

# Wendy A Gold

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8046667/publications.pdf>

Version: 2024-02-01

31  
papers

921  
citations

686830

13  
h-index

500791

28  
g-index

40  
all docs

40  
docs citations

40  
times ranked

2193  
citing authors

#	ARTICLE	IF	CITATIONS
1	Maternal acute and chronic inflammation in pregnancy is associated with common neurodevelopmental disorders: a systematic review. <i>Translational Psychiatry</i> , 2021, 11, 71.	2.4	158
2	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. <i>Brain</i> , 2014, 137, 44-56.	3.7	143
3	Rett Syndrome: A Genetic Update and Clinical Review Focusing on Comorbidities. <i>ACS Chemical Neuroscience</i> , 2018, 9, 167-176.	1.7	79
4	A novel gene family induced by acute inflammation in endothelial cells. <i>Gene</i> , 2004, 342, 85-95.	1.0	72
5	High-Throughput <i>In Vitro</i> , <i>Ex Vivo</i> , and <i>In Vivo</i> Screen of Adeno-Associated Virus Vectors Based on Physical and Functional Transduction. <i>Human Gene Therapy</i> , 2020, 31, 575-589.	1.4	65
6	Mitochondrial dysfunction in the skeletal muscle of a mouse model of Rett syndrome (RTT): Implications for the disease phenotype. <i>Mitochondrion</i> , 2014, 15, 10-17.	1.6	51
7	MeCP2 deficiency is associated with reduced levels of tubulin acetylation and can be restored using HDAC6 inhibitors. <i>Journal of Molecular Medicine</i> , 2015, 93, 63-72.	1.7	48
8	A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. <i>Human Genetics</i> , 2012, 131, 187-200.	1.8	46
9	Whole-exome sequencing identifies novel variants in PNPT1 causing oxidative phosphorylation defects and severe multisystem disease. <i>European Journal of Human Genetics</i> , 2017, 25, 79-84.	1.4	33
10	Maternal autoimmunity and inflammation are associated with childhood tics and obsessive-compulsive disorder: Transcriptomic data show common enriched innate immune pathways. <i>Brain, Behavior, and Immunity</i> , 2021, 94, 308-317.	2.0	32
11	Mutations in RARS cause a hypomyelination disorder akin to Pelizaeus's "Merzbacher disease. <i>European Journal of Human Genetics</i> , 2017, 25, 1134-1141.	1.4	26
12	Utility of next-generation sequencing technologies for the efficient genetic resolution of haematological disorders. <i>Clinical Genetics</i> , 2016, 89, 163-172.	1.0	18
13	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (KIF1A) in patients with developmental delay. <i>Journal of Medical Genetics</i> , 2019, 56, 111-116.	1.1	16
14	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. <i>PLoS ONE</i> , 2017, 12, e0178125.	1.1	16
15	Neurological Disorders Associated with WWOX Germline Mutations: A Comprehensive Overview. <i>Cells</i> , 2021, 10, 824.	1.8	15
16	Maternal thyroid autoimmunity associated with acute-onset neuropsychiatric disorders and global regression in offspring. <i>Developmental Medicine and Child Neurology</i> , 2019, 61, 984-988.	1.1	12
17	Whole Exome Sequencing Identifies the Genetic Basis of Late-Onset Leigh Syndrome in a Patient with MRI but Little Biochemical Evidence of a Mitochondrial Disorder. <i>JIMD Reports</i> , 2016, 32, 117-124.	0.7	11
18	Emerging evidence of Toll-like receptors as a putative pathway linking maternal inflammation and neurodevelopmental disorders in human offspring: A systematic review. <i>Brain, Behavior, and Immunity</i> , 2022, 99, 91-105.	2.0	11

#	ARTICLE	IF	CITATIONS
19	The Utility of Next-Generation Sequencing in Gene Discovery for Mutation-Negative Patients with Rett Syndrome. <i>Frontiers in Cellular Neuroscience</i> , 2015, 9, 266.	1.8	10
20	Pre-clinical Investigation of Rett Syndrome Using Human Stem Cell-Based Disease Models. <i>Frontiers in Neuroscience</i> , 2021, 15, 698812.	1.4	10
21	A novel mutation in <i>GMPPA</i> in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2246-2250.	0.7	9
22	WGCNA Identifies Translational and Proteasome-Ubiquitin Dysfunction in Rett Syndrome. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9954.	1.8	9
23	Genome-wide transcriptomic and proteomic studies of Rett syndrome mouse models identify common signaling pathways and cellular functions as potential therapeutic targets. <i>Human Mutation</i> , 2019, 40, 2184-2196.	1.1	8
24	Whole exome sequencing reveals a de novo missense variant in <i>EEF1A2</i> in a Rett syndrome-like patient. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 2476-2482.	0.2	8
25	Anti-Semaphorin 4D Rescues Motor, Cognitive, and Respiratory Phenotypes in a Rett Syndrome Mouse Model. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9465.	1.8	5
26	A simple and efficient toolset for analysing mitochondrial trafficking in neuronal cells. <i>Acta Histochemica</i> , 2018, 120, 797-805.	0.9	4
27	Pathogenicity of C-terminal mutations in <i>CDKL5</i> . <i>Journal of Pediatric Epilepsy</i> , 2015, 01, 185-186.	0.1	2
28	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A ( <i>KAT6A</i> ). <i>Journal of Genetics and Genomics</i> , 2020, 47, 650-654.	1.7	2
29	Tread carefully: A functional variant in the human NADPH oxidase 4 ( <i>NOX4</i> ) is not disease causing. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 382-387.	0.5	0
30	Front Cover, Volume 40, Issue 12. <i>Human Mutation</i> , 2019, 40, i.	1.1	0
31	Breaking Boundaries in the Brain—Advances in Editing Tools for Neurogenetic Disorders. <i>Frontiers in Genome Editing</i> , 2021, 3, 623519.	2.7	0