Aideen M Mcinerney-Leo

List of Publications by Year
in descending order

77 papers	2,795 citations	236612 25 h-index	182168 51 g-index
84 all docs	84 docs citations	84 times ranked	5296 citing authors

#	Article	IF	Citations
1	Midbrain dopamine and prefrontal function in humans: interaction and modulation by COMT genotype. Nature Neuroscience, 2005, 8, 594-596.	7.1	402
2	Parkinsonism among Gaucher disease carriers. Journal of Medical Genetics, 2004, 41, 937-940.	1.5	320
3	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196
4	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.	13.9	177
5	Short-Rib Polydactyly and Jeune Syndromes Are Caused by Mutations in WDR60. American Journal of Human Genetics, 2013, 93, 515-523.	2.6	116
6	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2013, 93, 932-944.	2.6	108
7	Clinical and positron emission tomography of Parkinson's disease caused byLRRK2. Annals of Neurology, 2005, 57, 453-456.	2.8	105
8	Turner syndrome: Four challenges across the lifespan. American Journal of Medical Genetics, Part A, 2005, 139A, 57-66.	0.7	99
9	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. Human Molecular Genetics, 2013, 22, 1625-1631.	1.4	87
10	SNCA multiplication is not a common cause of Parkinson disease or dementia with Lewy bodies. Neurology, 2004, 63, 554-556.	1.5	66
11	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	2.6	52
12	Mutations inLTBP3cause acromicric dysplasia and geleophysic dysplasia. Journal of Medical Genetics, 2016, 53, 457-464.	1.5	50
13	Genetic testing in Parkinson's disease. Movement Disorders, 2005, 20, 1-10.	2.2	48
14	Truth-telling and Turner Syndrome: The Importance of Diagnostic Disclosure. Journal of Pediatrics, 2006, 148, 102-107.	0.9	47
15	The emerging field of polygenic risk scores and perspective for use in clinical care. Human Molecular Genetics, 2020, 29, R165-R176.	1.4	46
16	Whole exome sequencing is an efficient, sensitive and specific method for determining the genetic cause of shortâ€rib thoracic dystrophies. Clinical Genetics, 2015, 88, 550-557.	1.0	45
17	Fryns Syndrome Associated with Recessive Mutations in PIGN in two Separate Families. Human Mutation, 2016, 37, 695-702.	1.1	43
18	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e1163-e1182.	1.8	43

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19	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, 221-227.	2.4	40
20	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. Human Molecular Genetics, 2015, 24, 1234-1242.	1.4	39
21	BRCA1/2 testing in hereditary breast and ovarian cancer families II: Impact on relationships. American Journal of Medical Genetics, Part A, 2005, 133A, 165-169.	0.7	36
22	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.	1.2	35
23	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMADâ€Ðependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. Journal of Bone and Mineral Research, 2020, 35, 92-105.	3.1	34
24	Prevalence of Parkinson's disease in populations of African ancestry: a review. Journal of the National Medical Association, 2004, 96, 974-9.	0.6	34
25	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.	1.6	30
26	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.	1.2	30
27	Factors Associated with Parental Adaptation to Children with an Undiagnosed Medical Condition. Journal of Genetic Counseling, 2017, 26, 829-840.	0.9	28
28	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.	0.8	27
29	Predicted Benign and Synonymous Variants in CYP11A1 Cause Primary Adrenal Insufficiency Through Missplicing. Journal of the Endocrine Society, 2019, 3, 201-221.	0.1	27
30	BRCA1/2 testing in hereditary breast and ovarian cancer families III: Risk perception and screening. American Journal of Medical Genetics, Part A, 2006, 140A, 2198-2206.	0.7	26
31	Comprehensive Screening of a North American Parkinsonâ€™s Disease Cohort for <i>LRRK2</i> Mutation. Neurodegenerative Diseases, 2007, 4, 386-391.	0.8	25
32	Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in osteogenesis imperfecta and Marfan syndrome. BoneKEy Reports, 2013, 2, 456.	2.7	24
33	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.	3.1	21
34	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.	0.8	20
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.	1.4	17
36	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. JCI Insight, 2021, 6, .	2.3	17

