Stephanie L Sherman

List of Publications by Year
in descending order

105 papers	6,791 citations	94433 37 h-index	64796 79 g-index
111 all docs	111 docs citations	111 times ranked	5024 citing authors

#	Article	IF	Citations
1	Stephen T. Warren, Ph.D. (1953â€“2021): A remembrance. American Journal of Human Genetics, 2022, 109, 3-11.	6.2	2
2	COVID-19 Vaccination of Individuals with Down Syndromeâ€”Data from the Trisomy 21 Research Society Survey on Safety, Efficacy, and Factors Associated with the Decision to Be Vaccinated. Vaccines, 2022, 10, 530.	4.4	8
3	Identification of PSMB5 as a genetic modifier of fragile Xâ€“associated tremor/ataxia syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	7
4	Men with an FMR1 premutation and their health education needs. Journal of Genetic Counseling, 2021, 30, 1156-1167.	1.6	0
5	Medical vulnerability of individuals with Down syndrome to severe COVID-19â€“data from the Trisomy 21 Research Society and the UK ISARIC4C survey. EClinicalMedicine, 2021, 33, 100769.	7.1	73
6	Analyses stratified by maternal age and recombination further characterize genes associated with maternal nondisjunction of chromosome 21. Prenatal Diagnosis, 2021, 41, 591-609.	2.3	4
7	Opportunities, barriers, and recommendations in Down syndrome research. Translational Science of Rare Diseases, 2021, 5, 99-129.	1.5	33
8	Refining the risk for fragile Xâ€“associated primary ovarian insufficiency (FXPOI) by FMR1 CGG repeat size. Genetics in Medicine, 2021, 23, 1648-1655.	2.4	20
9	Spoken language outcome measures for treatment studies in Down syndrome: feasibility, practice effects, test-retest reliability, and construct validity of variables generated from expressive language sampling. Journal of Neurodevelopmental Disorders, 2021, 13, 13.	3.1	18
10	Relationship between Apgar scores and long-term cognitive outcomes in individuals with Down syndrome. Scientific Reports, 2021, 11, 12707.	3.3	3
11	Comparison of COVID-19 and Non-COVID-19 Pneumonia in Down Syndrome. Journal of Clinical Medicine, 2021, 10, 3748.	2.4	7
12	Identifying susceptibility genes for primary ovarian insufficiency on the high-risk genetic background of a fragile X premutation. Fertility and Sterility, 2021, 116, 843-854.	1.0	5
13	Symptoms of Autism Spectrum Disorder in Individuals with Down Syndrome. Brain Sciences, 2021, 11, 1278.	2.3	9
14	Predictors of Comorbid Conditions in Women Who Carry an FMR1 Premutation. Frontiers in Psychiatry, 2021, 12, 715922.	2.6	4
15	COVID-19 in Children with Down Syndrome: Data from the Trisomy 21 Research Society Survey. Journal of Clinical Medicine, 2021, 10, 5125.	2.4	24
16	Clustering of comorbid conditions among women who carry an FMR1 premutation. Genetics in Medicine, 2020, 22, 758-766.	2.4	31
17	Analysis of the genomic expression profile in trisomy 18: insight into possible genes involved in the associated phenotypes. Human Molecular Genetics, 2020, 29, 238-247.	2.9	5
18	An empirical bayesian approach for testing gene expression fold change and its application in detecting global dosage effects. NAR Genomics and Bioinformatics, 2020, 2, lqaa072.	3.2	0

