

Margaret G Au

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

1,690
citations

430874

18
h-index

434195

31
g-index

35
all docs

35
docs citations

35
times ranked

2761
citing authors

#	ARTICLE	IF	CITATIONS
1	Oligogenic basis of isolated gonadotropin-releasing hormone deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 15140-15144.	7.1	313
2	TAC3/TACR3 Mutations Reveal Preferential Activation of Gonadotropin-Releasing Hormone Release by Neurokinin B in Neonatal Life Followed by Reversal in Adulthood. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2857-2867.	3.6	250
3	Congenital Idiopathic Hypogonadotropic Hypogonadism: Evidence of Defects in the Hypothalamus, Pituitary, and Testes. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3019-3027.	3.6	115
4	Olfactory Phenotypic Spectrum in Idiopathic Hypogonadotropic Hypogonadism: Pathophysiological and Genetic Implications. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E136-E144.	3.6	100
5	Expanding the Phenotype and Genotype of Female GnRH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E566-E576.	3.6	97
6	The Role of the Prokineticin 2 Pathway in Human Reproduction: Evidence from the Study of Human and Murine Gene Mutations. Endocrine Reviews, 2011, 32, 225-246.	20.1	95
7	GRIN1 mutation associated with intellectual disability alters NMDA receptor trafficking and function. Journal of Human Genetics, 2017, 62, 589-597.	2.3	81
8	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
9	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants in <i>KISS1</i> / <i>Kiss1</i> . Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1771-E1781.	3.6	59
10	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
11	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
12	When Genetic Load Does Not Correlate with Phenotypic Spectrum: Lessons from the GnRH Receptor (<i>GNRHR</i>). Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1798-E1807.	3.6	43
13	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
14	Genetic counseling for isolated GnRH deficiency. Molecular and Cellular Endocrinology, 2011, 346, 102-109.	3.2	37
15	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
16	An ancient founder mutation in PROKR2 impairs human reproduction. Human Molecular Genetics, 2012, 21, 4314-4324.	2.9	31
17	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
18	A Rapid and Sensitive Next-Generation Sequencing Method to Detect RB1 Mutations Improves Care for Retinoblastoma Patients and Their Families. Journal of Molecular Diagnostics, 2016, 18, 480-493.	2.8	26

#	ARTICLE	IF	CITATIONS
19	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. <i>Genetics in Medicine</i> , 2019, 21, 2036-2042.	2.4	23
20	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. <i>Genetics in Medicine</i> , 2020, 22, 878-888.	2.4	22
21	Evaluating co-created patient-facing materials to increase understanding of genetic test results. <i>Journal of Genetic Counseling</i> , 2021, 30, 598-605.	1.6	19
22	A Family History Demonstration Project Among Women in an Urban Appalachian Community. <i>Progress in Community Health Partnerships: Research, Education, and Action</i> , 2009, 3, 155-163.	0.3	18
23	Novel pathogenic <i>COX20</i> variants causing dysarthria, ataxia, and sensory neuropathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 154-160.	3.7	17
24	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. <i>Neurogenetics</i> , 2019, 20, 129-143.	1.4	16
25	<i>FARS2</i> mutations presenting with pure spastic paraplegia and lesions of the dentate nuclei. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1128-1133.	3.7	14
26	When moments matter: Finding answers with rapid exome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1027.	1.2	12
27	Decisions to Seek Healthcare Based on Family Health History Among Urban Appalachian Women. <i>Journal of Genetic Counseling</i> , 2009, 18, 534-550.	1.6	10
28	Non-Cystic Fibrosis-Related Meconium Ileus: GUCY2C-Associated Disease Discovered through Rapid Neonatal Whole-Exome Sequencing. <i>Journal of Pediatrics</i> , 2019, 211, 207-210.	1.8	5
29	<i>SEMA6B</i> variants cause intellectual disability and alter dendritic spine density and axon guidance. <i>Human Molecular Genetics</i> , 2022, 31, 3325-3340.	2.9	5
30	Expanding the Phenotype and Genotype of Female GnRH Deficiency. <i>Endocrine Reviews</i> , 2011, 32, 156-157.	20.1	1
31	GnRH-Deficient Phenotypes in Humans and Mice With Heterozygous Variants in <i>KISS1/Kiss1</i> . <i>Obstetrical and Gynecological Survey</i> , 2012, 67, 546-547.	0.4	0
32	Spermatogenesis and Assisted Fertility Treatment. , 2019, , 903-923.		0
33	Partners in care. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2630-2632.	1.2	0