Karen Usdin

List of Publications by Year
in descending order

80	3,412	109321 35	155660 55
papers	citations	h-index	g-index

80	80	80	2842
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The biological effects of simple tandem repeats: Lessons from the repeat expansion diseases: Table 1.. Genome Research, 2008, 18, 1011-1019.	5.5	217
2	Repeat-induced epigenetic changes in intron 1 of the frataxin gene and its consequences in Friedreich ataxia. Nucleic Acids Research, 2007, 35, 3383-3390.	14.5	185
3	Regional FMRP deficits and large repeat expansions into the full mutation range in a new Fragile X premutation mouse model. Gene, 2007, 395, 125-134.	2.2	182
4	Repeat instability during DNA repair: Insights from model systems. Critical Reviews in Biochemistry and Molecular Biology, 2015, 50, 142-167.	5.2	158
5	The fragile X syndrome repeats form RNA hairpins that do not activate the interferon-inducible protein kinase, PKR, but are cut by Dicer. Nucleic Acids Research, 2003, 31, 6243-6248.	14.5	115
6	The Development and Use of a DNA Polymerase Arrest Assay for the Evaluation of Parameters Affecting Intrastrand Tetraplex Formation. Journal of Biological Chemistry, 1996, 271, 20958-20964.	3.4	102
7	Repeat expansions confer WRN dependence in microsatellite-unstable cancers. Nature, 2020, 586, 292-298.	27.8	95
8	Interaction of the Transcription Factors USF1, USF2, and Î±-Pal/Nrf-1 with the FMR1 Promoter. Journal of Biological Chemistry, 2001, 276, 4357-4364.	3.4	84
9	Repeat Expansion Affects Both Transcription Initiation and Elongation in Friedreich Ataxia Cells. Journal of Biological Chemistry, 2011, 286, 4209-4215.	3.4	79
10	The distribution of repressive histone modifications on silenced FMR1 alleles provides clues to the mechanism of gene silencing in fragile X syndrome. Human Molecular Genetics, 2010, 19, 4634-4642.	2.9	76
11	Somatic Expansion in Mouse and Human Carriers of Fragile X Premutation Alleles. Human Mutation, 2013, 34, 157-166.	2.5	76
12	SIRT1 Inhibition Alleviates Gene Silencing in Fragile X Mental Retardation Syndrome. PLoS Genetics, 2008, 4, e1000017.	3.5	75
13	The Mismatch Repair Protein MSH2 is Rate Limiting for Repeat Expansion in a Fragile X Premutation Mouse Model. Human Mutation, 2014, 35, 129-136.	2.5	74
14	Ovarian Abnormalities in a Mouse Model of Fragile X Primary Ovarian Insufficiency. Journal of Histochemistry and Cytochemistry, 2012, 60, 439-456.	2.5	70
15	High-Throughput Screening to Identify Compounds That Increase Fragile X Mental Retardation Protein Expression in Neural Stem Cells Differentiated From Fragile X Syndrome Patient-Derived Induced Pluripotent Stem Cells. Stem Cells Translational Medicine, 2015, 4, 800-808.	3.3	70
16	DNA Secondary Structures and the Evolution of Hypervariable Tandem Arrays. Journal of Biological Chemistry, 1997, 272, 9517-9523.	3.4	66
17	Long Uninterrupted CGG Repeats within the First Exon of the Human FMR1 Gene Are Not Intrinsically Unstable in Transgenic Mice. Genomics, 1998, 50, 229-240.	2.9	58
18	Granulosa cell and oocyte mitochondrial abnormalities in a mouse model of fragile X primary ovarian insufficiency. Molecular Human Reproduction, 2016, 22, 384-396.	2.8	58

