

# De Novo Loss-of-Function Mutations in USP9X Cause a Syndrome with Developmental Delay and Congenital M

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Escape Artists of the X Chromosome. Trends in Genetics, 2016, 32, 348-359.	2.9	144
2	An Emerging Female Phenotype with Loss-of-Function Mutations in the <i>Aristaless</i> Related Homeodomain Transcription Factor <i>ARX</i> . Human Mutation, 2017, 38, 548-555.	1.1	10
3	Screen for reactivation of MeCP2 on the inactive X chromosome identifies the BMP/TGF- $\beta$ superfamily as a regulator of XIST expression. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 1619-1624.	3.3	51
4	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	1.1	3
6	Suppressors and activators of JAK-STAT signaling at diagnosis and relapse of acute lymphoblastic leukemia in Down syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E4030-E4039.	3.3	62
7	<i>DDX3X</i> mutations in two girls with a phenotype overlapping Toriello-Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	0.7	41
8	Two females with mutations in USP9X highlight the variable expressivity of the intellectual disability syndrome. European Journal of Medical Genetics, 2017, 60, 359-364.	0.7	12
9	Gonadal mosaicism of a novel IQSEC2 variant causing female limited intellectual disability and epilepsy. European Journal of Human Genetics, 2017, 25, 763-767.	1.4	14
10	When the Lyon(ized chromosome) roars: ongoing expression from an inactive X chromosome. Philosophical Transactions of the Royal Society B: Biological Sciences, 2017, 372, 20160355.	1.8	71
13	MEF2C loss-of-function mutation contributes to congenital heart defects. International Journal of Medical Sciences, 2017, 14, 1143-1153.	1.1	27
14	A novel NR2F2 loss-of-function mutation predisposes to congenital heart defect. European Journal of Medical Genetics, 2018, 61, 197-203.	0.7	27
15	Deubiquitinase USP9X Maintains Centriolar Satellite Integrity by Stabilizing Pericentriolar Material 1 Protein. Journal of Cell Science, 2019, 132, .	1.2	20
16	Phenotypic expansion in <i>DDX3X</i> – a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	1.7	66
17	SNX17 Recruits USP9X to Antagonize MIB1-Mediated Ubiquitination and Degradation of PCM1 during Serum-Starvation-Induced Ciliogenesis. Cells, 2019, 8, 1335.	1.8	17
18	Functional analysis of deubiquitylating enzymes in tumorigenesis and development. Biochimica Et Biophysica Acta: Reviews on Cancer, 2019, 1872, 188312.	3.3	48
19	SFI1 promotes centriole duplication by recruiting USP9X to stabilize the microcephaly protein STIL. Journal of Cell Biology, 2019, 218, 2185-2197.	2.3	18
20	CYLD Regulates Centriolar Satellites Proteostasis by Counteracting the E3 Ligase MIB1. Cell Reports, 2019, 27, 1657-1665.e4.	2.9	30
21	Whole-exome sequencing reveals novel USP9X variant in female fetus with isolated agenesis of the corpus callosum. Clinical Case Reports (discontinued), 2019, 7, 656-660.	0.2	2

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22	Crystal structure and activity-based labeling reveal the mechanisms for linkage-specific substrate recognition by deubiquitinase USP9X. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 7288-7297.	3.3	39
23	Female-restricted syndromic intellectual disability in a patient from Thailand. , 2019, 179, 758-761.		8
24	RNA sequencing identifies a novel <i>USP9X</i> - <i>USP6</i> promoter swap gene fusion in a primary aneurysmal bone cyst. Genes Chromosomes and Cancer, 2019, 58, 589-594.	1.5	27
25	Anorexia nervosa is associated with Neuronatin variants. Psychiatric Genetics, 2019, 29, 103-110.	0.6	16
26	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	2.6	84
27	Novel USP9X variants in two patients with X-linked intellectual disability. Human Genome Variation, 2019, 6, 49.	0.4	6
28	Neurodevelopmental and Psychiatric Symptoms in Patients with a Cyst Compressing the Cerebellum: an Ongoing Enigma. Cerebellum, 2020, 19, 16-29.	1.4	15
29	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.	0.7	42
30	Usp9X Controls Ankyrin-Repeat Domain Protein Homeostasis during Dendritic Spine Development. Neuron, 2020, 105, 506-521.e7.	3.8	34
31	Targeting E3 Ubiquitin Ligases and Deubiquitinases in Ciliopathy and Cancer. International Journal of Molecular Sciences, 2020, 21, 5962.	1.8	10
32	Experiences with offering pro bono medical genetics services in the West Indies: Benefits to patients, physicians, and the community. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1030-1041.	0.7	8
33	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	1.7	17
34	Understanding the Landscape of X-linked Variants Causing Intellectual Disability in Females Through Extreme X Chromosome Inactivation Skewing. Molecular Neurobiology, 2020, 57, 3671-3684.	1.9	21
35	Skewed X-Chromosome Inactivation and Compensatory Upregulation of Escape Genes Precludes Major Clinical Symptoms in a Female With a Large Xq Deletion. Frontiers in Genetics, 2020, 11, 101.	1.1	19
36	A de novo DDX3X Variant Is Associated With Syndromic Intellectual Disability: Case Report and Literature Review. Frontiers in Pediatrics, 2020, 8, 303.	0.9	6
37	The Ubiquitin System: a Regulatory Hub for Intellectual Disability and Autism Spectrum Disorder. Molecular Neurobiology, 2020, 57, 2179-2193.	1.9	23
38	Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. Journal of Medical Genetics, 2020, 57, 371-379.	1.5	23
39	X-linked diseases: susceptible females. Genetics in Medicine, 2020, 22, 1156-1174.	1.1	110