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37	<scp>COL1A1</scp> Câ€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.	1.0	15
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.	0.7	15
39	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.	1.2	15
40	Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. Frontiers in Endocrinology, 2020, 11, 628946.	1.5	15
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, 842-850.	0.9	14
42	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	1.4	14
43	Genomic Risk Score for Melanoma in a Prospective Study of Older Individuals. Journal of the National Cancer Institute, 2021, 113, 1379-1385.	3.0	14
44	Communicating polygenic risk scores in the familial breast cancer clinic. Patient Education and Counseling, 2021, 104, 2512-2521.	1.0	12
45	Study protocol: the Australian genetics and life insurance moratoriumâ€"monitoring the effectiveness and response (A-GLIMMER) project. BMC Medical Ethics, 2021, 22, 63.	1.0	12
46	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. PLoS ONE, 2020, 15, e0238529.	1.1	12
47	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.	1.2	11
48	Factors influencing cancer genetic somatic mutation test ordering by cancer physician. Journal of Translational Medicine, 2020, 18, 431.	1.8	11
49	Genetic testing in Parkinson's disease. Movement Disorders, 2005, 20, 908-909.	2.2	9
50	The Future of Precision Prevention for Advanced Melanoma. Frontiers in Medicine, 2021, 8, 818096.	1.2	7
51	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma I: Primary and Secondary Preventative Behaviours. Dermatology, 2021, 237, 806-815.	0.9	6
52	A step forward, but still inadequate: Australian health professionalsâ€™ views on the genetics and life insurance moratorium. Journal of Medical Genetics, 2022, 59, 817-826.	1.5	6
53	Compound heterozygous mutations in FBN1 in a large family with Marfan syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1116.	0.6	5
54	A Systematic Review on the Impact of Genetic Testing for Familial Melanoma II: Psychosocial Outcomes and Attitudes. Dermatology, 2021, 237, 816-826.	0.9	5

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55	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.	0.7	5
56	Anatomic Distribution of Cherry Angiomas in the General Population. Dermatology, 2022, 238, 18-26.	0.9	5
57	Uptake of invasive prenatal tests in pregnancies conceived via assisted reproductive technologies: the experience in Queensland, Australia. Prenatal Diagnosis, 2012, 32, 1049-1052.	1.1	4
58	Queensland Consumersâ€™ Awareness and Understanding of Clinical Genetics Services. Frontiers in Genetics, 2020, 11, 537743.	1.1	4
59	Waiting for a diagnosis in Rubinsteinâ€“Taybi: The journey from â€œignorance is blissâ€• to the value of â€œa labelâ€• American Journal of Medical Genetics, Part A, 2021, 185, 105-111.	0.7	3
60	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbBâ€™s role in oncogenesis. Human Molecular Genetics, 2021, 30, 2393-2401.	1.4	3
61	Point mutation in p14ARF-specific exon 1Î² of CDKN2A causing familial melanoma and astrocytoma. British Journal of Dermatology, 2018, 178, e263-e264.	1.4	2
62	CDKN2A testing threshold in a highâ€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.	1.3	2
63	Australian human research ethics committee membersâ€™ confidence in reviewing genomic research applications. European Journal of Human Genetics, 2021, 29, 1811-1818.	1.4	2
64	The impact of Marfan syndrome on an Aboriginal Australian family: â€˜I donâ€™t like it as much as I donâ€™t like cancerâ€™. Journal of Genetic Counseling, 2021, , .	0.9	2
65	Evaluation of a Genetics Education Program for Health Interpreters: A Pilot Study. Frontiers in Genetics, 2021, 12, 771892.	1.1	2
66	Causal Attributions in an Australian Aboriginal Family With Marfan Syndrome: A Qualitative Study. Frontiers in Genetics, 2020, 11, 461.	1.1	1
67	The ethical protection of genetic information: procedure analysis for psychologists. Clinical Psychologist, 0, , 1-10.	0.5	1
68	The personal touch: does the communication method affect response to melanoma genetic risk?. British Journal of Dermatology, 2019, 180, 1288-1289.	1.4	0
69	Use of the armâ€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.	0.7	0
70	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, .	0.1	0
71	Title is missing!. , 2020, 15, e0238529.		0
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73	Title is missing!. , 2020, 15, e0238529.		0
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