Margaret G Au

List of Publications by Year
in descending order

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	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
2	Evaluating coâ€created patientâ€facing materials to increase understanding of genetic test results. Journal of Genetic Counseling, 2021, 30, 598-605.	1.6	19
3	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
4	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
5	Partners in care. American Journal of Medical Genetics, Part A, 2021, 185, 2630-2632.	1.2	0
6	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
7	When moments matter: Finding answers with rapid exome sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1027.	1.2	12
8	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
9	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
10	Non-Cystic Fibrosisâ€'Related Meconium Ileus: GUCY2C-Associated Disease Discovered through Rapid Neonatal Whole-Exome Sequencing. Journal of Pediatrics, 2019, 211, 207-210.	1.8	5
11	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
12	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	1.4	16
13	Spermatogenesis and Assisted Fertility Treatment. , 2019, , 903-923.		0
14	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
15	Novel pathogenic <scp>COX</scp>20 variants causing dysarthria, ataxia, and sensory neuropathy. Annals of Clinical and Translational Neurology, 2019, 6, 154-160.	3.7	17
16	FARS2 mutations presenting with pure spastic paraplegia and lesions of the dentate nuclei. Annals of Clinical and Translational Neurology, 2018, 5, 1128-1133.	3.7	14
17	GRIN1 mutation associated with intellectual disability alters NMDA receptor trafficking and function. Journal of Human Genetics, 2017, 62, 589-597.	2.3	81
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	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
20	When Genetic Load Does Not Correlate with Phenotypic Spectrum: Lessons from the GnRH Receptor (GNRHR). Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1798-E1807.	3.6	43
21	Olfactory Phenotypic Spectrum in Idiopathic Hypogonadotropic Hypogonadism: Pathophysiological and Genetic Implications. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E136-E144.	3.6	100
22	An ancient founder mutation in PROKR2 impairs human reproduction. Human Molecular Genetics, 2012, 21, 4314-4324.	2.9	31
23	GnRH-Deficient Phenotypes in Humans and Mice With Heterozygous Variants in KISS1/Kiss1. Obstetrical and Gynecological Survey, 2012, 67, 546-547.	0.4	0
24	Genetic counseling for isolated GnRH deficiency. Molecular and Cellular Endocrinology, 2011, 346, 102-109.	3.2	37
25	Expanding the Phenotype and Genotype of Female GnRH Deficiency. Endocrine Reviews, 2011, 32, 156-157.	20.1	1
26	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
27	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants inKISS1/Kiss1. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1771-E1781.	3.6	59
28	Expanding the Phenotype and Genotype of Female GnRH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E566-E576.	3.6	97
29	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
30	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
31	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
32	A Family History Demonstration Project Among Women in an Urban Appalachian Community. Progress in Community Health Partnerships: Research, Education, and Action, 2009, 3, 155-163.	0.3	18
33	Decisions to Seek Healthcare Based on Family Health History Among Urban Appalachian Women. Journal of Genetic Counseling, 2009, 18, 534-550.	1.6	10