total-body photography with sequential digital dermoscopy in a high-risk melanoma cohort: protocol for a randomised controlled trial. <i>BMJ Open</i> , <b>2019</b> , 9, e032969	3	10
39	Comprehensive genetic screening: The prevalence of maturity-onset diabetes of the young gene variants in a population-based childhood diabetes cohort. <i>Pediatric Diabetes</i> , <b>2019</b> , 20, 57-64	3.6	13
38	Predicted Benign and Synonymous Variants in Cause Primary Adrenal Insufficiency Through Missplicing. <i>Journal of the Endocrine Society</i> , <b>2019</b> , 3, 201-221	0.4	13
37	Mutations That Alter the Carboxy-Terminal-Propeptide Cleavage Site of the Chains of Type I Procollagen Are Associated With a Unique Osteogenesis Imperfecta Phenotype. <i>Journal of Bone and Mineral Research</i> , <b>2018</b> , 33, 1260-1271	6.3	20
36	Whole-exome sequencing for mutation detection in pediatric disorders of insulin secretion: Maturity onset diabetes of the young and congenital hyperinsulinism. <i>Pediatric Diabetes</i> , <b>2018</b> , 19, 656-662	3.6	13
35	Point mutation in p14 -specific exon 1 of CDKN2A causing familial melanoma and astrocytoma. <i>British Journal of Dermatology</i> , <b>2018</b> , 178, e263-e264	4	0
34	Factors Associated with Parental Adaptation to Children with an Undiagnosed Medical Condition. <i>Journal of Genetic Counseling</i> , <b>2017</b> , 26, 829-840	2.5	21
33	Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1698-1704	2.5	9
32	Low first-trimester PAPP-A in IVF (fresh and frozen-thawed) pregnancies, likely due to a biological cause. <i>Journal of Assisted Reproduction and Genetics</i> , <b>2017</b> , 34, 1367-1375	3.4	7
31	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. <i>New England Journal of Medicine</i> , <b>2017</b> , 377, 544-552	59.2	114
30	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 392-406	11	34
29	Fryns Syndrome Associated with Recessive Mutations in PIGN in two Separate Families. <i>Human Mutation</i> , <b>2016</b> , 37, 695-702	4.7	34
28	Mutations in human C2CD3 cause skeletal dysplasia and provide new insights into phenotypic and cellular consequences of altered C2CD3 function. <i>Scientific Reports</i> , <b>2016</b> , 6, 24083	4.9	24
27	Mutations in LTBP3 cause acromicric dysplasia and geleophysic dysplasia. <i>Journal of Medical Genetics</i> , <b>2016</b> , 53, 457-64	5.8	34
26	Psychological Impact of Predictive Genetic Testing in VCP Inclusion Body Myopathy, Paget Disease of Bone and Frontotemporal Dementia. <i>Journal of Genetic Counseling</i> , <b>2015</b> , 24, 842-50	2.5	12
25	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1234-42	5.6	30
24	COL1A1 C-propeptide cleavage site mutation causes high bone mass, bone fragility and jaw lesions: a new cause of gnathodiaphyseal dysplasia?. <i>Clinical Genetics</i> , <b>2015</b> , 88, 49-55	4	12

23	Whole exome sequencing is an efficient, sensitive and specific method for determining the genetic cause of short-rib thoracic dystrophies. <i>Clinical Genetics</i> , <b>2015</b> , 88, 550-7	4	30
22	The IFITM5 mutation c.-14C > T results in an elongated transcript expressed in human bone; and causes varying phenotypic severity of osteogenesis imperfecta type V. <i>BMC Musculoskeletal Disorders</i> , <b>2014</b> , 15, 107	2.8	15
21	Whole exome sequencing is an efficient and sensitive method for detection of germline mutations in patients with pheochromocytomas and paragangliomas. <i>Clinical Endocrinology</i> , <b>2014</b> , 80, 25-33	3.4	32
20	Short-rib polydactyly and Jeune syndromes are caused by mutations in WDR60. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 515-23	11	92
19	Mutations in the gene encoding IFT dynein complex component WDR34 cause Jeune asphyxiating thoracic dystrophy. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 932-44	11	84
18	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 915-25	11	155
17	Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in osteogenesis imperfecta and Marfan syndrome. <i>BoneKEy Reports</i> , <b>2013</b> , 2, 456		17
16	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1625-31	5.6	70
15	Uptake of invasive prenatal tests in pregnancies conceived via assisted reproductive technologies: the experience in Queensland, Australia. <i>Prenatal Diagnosis</i> , <b>2012</b> , 32, 1049-52	3.2	3
14	Comprehensive screening of a North American Parkinson $\beta$ disease cohort for LRRK2 mutation. <i>Neurodegenerative Diseases</i> , <b>2007</b> , 4, 386-91	2.3	23
13	BRCA1/2 testing in hereditary breast and ovarian cancer families III: risk perception and screening. <i>American Journal of Medical Genetics, Part A</i> , <b>2006</b> , 140, 2198-206	2.5	26
12	Truth-telling and Turner Syndrome: the importance of diagnostic disclosure. <i>Journal of Pediatrics</i> , <b>2006</b> , 148, 102-7	3.6	38
11	Midbrain dopamine and prefrontal function in humans: interaction and modulation by COMT genotype. <i>Nature Neuroscience</i> , <b>2005</b> , 8, 594-6	25.5	360
10	BRCA1/2 testing in hereditary breast and ovarian cancer families II: impact on relationships. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 133A, 165-9	2.5	32
9	Turner syndrome: four challenges across the lifespan. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 139A, 57-66	2.5	67
8	Clinical and positron emission tomography of Parkinson $\beta$ disease caused by LRRK2. <i>Annals of Neurology</i> , <b>2005</b> , 57, 453-6	9.4	88
7	Genetic testing in Parkinson $\beta$ disease. <i>Movement Disorders</i> , <b>2005</b> , 20, 1-10	7	38
6	Genetic testing in Parkinson $\beta$ disease. <i>Movement Disorders</i> , <b>2005</b> , 20, 908-9	7	9

5	SNCA multiplication is not a common cause of Parkinson disease or dementia with Lewy bodies. <i>Neurology</i> , <b>2004</b> , 63, 554-6	6.5	56
4	Parkinsonism among Gaucher disease carriers. <i>Journal of Medical Genetics</i> , <b>2004</b> , 41, 937-40	5.8	257
3	BRCA1/2 testing in hereditary breast and ovarian cancer families: effectiveness of problem-solving training as a counseling intervention. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 130A, 221-7		34
2	Prevalence of Parkinson disease in populations of African ancestry: a review. <i>Journal of the National Medical Association</i> , <b>2004</b> , 96, 974-9	2.3	30
1	The ethical protection of genetic information: procedure analysis for psychologists. <i>Clinical Psychologist</i> , 1-10	1.6	1