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19	Mouse models of the fragile X premutation and fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 25.	3.1	57
20	Use of model systems to understand the etiology of fragile X-associated primary ovarian insufficiency (FXPOI). Journal of Neurodevelopmental Disorders, 2014, 6, 26.	3.1	55
21	The Repeat Expansion Diseases: The dark side of DNA repair. DNA Repair, 2015, 32, 96-105.	2.8	55
22	Potassium bromate, a potent DNA oxidizing agent, exacerbates germline repeat expansion in a fragile X premutation mouse model. Human Mutation, 2010, 31, n/a-n/a.	2.5	52
23	Identification of Fragile X Syndrome Specific Molecular Markers in Human Fibroblasts: A Useful Model to Test the Efficacy of Therapeutic Drugs. Human Mutation, 2014, 35, 1485-1494.	2.5	52
24	MutsÎ² generates both expansions and contractions in a mouse model of the Fragile X-associated disorders. Human Molecular Genetics, 2015, 24, ddv408.	2.9	52
25	ATR protects the genome against CGG{middle dot}CCG-repeat expansion in Fragile X premutation mice. Nucleic Acids Research, 2007, 36, 1050-1056.	14.5	50
26	FAN1 protects against repeat expansions in a Fragile X mouse model. DNA Repair, 2018, 69, 1-5.	2.8	50
27	Long CGG-repeat tracts are toxic to human cells: Implications for carriers of Fragile X premutation alleles. FEBS Letters, 2005, 579, 2702-2708.	2.8	49
28	Impaired activity-dependent FMRP translation and enhanced mGluR-dependent LTD in Fragile X premutation mice. Human Molecular Genetics, 2013, 22, 1180-1192.	2.9	48
29	Chromatin Remodeling in the Noncoding Repeat Expansion Diseases. Journal of Biological Chemistry, 2009, 284, 7413-7417.	3.4	47
30	CGG-repeat dynamics and FMR1 gene silencing in fragile X syndrome stem cells and stem cell-derived neurons. Molecular Autism, 2016, 7, 42.	4.9	47
31	Chromosome fragility and the abnormal replication of the FMR1 locus in fragile X syndrome. Human Molecular Genetics, 2014, 23, 2940-2952.	2.9	45
32	A MutSÎ²-Dependent Contribution of MutSÎ± to Repeat Expansions in Fragile X Premutation Mice?. PLoS Genetics, 2016, 12, e1006190.	3.5	44
33	MutLÎ³ promotes repeat expansion in a Fragile X mouse model while EXO1 is protective. PLoS Genetics, 2018, 14, e1007719.	3.5	43
34	Heterozygosity for a Hypomorphic PolÎ² Mutation Reduces the Expansion Frequency in a Mouse Model of the Fragile X-Related Disorders. PLoS Genetics, 2015, 11, e1005181.	3.5	41
35	Sustained expression ofFMR1mRNA from reactivated fragile X syndrome alleles after treatment with small molecules that prevent trimethylation of H3K27. Human Molecular Genetics, 2016, 25, 3689-3698.	2.9	38
36	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 23.	3.1	36

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37	A Set of Assays for the Comprehensive Analysis of FMR1 Alleles in the Fragile Xâ€“Related Disorders. Journal of Molecular Diagnostics, 2016, 18, 762-774.	2.8	35
38	The roles of Sp1, Sp3, USF1/USF2 and NRF-1 in the regulation and three-dimensional structure of the Fragile X mental retardation gene promoter. Biochemical Journal, 2005, 386, 297-303.	3.7	33
39	Repeat-mediated genetic and epigenetic changes at the FMR1 locus in the Fragile X-related disorders. Frontiers in Genetics, 2014, 5, 226.	2.3	33
40	All three mammalian MutL complexes are required for repeat expansion in a mouse cell model of the Fragile X-related disorders. PLoS Genetics, 2020, 16, e1008902.	3.5	33
41	ATM and ATR protect the genome against two different types of tandem repeat instability in Fragile X premutation mice. Nucleic Acids Research, 2009, 37, 6371-6377.	14.5	32
42	Polycomb group complexes are recruited to reactivated FMR1 alleles in Fragile X syndrome in response to FMR1 transcription. Human Molecular Genetics, 2014, 23, 6575-6583.	2.9	31
43	Gender and Cell-Type-Specific Effects of the Transcription-Coupled Repair Protein, ERCC6/CSB, on Repeat Expansion in a Mouse Model of the Fragile X-Related Disorders. Human Mutation, 2014, 35, 341-349.	2.5	30
44	Tetraplex formation by the progressive myoclonus epilepsy type-1 repeat: implications for instability in the repeat expansion diseases. FEBS Letters, 2001, 491, 184-187.	2.8	29
45	Is Friedreich ataxia an epigenetic disorder?. Clinical Epigenetics, 2012, 4, 2.	4.1	29
46	X inactivation plays a major role in the gender bias in somatic expansion in a mouse model of the fragile X-related disorders: implications for the mechanism of repeat expansion. Human Molecular Genetics, 2014, 23, 4985-4994.	2.9	29
47	Repeat-mediated epigenetic dysregulation of the FMR1 gene in the fragile X-related disorders. Frontiers in Genetics, 2015, 6, 192.	2.3	29
48	Timing of Expansion of Fragile X Premutation Alleles During Intergenerational Transmission in a Mouse Model of the Fragile X-Related Disorders. Frontiers in Genetics, 2018, 9, 314.	2.3	24
49	The Mouse Ms6-hm Hypervariable Microsatellite Forms a Hairpin and Two Unusual Tetraplexes. Journal of Biological Chemistry, 1998, 273, 30742-30749.	3.4	23
50	The role of DNA damage response pathways in chromosome fragility in Fragile X syndrome. Nucleic Acids Research, 2009, 37, 4385-4392.	14.5	21
51	Ancient repeated DNA elements and the regulation of the human frataxin promoter. Genomics, 2005, 85, 221-230.	2.9	20
52	Fragile X syndrome and Friedreichâ€™s ataxia: two different paradigms for repeat induced transcript insufficiency. Brain Research Bulletin, 2001, 56, 367-373.	3.0	19
53	The Transcription-Coupled Repair Protein ERCC6/CSB Also Protects Against Repeat Expansion in a Mouse Model of the Fragile X Premutation. Human Mutation, 2015, 36, 482-487.	2.5	19
54	Repeat Instability in the Fragile X-Related Disorders: Lessons from a Mouse Model. Brain Sciences, 2019, 9, 52.	2.3	19