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19	Gestational age is related to symptoms of attention-deficit/hyperactivity disorder in late-preterm to full-term children and adolescents with down syndrome. Scientific Reports, 2020, 10, 20345.	3.3	11
20	Identifying genetic factors that contribute to the increased risk of congenital heart defects in infants with Down syndrome. Scientific Reports, 2020, 10, 18051.	3.3	14
21	Study of telomere length in men who carry a fragile X premutation or full mutation allele. Human Genetics, 2020, 139, 1531-1539.	3.8	1
22	Expressive language sampling as a source of outcome measures for treatment studies in fragile X syndrome: feasibility, practice effects, test-retest reliability, and construct validity. Journal of Neurodevelopmental Disorders, 2020, 12, 10.	3.1	32
23	Down syndrome. Nature Reviews Disease Primers, 2020, 6, 9.	30.5	376
24	Health knowledge of women with a fragile X premutation: Improving understanding with targeted educational material. Journal of Genetic Counseling, 2020, 29, 983-991.	1.6	7
25	Inherited genetic susceptibility to acute lymphoblastic leukemia in Down syndrome. Blood, 2019, 134, 1227-1237.	1.4	37
26	Characteristics Associated with Autism Spectrum Disorder Risk in Individuals with Down Syndrome. Journal of Autism and Developmental Disorders, 2019, 49, 3543-3556.	2.7	27
27	Preventive care services and health behaviors in children with fragile X syndrome. Disability and Health Journal, 2019, 12, 564-573.	2.8	4
28	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
29	Feasibility of an app-based mindfulness intervention among women with an FMR1 premutation experiencing maternal stress. Research in Developmental Disabilities, 2019, 89, 76-82.	2.2	7
30	A candidate gene analysis and GWAS for genes associated with maternal nondisjunction of chromosome 21. PLoS Genetics, 2019, 15, e1008414.	3.5	25
31	P2â€030: THE LIFEâ€ÐSR STUDY. Alzheimer's and Dementia, 2019, 15, .	0.8	0
32	Analysis of Copy Number Variants on Chromosome 21 in Down Syndrome-Associated Congenital Heart Defects. G3: Genes, Genomes, Genetics, 2018, 8, 105-111.	1.8	13
33	Associations Between Medical History, Cognition, and Behavior in Youth With Down Syndrome: A Report From the Down Syndrome Cognition Project. American Journal on Intellectual and Developmental Disabilities, 2018, 123, 514-528.	1.6	25
34	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
35	Newton E. Morton (1929â€“2018). American Journal of Human Genetics, 2018, 102, 1011-1017.	6.2	0
36	The Arizona Cognitive Test Battery for Down Syndrome: Test-Retest Reliability and Practice Effects. American Journal on Intellectual and Developmental Disabilities, 2017, 122, 215-234.	1.6	35

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37	Parental Perspectives on Pharmacological Clinical Trials: a Qualitative Study in Down Syndrome and Fragile X Syndrome. Journal of Genetic Counseling, 2017, 26, 1333-1340.	1.6	11
38	Autism Spectrum Disorder in Fragile X Syndrome: Cooccurring Conditions and Current Treatment. Pediatrics, 2017, 139, S194-S206.	2.1	186
39	FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. Pediatrics, 2017, 139, S183-S193.	2.1	39
40	Women who carry a fragile X premutation are biologically older than noncarriers as measured by telomere length. American Journal of Medical Genetics, Part A, 2017, 173, 2985-2994.	1.2	6
41	Low-level maternal exposure to nicotine associates with significant metabolic perturbations in second-trimester amniotic fluid. Environment International, 2017, 107, 227-234.	10.0	15
42	Variation in the Zinc Finger of PRDM9 is Associated with the Absence of Recombination along Nondisjoined Chromosomes 21 of Maternal Origin. Journal of Down Syndrome & Chromosome Abnormalities, 2016, 02, .	0.1	7
43	Regionally Smoothed Meta-Analysis Methods for GWAS Datasets. Genetic Epidemiology, 2016, 40, 154-160.	1.3	6
44	Genome-Wide Association Study of Meiotic Recombination Phenotypes. G3: Genes, Genomes, Genetics, 2016, 6, 3995-4007.	1.8	9
45	Influence of CHDs on psycho-social and neurodevelopmental outcomes in children with Down syndrome. Cardiology in the Young, 2016, 26, 250-256.	0.8	25
46	Cliniciansâ€™ experiences with the fragile X clinical and research consortium. American Journal of Medical Genetics, Part A, 2016, 170, 3138-3143.	1.2	6
47	Importance of a specialty clinic for individuals with fragile X syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3144-3149.	1.2	7
48	Reproductive and gynecologic care of women with fragile X primary ovarian insufficiency (FXPOI). Menopause, 2016, 23, 993-999.	2.0	45
49	Improving Health Education for Women Who Carry an FMR1 Premutation. Journal of Genetic Counseling, 2016, 25, 228-238.	1.6	6
50	Genotype/Phenotype Relationships in FXTAS. , 2016, , 129-160.		1
51	Genome-Wide Association Study of Down Syndrome-Associated Atrioventricular Septal Defects. G3: Genes, Genomes, Genetics, 2015, 5, 1961-1971.	1.8	28
52	Contribution of copy-number variation to Down syndromeâ€“associated atrioventricular septal defects. Genetics in Medicine, 2015, 17, 554-560.	2.4	24
53	Cognitive aspects of Fragile X syndrome. Wiley Interdisciplinary Reviews: Cognitive Science, 2014, 5, 501-508.	2.8	32
54	Evidence for dysregulation of genome-wide recombination in oocytes with nondisjoined chromosomes 21. Human Molecular Genetics, 2014, 23, 408-417.	2.9	17

