Sarah L Nolin

List of Publications by Year
in descending order

41 papers	2,048 citations	430874 18 h-index	395702 33 g-index
41 all docs	41 docs citations	41 times ranked	1250 citing authors

#	Article	IF	Citations
1	Fragile X premutation is a significant risk factor for premature ovarian failure: The international collaborative POF in fragile X study?preliminary data. American Journal of Medical Genetics Part A, 1999, 83, 322-325.	2.4	405
2	Expansion of the Fragile X CGG Repeat in Females with Premutation or Intermediate Alleles. American Journal of Human Genetics, 2003, 72, 454-464.	6.2	345
3	Mosaicism in fragile X affected males. American Journal of Medical Genetics Part A, 1994, 51, 509-512.	2.4	158
4	Fragile X full mutation expansions are inhibited by one or more AGG interruptions in premutation carriers. Genetics in Medicine, 2015, 17, 358-364.	2.4	119
5	Fragile X AGG analysis provides new risk predictions for 45â€“69 repeat alleles. American Journal of Medical Genetics, Part A, 2013, 161, 771-778.	1.2	110
6	Fragile X analysis of 1112 prenatal samples from 1991 to 2010. Prenatal Diagnosis, 2011, 31, 925-931.	2.3	86
7	The Fragile X Mental Retardation Protein FMRP Binds Elongation Factor 1A mRNA and Negatively Regulates Its Translation in Vivo. Journal of Biological Chemistry, 2003, 278, 15669-15678.	3.4	75
8	Mosaicism for theFMR1 gene influences adaptive skills development in fragile X-affected males. American Journal of Medical Genetics Part A, 1996, 64, 365-369.	2.4	62
9	Prenatal diagnosis and carrier screening for fragile X by PCR. , 1996, 64, 191-195.		57
10	Examination of Factors Associated with Instability of the FMR1 CGG Repeat. American Journal of Human Genetics, 1998, 63, 776-785.	6.2	48
11	Cis-acting DNA sequence at a replication origin promotes repeat expansion to fragile X full mutation. Journal of Cell Biology, 2014, 206, 599-607.	5.2	48
12	Tissue differences in fragile X mosaics: Mosaicism in blood cells may differ greatly from skin. , 1996, 64, 296-301.		47
13	Fragile X Screening by Quantification of FMRP in Dried Blood Spots by a Luminex Immunoassay. Journal of Molecular Diagnostics, 2013, 15, 508-517.	2.8	47
14	The role of AGG interruptions in fragile X repeat expansions: a twenty-year perspective. Frontiers in Genetics, 2014, 5, 244.	2.3	47
15	Survey of the Fragile X Syndrome CGG Repeat and the Short-Tandem-Repeat and Single-Nucleotide-Polymorphism Haplotypes in an African American Population. American Journal of Human Genetics, 2000, 66, 480-493.	6.2	45
16	Expansions and contractions of the FMR1 CGG repeat in 5,508 transmissions of normal, intermediate, and premutation alleles. American Journal of Medical Genetics, Part A, 2019, 179, 1148-1156.	1.2	42
17	FMR1 CGG-Repeat Instability in Single Sperm and Lymphocytes of Fragile-X Premutation Males. American Journal of Human Genetics, 1999, 65, 680-688.	6.2	34
18	Reverse mutations in the fragile X syndrome. , 1996, 64, 287-292.		33

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19	Fragile X screening program in New York State. American Journal of Medical Genetics Part A, 1991, 38, 251-255.	2.4	26
20	Fragile X prenatal analyses show full mutation females at high risk for mosaic Turner syndrome: Fragile X leads to chromosome loss. American Journal of Medical Genetics, Part A, 2009, 149A, 2152-2157.	1.2	25
21	Fragile X full mutation alleles composed of few alleles: Implications for CGG repeat expansion. American Journal of Medical Genetics, Part A, 2008, 146A, 60-65.	1.2	18
22	Fragile X protein in newborn dried blood spots. BMC Medical Genetics, 2014, 15, 119.	2.1	18
23	Large-scale copy number variants (CNVs): Distribution in normal subjects and FISH/real-time qPCR analysis. BMC Genomics, 2007, 8, 167.	2.8	17
24	Terminal deletions of the long arm of chromosome X that include the FMR1 gene in female patients: A case series. , 2011, 155, 870-874.		16
25	Absence of AGG Interruptions Is a Risk Factor for Full Mutation Expansion Among Israeli FMR1 Premutation Carriers. Frontiers in Genetics, 2018, 9, 606.	2.3	14
26	Distal duplication 14q: Report of three cases and further delineation of the syndrome. Human Genetics, 1984, 68, 159-164.	3.8	13
27	New York State screening program for fragile X syndrome: A progress report. American Journal of Medical Genetics Part A, 1992, 43, 328-332.	2.4	11
28	Molecular Analysis of Fragile X Syndrome. Current Protocols in Human Genetics, 2003, 38, Unit9.5.	3.5	11
29	Prenatal fragile X detection using cytoplasmic and nuclear-specific monoclonal antibodies. , 1999, 83, 342-346.		10
30	FXPOI: Pattern of AGG Interruptions Does not Show an Association With Age at Amenorrhea Among Women With a Premutation. Frontiers in Genetics, 2018, 9, 292.	2.3	10
31	Deficits in Prenatal Serine Biosynthesis Underlie the Mitochondrial Dysfunction Associated with the Autism-Linked FMR1 Gene. International Journal of Molecular Sciences, 2021, 22, 5886.	4.1	10
32	Isolation and regional localization by insitu hybridization of a unique gene segment to chromosome 21. Biochemical and Biophysical Research Communications, 1984, 121, 380-385.	2.1	8
33	Accelerated prenatal diagnosis of fragile X syndrome by polymerase chain reaction restriction fragment detection. , 1999, 83, 338-341.		8
34	Localization of Chromosome 21 Probes by in Situ Hybridization. Annals of the New York Academy of Sciences, 1985, 450, 69-83.	3.8	7
35	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
36	Global transcriptome dysregulation in second trimester fetuses with FMR1 expansions. Prenatal Diagnosis, 2017, 37, 43-52.	2.3	5

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37	Fragile X premutation is a significant risk factor for premature ovarian failure: The international collaborative POF in fragile X studyâ€"preliminary data. American Journal of Medical Genetics Part A, 1999, 83, 322-325.	2.4	3
38	In situ nick translation of the fragile X region. American Journal of Medical Genetics Part A, 1988, 30, 443-450.	2.4	2
39	Invited Commentary: Apparent FMR1 Allele Instability in Non-Fragile X Males. Genetic Testing and Molecular Biomarkers, 2000, 4, 241-242.	1.7	1
40	Cloning the ends of size selected Sfi I fragments. American Journal of Medical Genetics Part A, 1991, 38, 384-390.	2.4	0
41	Molecular carrier testing for the fragile X syndrome: Issues for genetic counselors. Journal of Genetic Counseling, 1994, 3, 233-244.	1.6	0