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55	A point mutation in the nuclease domain of MLH3 eliminates repeat expansions in a mouse stem cell model of the Fragile X-related disorders. Nucleic Acids Research, 2020, 48, 7856-7863.	14.5	19
56	Double-strand break repair plays a role in repeat instability in a fragile X mouse model. DNA Repair, 2019, 74, 63-69.	2.8	18
57	Chromatin changes in the development and pathology of the Fragile X-associated disorders and Friedreich ataxia. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2012, 1819, 802-810.	1.9	17
58	Ups and Downs: Mechanisms of Repeat Instability in the Fragile X-Related Disorders. Genes, 2016, 7, 70.	2.4	16
59	Recent advances in assays for the fragile X-related disorders. Human Genetics, 2017, 136, 1313-1327.	3.8	16
60	Evidence for chromosome fragility at the frataxin locus in Friedreich ataxia. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 781, 14-21.	1.0	15
61	Modifiers of Somatic Repeat Instability in Mouse Models of Friedreich Ataxia and the Fragile X-Related Disorders: Implications for the Mechanism of Somatic Expansion in Huntingtonâ€™s Disease. Journal of Huntington's Disease, 2021, 10, 149-163.	1.9	15
62	Small Molecules Targeting H3K9 Methylation Prevent Silencing of Reactivated FMR1 Alleles in Fragile X Syndrome Patient Derived Cells. Genes, 2020, 11, 356.	2.4	12
63	(Dys)function Follows Form: Nucleic Acid Structure, Repeat Expansion, and Disease Pathology in FMR1 Disorders. International Journal of Molecular Sciences, 2021, 22, 9167.	4.1	11
64	Both cis and trans-acting genetic factors drive somatic instability in female carriers of the FMR1 premutation. Scientific Reports, 2022, 12, .	3.3	11
65	Improved Assays for AGG Interruptions in Fragile X Premutation Carriers. Journal of Molecular Diagnostics, 2017, 19, 828-835.	2.8	10
66	Pharmacological Reactivation of the Silenced FMR1 Gene as a Targeted Therapeutic Approach for Fragile X Syndrome. Brain Sciences, 2019, 9, 39.	2.3	10
67	Common Threads: Aphidicolin-Inducible and Folate-Sensitive Fragile Sites in the Human Genome. Frontiers in Genetics, 2021, 12, 708860.	2.3	9
68	FAN1â€™s protection against CGG repeat expansion requires its nuclease activity and is FANCD2-independent. Nucleic Acids Research, 2021, 49, 11643-11652.	14.5	9
69	Molecular analysis of FMR1 alleles for fragile X syndrome diagnosis and patient stratification. Expert Review of Molecular Diagnostics, 2020, 20, 363-365.	3.1	8
70	Fragile X syndrome in a male with methylated premutation alleles and no detectable methylated full mutation alleles. American Journal of Medical Genetics, Part A, 2019, 179, 2132-2137.	1.2	7
71	Mechanisms of Genome Instability in the Fragile X-Related Disorders. Genes, 2021, 12, 1633.	2.4	6
72	Isolation and Analysis of the CGG-Repeat Size in Male and Female Gametes from a Fragile X Mouse Model. Methods in Molecular Biology, 2020, 2056, 173-186.	0.9	5

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73	CGG Repeat Expansion, and Elevated Fmr1 Transcription and Mitochondrial Copy Number in a New Fragile X PM Mouse Embryonic Stem Cell Model. Frontiers in Cell and Developmental Biology, 2020, 8, 482.	3.7	4
74	Assays for Determining Repeat Number, Methylation Status, and AGG Interruptions in the Fragile X-Related Disorders. Methods in Molecular Biology, 2019, 1942, 49-59.	0.9	2
75	Bending the Rules: Unusual Nucleic Acid Structures and Disease Pathology in the Repeat Expansion Diseases. , 2006, , 617-635.		2
76	Stool is a sensitive and noninvasive source of DNA for monitoring expansion in repeat expansion disease mouse models. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	1
77	Model Systems for Understanding FXPOI. , 2016, , 225-240.		0
78	Mechanisms of Repeat Instability in Fragile X Syndrome. , 2017, , 77-102.		0
79	Epigenetic dysregulation in the fragile X-related disorders. , 2019, , 261-283.		0
80	Editorial: Proceedings of the â€œFourth International Conference of the FMR1 Premutation: Basic Mechanisms, Clinical Involvement and Therapyâ€• Frontiers in Molecular Biosciences, 2021, 8, 671875.	3.5	0