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	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
2	Both cis and trans-acting genetic factors drive somatic instability in female carriers of the FMR1 premutation. Scientific Reports, 2022, 12, .	3.3	11
3	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
4	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
5	(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
6	Common Threads: Aphidicolin-Inducible and Folate-Sensitive Fragile Sites in the Human Genome. Frontiers in Genetics, 2021, 12, 708860.	2.3	9
7	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
8	Mechanisms of Genome Instability in the Fragile X-Related Disorders. Genes, 2021, 12, 1633.	2.4	6
9	Repeat expansions confer WRN dependence in microsatellite-unstable cancers. Nature, 2020, 586, 292-298.	27.8	95
10	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
11	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
12	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
13	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
14	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
15	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
16	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
17	Epigenetic dysregulation in the fragile X-related disorders. , 2019, , 261-283.		0
18	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

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19	Repeat Instability in the Fragile X-Related Disorders: Lessons from a Mouse Model. Brain Sciences, 2019, 9, 52.	2.3	19
20	Pharmacological Reactivation of the Silenced FMR1 Gene as a Targeted Therapeutic Approach for Fragile X Syndrome. Brain Sciences, 2019, 9, 39.	2.3	10
21	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
22	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
23	MutLÎ³ promotes repeat expansion in a Fragile X mouse model while EXO1 is protective. PLoS Genetics, 2018, 14, e1007719.	3.5	43
24	FAN1 protects against repeat expansions in a Fragile X mouse model. DNA Repair, 2018, 69, 1-5.	2.8	50
25	Improved Assays for AGG Interruptions in Fragile X Premutation Carriers. Journal of Molecular Diagnostics, 2017, 19, 828-835.	2.8	10
26	Recent advances in assays for the fragile X-related disorders. Human Genetics, 2017, 136, 1313-1327.	3.8	16
27	Mechanisms of Repeat Instability in Fragile X Syndrome. , 2017, , 77-102.		0
28	Ups and Downs: Mechanisms of Repeat Instability in the Fragile X-Related Disorders. Genes, 2016, 7, 70.	2.4	16
29	A MutSÎ²-Dependent Contribution of MutSÎ± to Repeat Expansions in Fragile X Premutation Mice?. PLoS Genetics, 2016, 12, e1006190.	3.5	44
30	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
31	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
32	Model Systems for Understanding FXPOI. , 2016, , 225-240.		0
33	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
34	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
35	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
36	Repeat-mediated epigenetic dysregulation of the FMR1 gene in the fragile X-related disorders. Frontiers in Genetics, 2015, 6, 192.	2.3	29

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37	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
38	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
39	Repeat instability during DNA repair: Insights from model systems. Critical Reviews in Biochemistry and Molecular Biology, 2015, 50, 142-167.	5.2	158
40	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
41	The Repeat Expansion Diseases: The dark side of DNA repair. DNA Repair, 2015, 32, 96-105.	2.8	55
42	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
43	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
44	Chromosome fragility and the abnormal replication of the FMR1 locus in fragile X syndrome. Human Molecular Genetics, 2014, 23, 2940-2952.	2.9	45
45	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
46	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
47	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
48	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
49	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
50	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 23.	3.1	36
51	Mouse models of the fragile X premutation and fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 25.	3.1	57
52	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
53	Somatic Expansion in Mouse and Human Carriers of Fragile X Premutation Alleles. Human Mutation, 2013, 34, 157-166.	2.5	76
54	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

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55	Ovarian Abnormalities in a Mouse Model of Fragile X Primary Ovarian Insufficiency. Journal of Histochemistry and Cytochemistry, 2012, 60, 439-456.	2.5	70
56	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
57	Is Friedreich ataxia an epigenetic disorder?. Clinical Epigenetics, 2012, 4, 2.	4.1	29
58	Repeat Expansion Affects Both Transcription Initiation and Elongation in Friedreich Ataxia Cells. Journal of Biological Chemistry, 2011, 286, 4209-4215.	3.4	79
59	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
60	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
61	Chromatin Remodeling in the Noncoding Repeat Expansion Diseases. Journal of Biological Chemistry, 2009, 284, 7413-7417.	3.4	47
62	The role of DNA damage response pathways in chromosome fragility in Fragile X syndrome. Nucleic Acids Research, 2009, 37, 4385-4392.	14.5	21
63	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
64	The biological effects of simple tandem repeats: Lessons from the repeat expansion diseases: Table 1.. Genome Research, 2008, 18, 1011-1019.	5.5	217
65	SIRT1 Inhibition Alleviates Gene Silencing in Fragile X Mental Retardation Syndrome. PLoS Genetics, 2008, 4, e1000017.	3.5	75
66	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
67	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
68	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
69	Bending the Rules: Unusual Nucleic Acid Structures and Disease Pathology in the Repeat Expansion Diseases. , 2006, , 617-635.		2
70	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
71	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
72	Ancient repeated DNA elements and the regulation of the human frataxin promoter. Genomics, 2005, 85, 221-230.	2.9	20

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73	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
74	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
75	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
76	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
77	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
78	The Mouse Ms6-hm Hypervariable Microsatellite Forms a Hairpin and Two Unusual Tetraplexes. Journal of Biological Chemistry, 1998, 273, 30742-30749.	3.4	23
79	DNA Secondary Structures and the Evolution of Hypervariable Tandem Arrays. Journal of Biological Chemistry, 1997, 272, 9517-9523.	3.4	66
80	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