

# Lachlan A Jolly

## List of Publications by Year in descending order

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Version: 2024-02-01

35  
papers

1,679  
citations

304743

22  
h-index

395702

33  
g-index

35  
all docs

35  
docs citations

35  
times ranked

2960  
citing authors

#	ARTICLE	IF	CITATIONS
1	La FAM fatale: USP9X in development and disease. Cellular and Molecular Life Sciences, 2015, 72, 2075-2089.	5.4	145
2	Mutations in USP9X Are Associated with X-Linked Intellectual Disability and Disrupt Neuronal Cell Migration and Growth. American Journal of Human Genetics, 2014, 94, 470-478.	6.2	117
3	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	6.2	111
4	The UPF3B gene, implicated in intellectual disability, autism, ADHD and childhood onset schizophrenia regulates neural progenitor cell behaviour and neuronal outgrowth. Human Molecular Genetics, 2013, 22, 4673-4687.	2.9	101
5	Viperin is an important host restriction factor in control of Zika virus infection. Scientific Reports, 2017, 7, 4475.	3.3	98
6	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. Molecular Psychiatry, 2012, 17, 1103-1115.	7.9	97
7	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95
8	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
9	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	6.2	82
10	Loss of Usp9x Disrupts Cortical Architecture, Hippocampal Development and TGF $\beta$ -Mediated Axonogenesis. PLoS ONE, 2013, 8, e68287.	2.5	77
11	The FAM Deubiquitylating Enzyme Localizes to Multiple Points of Protein Trafficking in Epithelia, where It Associates with E-cadherin and $\beta$ -catenin. Molecular Biology of the Cell, 2004, 15, 1591-1599.	2.1	75
12	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	3.5	66
13	A Upf3b-mutant mouse model with behavioral and neurogenesis defects. Molecular Psychiatry, 2018, 23, 1773-1786.	7.9	54
14	USP9X Enhances the Polarity and Self-Renewal of Embryonic Stem Cell-derived Neural Progenitors. Molecular Biology of the Cell, 2009, 20, 2015-2029.	2.1	52
15	Pcdh19 Loss-of-Function Increases Neuronal Migration In Vitro but is Dispensable for Brain Development in Mice. Scientific Reports, 2016, 6, 26765.	3.3	52
16	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	2.9	47
17	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	8.1	43
18	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor $\beta$ Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42

#	ARTICLE	IF	CITATIONS
19	PCDH19 regulation of neural progenitor cell differentiation suggests asynchrony of neurogenesis as a mechanism contributing to PCDH19 Girls Clustering Epilepsy. <i>Neurobiology of Disease</i> , 2018, 116, 106-119.	4.4	39
20	Usp9X Controls Ankyrin-Repeat Domain Protein Homeostasis during Dendritic Spine Development. <i>Neuron</i> , 2020, 105, 506-521.e7.	8.1	34
21	Increased <i>STAG2</i> dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015, 24, 7171-7181.	2.9	28
22	USP9X deubiquitylating enzyme maintains RAPTOR protein levels, mTORC1 signalling and proliferation in neural progenitors. <i>Scientific Reports</i> , 2017, 7, 391.	3.3	27
23	Homozygous mutation of <i>STXBP5L</i> explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	2.9	25
24	Loss of <i>Usp9x</i> disrupts cell adhesion, and components of the Wnt and Notch signaling pathways in neural progenitors. <i>Scientific Reports</i> , 2017, 7, 8109.	3.3	24
25	Missense variant contribution to <i>USP9X</i> -female syndrome. <i>Npj Genomic Medicine</i> , 2020, 5, 53.	3.8	17
26	Loss of <i>FMR2</i> further emphasizes the link between deregulation of immediate early response genes <i>FOS</i> and <i>JUN</i> and intellectual disability. <i>Human Molecular Genetics</i> , 2013, 22, 2984-2991.	2.9	10
27	A synonymous <i>UPF3B</i> variant causing a speech disorder implicates NMD as a regulator of neurodevelopmental disorder gene networks. <i>Human Molecular Genetics</i> , 2020, 29, 2568-2578.	2.9	9
28	The DUB Club: Deubiquitinating Enzymes and Neurodevelopmental Disorders. <i>Biological Psychiatry</i> , 2022, 92, 614-625.	1.3	8
29	Abnormal Behavior and Cortical Connectivity Deficits in Mice Lacking <i>Usp9x</i> . <i>Cerebral Cortex</i> , 2021, 31, 1763-1775.	2.9	5
30	<i>UPF3B</i> Gene and Nonsense-Mediated mRNA Decay in Autism Spectrum Disorders. , 2014, , 1663-1678.		3
31	Robust imaging and gene delivery to study human lymphoblastoid cell lines. <i>Journal of Human Genetics</i> , 2018, 63, 945-955.	2.3	2
32	Vav Proteins in Development of the Brain: A Potential Relationship to the Pathogenesis of Congenital Zika Syndrome?. <i>Viruses</i> , 2022, 14, 386.	3.3	2
33	Protocadherin Mutations in Neurodevelopmental Disorders. , 2016, , 221-231.		1
34	Integrated in silico and experimental assessment of disease relevance of <i>PCDH19</i> missense variants. <i>Human Mutation</i> , 2021, 42, 1030-1041.	2.5	1
35	Impaired neural differentiation of MPS IIIA patient induced pluripotent stem cell-derived neural progenitor cells. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100811.	1.1	1