Wendy A Gold

List of Publications by Year in descending order

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WENDY & COLD

#	Article	IF	CITATIONS
1	Maternal acute and chronic inflammation in pregnancy is associated with common neurodevelopmental disorders: a systematic review. Translational Psychiatry, 2021, 11, 71.	4.8	158
2	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	7.6	143
3	Rett Syndrome: A Genetic Update and Clinical Review Focusing on Comorbidities. ACS Chemical Neuroscience, 2018, 9, 167-176.	3.5	79
4	A novel gene family induced by acute inflammation in endothelial cells. Gene, 2004, 342, 85-95.	2.2	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. Human Gene Therapy, 2020, 31, 575-589.	2.7	65
6	Mitochondrial dysfunction in the skeletal muscle of a mouse model of Rett syndrome (RTT): Implications for the disease phenotype. Mitochondrion, 2014, 15, 10-17.	3.4	51
7	MeCP2 deficiency is associated with reduced levels of tubulin acetylation and can be restored using HDAC6 inhibitors. Journal of Molecular Medicine, 2015, 93, 63-72.	3.9	48
8	A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. Human Genetics, 2012, 131, 187-200.	3.8	46
9	Whole-exome sequencing identifies novel variants in PNPT1 causing oxidative phosphorylation defects and severe multisystem disease. European Journal of Human Genetics, 2017, 25, 79-84.	2.8	33
10	Maternal autoimmunity and inflammation are associated with childhood tics and obsessive-compulsive disorder: Transcriptomic data show common enriched innate immune pathways. Brain, Behavior, and Immunity, 2021, 94, 308-317.	4.1	32
11	Mutations in RARS cause a hypomyelination disorder akin to Pelizaeus–Merzbacher disease. European Journal of Human Genetics, 2017, 25, 1134-1141.	2.8	26
12	Utility of nextâ€generation sequencing technologies for the efficient genetic resolution of haematological disorders. Clinical Genetics, 2016, 89, 163-172.	2.0	18
13	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1	0.784314 2.5	rgBT /Overlo
14	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. PLoS ONE, 2017, 12, e0178125.	2.5	16
15	Neurological Disorders Associated with WWOX Germline Mutations—A Comprehensive Overview. Cells, 2021, 10, 824.	4.1	15
16	Maternal thyroid autoimmunity associated with acuteâ€onset neuropsychiatric disorders and global regression in offspring. Developmental Medicine and Child Neurology, 2019, 61, 984-988.	2.1	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. JIMD Reports, 2016, 32, 117-124.	1.5	11
18	Emerging evidence of Toll-like receptors as a putative pathway linking maternal inflammation and neurodevelopmental disorders in human offspring: A systematic review. Brain, Behavior, and Immunity, 2022, 99, 91-105.	4.1	11

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19	The Utility of Next-Generation Sequencing in Gene Discovery for Mutation-Negative Patients with Rett Syndrome. Frontiers in Cellular Neuroscience, 2015, 9, 266.	3.7	10
20	Pre-clinical Investigation of Rett Syndrome Using Human Stem Cell-Based Disease Models. Frontiers in Neuroscience, 2021, 15, 698812.	2.8	10
21	A novel mutation in <i>GMPPA</i> in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. American Journal of Medical Genetics, Part A, 2017, 173, 2246-2250.	1.2	9
22	WGCNA Identifies Translational and Proteasome-Ubiquitin Dysfunction in Rett Syndrome. International Journal of Molecular Sciences, 2021, 22, 9954.	4.1	9
23	Genomeâ€wide transcriptomic and proteomic studies of Rett syndrome mouse models identify common signaling pathways and cellular functions as potential therapeutic targets. Human Mutation, 2019, 40, 2184-2196.	2.5	8
24	Whole exome sequencing reveals a de novo missense variant in <i>EEF1A2</i> in a Rett syndromeâ€like patient. Clinical Case Reports (discontinued), 2019, 7, 2476-2482.	0.5	8
25	Anti-Semaphorin 4D Rescues Motor, Cognitive, and Respiratory Phenotypes in a Rett Syndrome Mouse Model. International Journal of Molecular Sciences, 2021, 22, 9465.	4.1	5
26	A simple and efficient toolset for analysing mitochondrial trafficking in neuronal cells. Acta Histochemica, 2018, 120, 797-805.	1.8	4
27	Pathogenicity of C-terminal mutations in CDKL5. Journal of Pediatric Epilepsy, 2015, 01, 185-186.	0.2	2
28	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A (KAT6A). Journal of Genetics and Genomics, 2020, 47, 650-654.	3.9	2
29	Tread carefully: A functional variant in the human NADPH oxidase 4 (NOX4) is not disease causing. Molecular Genetics and Metabolism, 2018, 123, 382-387.	1.1	0
30	Front Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, i.	2.5	0
31	Breaking Boundaries in the Brain—Advances in Editing Tools for Neurogenetic Disorders. Frontiers in Genome Editing, 2021, 3, 623519.	5.2	0