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40	Abnormal Behavior and Cortical Connectivity Deficits in Mice Lacking <i>Usp9x</i> . <i>Cerebral Cortex</i> , 2021, 31, 1763-1775.	1.6	5
42	Singapore Undiagnosed Disease Program: Genomic Analysis aids Diagnosis and Clinical Management. <i>Archives of Disease in Childhood</i> , 2021, 106, 31-37.	1.0	17
44	The deubiquitylase USP9X controls ribosomal stalling. <i>Journal of Cell Biology</i> , 2021, 220, .	2.3	20
45	Novel USP9X variant associated with syndromic intellectual disability in a female: A case study and review. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1569-1574.	0.7	7
46	The deubiquitinase Usp9x regulates PRC2-mediated chromatin reprogramming during mouse development. <i>Nature Communications</i> , 2021, 12, 1865.	5.8	11
47	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. <i>Journal of Medical Genetics</i> , 2022, 59, 559-567.	1.5	25
48	Clinical and genetic heterogeneity of primary ciliopathies (Review). <i>International Journal of Molecular Medicine</i> , 2021, 48, .	1.8	33
49	Deubiquitylases in developmental ubiquitin signaling and congenital diseases. <i>Cell Death and Differentiation</i> , 2021, 28, 538-556.	5.0	27
51	USP9X counteracts differential ubiquitination of NPHP5 by MARCH7 and BBS11 to regulate ciliogenesis. <i>PLoS Genetics</i> , 2017, 13, e1006791.	1.5	27
52	Heterozygous loss of function of <i>IQSEC2</i> leads to increased activated Arf6 and severe neurocognitive seizure phenotype in females. <i>Life Science Alliance</i> , 2019, 2, e201900386.	1.3	18
56	Clinical features of a male with a <i>USP9X</i> variant associated with intellectual disability: A case study and review of reported cases. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 672-675.	0.7	3
57	Function of Centriolar Satellites and Regulation by Post-Translational Modifications. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 780502.	1.8	5
58	Single-cell RNA sequencing reveals that <i>BMPR2</i> mutation regulates right ventricular function via <i>ID</i> genes. <i>European Respiratory Journal</i> , 2022, 60, 2100327.	3.1	5
59	Compound genetic etiology in a patient with a syndrome including diabetes, intellectual deficiency and distichiasis. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 86.	1.2	0
60	Reciprocal Xp11.4p11.3 microdeletion/microduplication spanning <i>USP9X</i> , <i>DDX3X</i> , and <i>CASK</i> genes in two patients with syndromic intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1836-1847.	0.7	4
61	Pigmentary Mosaicism. <i>Clinics in Dermatology</i> , 2022, , .	0.8	3
62	Selective Xi reactivation and alternative methods to restore MECP2 function in Rett syndrome. <i>Trends in Genetics</i> , 2022, 38, 920-943.	2.9	13
63	Exome and RNA-seq analyses of an incomplete penetrance variant in <i>USP9X</i> in female-specific syndromic intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	1

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64	Regulation of Cell Delamination During Cortical Neurodevelopment and Implication for Brain Disorders. <i>Frontiers in Neuroscience</i> , 2022, 16, 824802.	1.4	3
65	The DUB Club: Deubiquitinating Enzymes and Neurodevelopmental Disorders. <i>Biological Psychiatry</i> , 2022, 92, 614-625.	0.7	8
78	Monogenic causes of pigmentary mosaicism. <i>Human Genetics</i> , 2022, , .	1.8	2
79	Roles and mechanisms of ankyrin-G in neuropsychiatric disorders. <i>Experimental and Molecular Medicine</i> , 2022, 54, 867-877.	3.2	11
80	SAGA-Dependent Histone H2Bub1 Deubiquitination Is Essential for Cellular Ubiquitin Balance during Embryonic Development. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7459.	1.8	5
81	Comparison of chromatin accessibility landscapes during early development of prefrontal cortex between rhesus macaque and human. <i>Nature Communications</i> , 2022, 13, .	5.8	7
82	Identifying phenotypic expansions for congenital diaphragmatic hernia plus (<scp>CDH</scp>+) using <scp>DECIPHER</scp> data. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2958-2968.	0.7	2
83	Exome sequencing revealed USP9X and COL2A1 mutations in a large family with multiple epiphyseal dysplasia. <i>Bone</i> , 2022, 163, 116508.	1.4	2
84	Epigenetics of X-chromosome Inactivation. , 2023, , 419-441.		2
85	A patient with mosaic USP9X gene variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104638.	0.7	0
86	Novel Variant in the USP9X Gene Is Associated with Congenital Heart Disease in a Male Patient: A Case Report and Literature Review. <i>Molecular Syndromology</i> , 2023, 14, 158-163.	0.3	1
87	Molecular diagnosis and novel genes and phenotypes in a pediatric thoracic insufficiency cohort. <i>Scientific Reports</i> , 2023, 13, .	1.6	0
88	A targeted multi-proteomics approach generates a blueprint of the ciliary ubiquitinome. <i>Frontiers in Cell and Developmental Biology</i> , 0, 11, .	1.8	11
89	Dental anomalies in syndromes displaying hypertrichosis in the clinical spectrum. <i>Brazilian Oral Research</i> , 0, 37, .	0.6	0
93	Redefining cerebral palsies as a diverse group of neurodevelopmental disorders with genetic aetiology. <i>Nature Reviews Neurology</i> , 2023, 19, 542-555.	4.9	2