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55	Approaches to identify genetic variants that influence the risk for onset of fragile X-associated primary ovarian insufficiency (FXPOI): a preliminary study. Frontiers in Genetics, 2014, 5, 260.	2.3	15
56	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
57	The cognitive neuropsychological phenotype of carriers of the FMR1 premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 28.	3.1	74
58	Associated features in females with an FMR1 premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 30.	3.1	116
59	An Examination of the Relationship between Hotspots and Recombination Associated with Chromosome 21 Nondisjunction. PLoS ONE, 2014, 9, e99560.	2.5	15
60	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
61	Maternal Age and Oocyte Aneuploidy: Lessons Learned from Trisomy 21. , 2013, , 69-85.		3
62	Genetic basis for primary ovarian insufficiency. , 2013, , 394-408.		1
63	Olfactory dysfunction in fragile X tremor ataxia syndrome. Movement Disorders, 2012, 27, 1556-1559.	3.9	16
64	Depression and anxiety symptoms among women who carry the FMR1 premutation: Impact of raising a child with fragile X syndrome is moderated by CRHR1 polymorphisms. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 549-559.	1.7	32
65	Altered patterns of multiple recombinant events are associated with nondisjunction of chromosome 21. Human Genetics, 2012, 131, 1039-1046.	3.8	39
66	The FMR1 Premutation and Attention-Deficit Hyperactivity Disorder (ADHD): Evidence for a Complex Inheritance. Behavior Genetics, 2012, 42, 415-422.	2.1	41
67	Neuropsychological findings from older premutation carrier males and their noncarrier siblings from families with fragile X syndrome.. Neuropsychology, 2011, 25, 404-411.	1.3	19
68	New clinical findings in the fragile X-associated tremor ataxia syndrome (FXTAS). Neurogenetics, 2011, 12, 123-135.	1.4	67
69	Fragile X analysis of 1112 prenatal samples from 1991 to 2010. Prenatal Diagnosis, 2011, 31, 925-931.	2.3	86
70	Coâ€occurring diagnoses among FMR1 premutation allele carriers. Clinical Genetics, 2010, 77, 374-381.	2.0	80
71	Genotype/Phenotype Relationships in FXTAS. , 2010, , 95-122.		1
72	Is there evidence for neuropsychological and neurobehavioral phenotypes among adults without FXTAS who carry the FMR1 premutation? A review of current literature. Genetics in Medicine, 2009, 11, 79-89.	2.4	43

