Aideen M Mcinerney-Leo

List of Publications by Citations

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	Midbrain dopamine and prefrontal function in humans: interaction and modulation by COMT genotype. *Nature Neuroscience*, **2005**, 8, 594-6	25.5	360
75	Parkinsonism among Gaucher disease carriers. *Journal of Medical Genetics*, **2004**, 41, 937-40	5.8	257
74	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. *American Journal of Human Genetics*, **2013**, 93, 915-25	11	155
73	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. *New England Journal of Medicine*, **2017**, 377, 544-552	59.2	114
72	Short-rib polydactyly and Jeune syndromes are caused by mutations in WDR60. *American Journal of Human Genetics*, **2013**, 93, 515-23	11	92
71	Clinical and positron emission tomography of Parkinson disease caused by LRRK2. *Annals of Neurology*, **2005**, 57, 453-6	9.4	88
70	Mutations in the gene encoding IFT dynein complex component WDR34 cause Jeune asphyxiating thoracic dystrophy. *American Journal of Human Genetics*, **2013**, 93, 932-44	11	84
69	OR28-3 A Mutation in KCNK16 Segregating with Autosomal Dominant Non-Ketotic Diabetes Drastically Increases TALK-1 Membrane Current: A Novel Gene for MODY?. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
68	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. *Human Molecular Genetics*, **2013**, 22, 1625-31	5.6	70
67	Turner syndrome: four challenges across the lifespan. *American Journal of Medical Genetics, Part A*, **2005**, 139A, 57-66	2.5	67
66	SNCA multiplication is not a common cause of Parkinson disease or dementia with Lewy bodies. *Neurology*, **2004**, 63, 554-6	6.5	56
65	Truth-telling and Turner Syndrome: the importance of diagnostic disclosure. *Journal of Pediatrics*, **2006**, 148, 102-7	3.6	38
64	Genetic testing in Parkinson disease. *Movement Disorders*, **2005**, 20, 1-10	7	38
63	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. *American Journal of Human Genetics*, **2016**, 99, 392-406	11	34
62	BRCA1/2 testing in hereditary breast and ovarian cancer families: effectiveness of problem-solving training as a counseling intervention. *American Journal of Medical Genetics Part A*, **2004**, 130A, 221-7		34
61	Fryns Syndrome Associated with Recessive Mutations in PIGN in two Separate Families. *Human Mutation*, **2016**, 37, 695-702	4.7	34
60	Mutations in LTBP3 cause acromicric dysplasia and geleophysic dysplasia. *Journal of Medical Genetics*, **2016**, 53, 457-64	5.8	34

59	Whole exome sequencing is an efficient and sensitive method for detection of germline mutations in patients with phaeochromcytomas and paragangliomas. *Clinical Endocrinology*, **2014**, 80, 25-33	3.4	32
58	BRCA1/2 testing in hereditary breast and ovarian cancer families II: impact on relationships. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 165-9	2.5	32
57	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. *Human Molecular Genetics*, **2015**, 24, 1234-42	5.6	30
56	Whole exome sequencing is an efficient, sensitive and specific method for determining the genetic cause of short-rib thoracic dystrophies. *Clinical Genetics*, **2015**, 88, 550-7	4	30
55	Prevalence of Parkinson's disease in populations of African ancestry: a review. *Journal of the National Medical Association*, **2004**, 96, 974-9	2.3	30
54	BRCA1/2 testing in hereditary breast and ovarian cancer families III: risk perception and screening. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2198-206	2.5	26
53	Mutations in human C2CD3 cause skeletal dysplasia and provide new insights into phenotypic and cellular consequences of altered C2CD3 function. *Scientific Reports*, **2016**, 6, 24083	4.9	24
52	Comprehensive screening of a North American Parkinson's disease cohort for LRRK2 mutation. *Neurodegenerative Diseases*, **2007**, 4, 386-91	2.3	23
51	Factors Associated with Parental Adaptation to Children with an Undiagnosed Medical Condition. *Journal of Genetic Counseling*, **2017**, 26, 829-840	2.5	21
50	Mutations That Alter the Carboxy-Terminal-Propeptide Cleavage Site of the Chains of Type I Procollagen Are Associated With a Unique Osteogenesis Imperfecta Phenotype. *Journal of Bone and Mineral Research*, **2018**, 33, 1260-1271	6.3	20
49	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. *Journal of Bone and Mineral Research*, **2020**, 35, 92-105	6.3	19
48	Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in osteogenesis imperfecta and Marfan syndrome. *BoneKEy Reports*, **2013**, 2, 456		17
47	The emerging field of polygenic risk scores and perspective for use in clinical care. *Human Molecular Genetics*, **2020**, 29, R165-R176	5.6	16
46	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. *BMC Musculoskeletal Disorders*, **2014**, 15, 107	2.8	15
45	Whole-exome sequencing for mutation detection in pediatric disorders of insulin secretion: Maturity onset diabetes of the young and congenital hyperinsulinism. *Pediatric Diabetes*, **2018**, 19, 656-662	3.6	13
44	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 1163-1182	5.6	13
43	Comprehensive genetic screening: The prevalence of maturity-onset diabetes of the young gene variants in a population-based childhood diabetes cohort. *Pediatric Diabetes*, **2019**, 20, 57-64	3.6	13
42	Predicted Benign and Synonymous Variants in Cause Primary Adrenal Insufficiency Through Missplicing. *Journal of the Endocrine Society*, **2019**, 3, 201-221	0.4	13

41	Psychological Impact of Predictive Genetic Testing in VCP Inclusion Body Myopathy, Paget Disease of Bone and Frontotemporal Dementia. *Journal of Genetic Counseling*, **2015**, 24, 842-50	2.5	12
40	COL1A1 C-propeptide cleavage site mutation causes high bone mass, bone fragility and jaw lesions: a new cause of gnathodiaphyseal dysplasia?. *Clinical Genetics*, **2015**, 88, 49-55	4	12
39	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. *BMJ Open*, **2019**, 9, e032969	3	10
38	Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1698-1704	2.5	9
37	Genetic testing in Parkinson's disease. *Movement Disorders*, **2005**, 20, 908-9	7	9
36	Low first-trimester PAPP-A in IVF (fresh and frozen-thawed) pregnancies, likely due to a biological cause. *Journal of Assisted Reproduction and Genetics*, **2017**, 34, 1367-1375	3.4	7
35	The interplay of sun damage and genetic risk in Australian multiple and single primary melanoma cases and controls. *British Journal of Dermatology*, **2020**, 183, 357-366	4	7
34	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. *PLoS ONE*, **2020**, 15, e0238529	3.7	5
33	Compound heterozygous mutations in FBN1 in a large family with Marfan syndrome. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1116	2.3	4
32	Factors influencing cancer genetic somatic mutation test ordering by cancer physician. *Journal of Translational Medicine*, **2020**, 18, 431	8.5	4
31	Genomic Risk Score for Melanoma in a Prospective Study of Older Individuals. *Journal of the National Cancer Institute*, **2021**, 113, 1379-1385	9.7	4
30	Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. *Frontiers in Endocrinology*, **2020**, 11, 628946	5.7	4
29	Queensland ConsumersPAwareness and Understanding of Clinical Genetics Services. *Frontiers in Genetics*, **2020**, 11, 537743	4.5	3
28	Uptake of invasive prenatal tests in pregnancies conceived via assisted reproductive technologies: the experience in Queensland, Australia. *Prenatal Diagnosis*, **2012**, 32, 1049-52	3.2	3
27	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. *Human Molecular Genetics*, **2020**, 29, 3662-3678	5.6	3
26	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. *JCI Insight*, **2021**, 6,	9.9	3
25	Communicating polygenic risk scores in the familial breast cancer clinic. *Patient Education and Counseling*, **2021**, 104, 2512-2521	3.1	2
24	Causal Attributions in an Australian Aboriginal Family With Marfan Syndrome: A Qualitative Study. *Frontiers in Genetics*, **2020**, 11, 461	4.5	1

23	The ethical protection of genetic information: procedure analysis for psychologists. *Clinical Psychologist*,1-10	1.6	1
22	The impact of Marfan syndrome on an Aboriginal Australian family: P donP like it as much as I donP like cancerP. *Journal of Genetic Counseling*, **2021**,	2.5	1
21	Study protocol: the Australian genetics and life insurance moratorium-monitoring the effectiveness and response (A-GLIMMER) project. *BMC Medical Ethics*, **2021**, 22, 63	2.9	1
20	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma I: Primary and Secondary Preventative Behaviours. *Dermatology*, **2021**, 237, 806-815	4.4	1
19	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma II: Psychosocial Outcomes and Attitudes. *Dermatology*, **2021**, 237, 816-826	4.4	1
18	Point mutation in p14 -specific exon 1 of CDKN2A causing familial melanoma and astrocytoma. *British Journal of Dermatology*, **2018**, 178, e263-e264	4	0
17	The Future of Precision Prevention for Advanced Melanoma.. *Frontiers in Medicine*, **2021**, 8, 818096	4.9	0
16	Evaluation of a Genetics Education Program for Health Interpreters: A Pilot Study.. *Frontiers in Genetics*, **2021**, 12, 771892	4.5	0
15	A family with partially penetrant multicentric carpotarsal osteolysis due to gonadal mosaicism: First reported case. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2477-2481	2.5	0
14	Anatomic Distribution of Cherry Angiomas in the General Population. *Dermatology*, **2021**, 1-9	4.4	0
13	Waiting for a diagnosis in Rubinstein-Taybi: The journey from "ignorance is bliss" to the value of "a label". *American Journal of Medical Genetics, Part A*, **2021**, 185, 105-111	2.5	0
12	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbBP role in oncogenesis. *Human Molecular Genetics*, **2021**, 30, 2393-2401	5.6	0
11	Australian human research ethics committee membersP confidence in reviewing genomic research applications. *European Journal of Human Genetics*, **2021**, 29, 1811-1818	5.3	0
10	The personal touch: does the communication method affect response to melanoma genetic risk?. *British Journal of Dermatology*, **2019**, 180, 1288-1289	4	
9	CDKN2A testing threshold in a high-risk Australian melanoma cohort: number of primaries, family history and young age of onset impact risk. *Journal of the European Academy of Dermatology and Venereology*, **2020**, 34, e797-e798	4.6	
8	Use of the arm-span to height ratio as a criterion for Marfan syndrome in Aboriginal Australians: Diagnostically challenging. *American Journal of Medical Genetics, Part A*, **2020**, 182, 829-830	2.5	
7	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
6	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		

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1 Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529