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73	Genetic Analysis of Variation in Human Meiotic Recombination. PLoS Genetics, 2009, 5, e1000648.	3.5	142
74	Incidence of Fragile X Syndrome by Newborn Screening for Methylated FMR1 DNA. American Journal of Human Genetics, 2009, 85, 503-514.	6.2	347
75	Investigation of Phenotypes Associated with Mood and Anxiety Among Male and Female Fragile X Premutation Carriers. Behavior Genetics, 2008, 38, 493-502.	2.1	65
76	No Evidence for a Difference in Neuropsychological Profile among Carriers and Noncarriers of the FMR1 Premutation in Adults under the Age of 50. American Journal of Human Genetics, 2008, 83, 692-702.	6.2	71
77	New Insights into Human Nondisjunction of Chromosome 21 in Oocytes. PLoS Genetics, 2008, 4, e1000033.	3.5	146
78	Smarter clustering methods for SNP genotype calling. Bioinformatics, 2008, 24, 2665-2671.	4.1	18
79	Clinical significance of tri-nucleotide repeats in Fragile X testing: A clarification of American College of Medical Genetics guidelines. Genetics in Medicine, 2008, 10, 845-847.	2.4	58
80	The National down Syndrome Project: Design and Implementation. Public Health Reports, 2007, 122, 62-72.	2.5	67
81	The FMR1 premutation and reproduction. Fertility and Sterility, 2007, 87, 456-465.	1.0	360
82	Epidemiology of Down syndrome. Mental Retardation and Developmental Disabilities Research Reviews, 2007, 13, 221-227.	3.6	353
83	Examination of the Effect of the Polymorphic CGG Repeat in the FMR1 Gene on Cognitive Performance. Behavior Genetics, 2005, 35, 435-445.	2.1	49
84	Fragile X syndrome: Diagnostic and carrier testing. Genetics in Medicine, 2005, 7, 584-587.	2.4	247
85	Association between Maternal Age and Meiotic Recombination for Trisomy 21. American Journal of Human Genetics, 2005, 76, 91-99.	6.2	107
86	A study of the distributional characteristics of FMR1 transcript levels in 238 individuals. Human Genetics, 2004, 114, 439-447.	3.8	119
87	Linkage disequilibrium mapping in trisomic populations: Analytical approaches and an application to congenital heart defects in Down syndrome. Genetic Epidemiology, 2004, 27, 240-251.	1.3	18
88	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
89	FMR1 and the fragile X syndrome: Human genome epidemiology review. Genetics in Medicine, 2001, 3, 359-371.	2.4	556
90	Premature ovarian failure in the fragile X syndrome. American Journal of Medical Genetics Part A, 2000, 97, 189-194.	2.4	396

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91	Down syndrome: genetic recombination and the origin of the extra chromosome 21. Clinical Genetics, 2000, 57, 95-100.	2.0	125
92	Premature Ovarian Failure among Fragile X Premutation Carriers: Parent-of-Origin Effect?. American Journal of Human Genetics, 2000, 67, 11-13.	6.2	61
93	The relation of the dopamine transporter gene (DAT1) to symptoms of internalizing disorders in children. Behavior Genetics, 1998, 28, 215-225.	2.1	119
94	Testing for contributions of mitochondrial DNA mutations to complex diseases. , 1998, 15, 451-469.		4
95	FISH studies of the sperm of fathers of paternally derived cases of trisomy 21: no evidence for an increase in aneuploidy. Human Genetics, 1998, 103, 654-657.	3.8	17
96	Sequential sib-pair and association studies to detect genes in quantitative traits. Genetic Epidemiology, 1997, 14, 885-890.	1.3	1
97	The method of sib-pair linkage analysis in context of case-control design. Genetic Epidemiology, 1997, 14, 939-944.	1.3	3
98	Examination of factors that influence the expansion of the fragile X mutation in a sample of conceptuses from known carrier females. , 1996, 64, 256-260.		30
99	Survey of the fragile X syndrome and the fragile X E syndrome in a special education needs population. , 1996, 64, 428-433.		56
100	Susceptible chiasmate configurations of chromosome 21 predispose to nonâ€“disjunction in both maternal meiosis I and meiosis II. Nature Genetics, 1996, 14, 400-405.	21.4	362
101	Modeling the natural history of the fragile X gene. Mental Retardation and Developmental Disabilities Research Reviews, 1995, 1, 263-268.	3.6	3
102	CGG-repeat polymorphism of the BCR gene rules out predisposing alleles leading to the philadelphia chromosome. Genes Chromosomes and Cancer, 1994, 9, 141-144.	2.8	4
103	Concordance and recessive inheritance of Leber congenital amaurosis. American Journal of Medical Genetics Part A, 1993, 46, 275-277.	2.4	16
104	Human genes containing polymorphic trinucleotide repeats. Nature Genetics, 1992, 2, 186-191.	21.4	160
105	Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. American Journal of Medical Genetics Part A, 1992, 43, 237-243.	2.4	82