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	FMR1 and the fragile X syndrome: Human genome epidemiology review. Genetics in Medicine, 2001, 3, 359-371.	2.4	556
2	Premature ovarian failure in the fragile X syndrome. American Journal of Medical Genetics Part A, 2000, 97, 189-194.	2.4	396
3	Down syndrome. Nature Reviews Disease Primers, 2020, 6, 9.	30.5	376
4	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
5	The FMR1 premutation and reproduction. Fertility and Sterility, 2007, 87, 456-465.	1.0	360
6	Epidemiology of Down syndrome. Mental Retardation and Developmental Disabilities Research Reviews, 2007, 13, 221-227.	3.6	353
7	Incidence of Fragile X Syndrome by Newborn Screening for Methylated FMR1 DNA. American Journal of Human Genetics, 2009, 85, 503-514.	6.2	347
8	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
9	Fragile X syndrome: Diagnostic and carrier testing. Genetics in Medicine, 2005, 7, 584-587.	2.4	247
10	Autism Spectrum Disorder in Fragile X Syndrome: Cooccurring Conditions and Current Treatment. Pediatrics, 2017, 139, S194-S206.	2.1	186
11	Human genes containing polymorphic trinucleotide repeats. Nature Genetics, 1992, 2, 186-191.	21.4	160
12	New Insights into Human Nondisjunction of Chromosome 21 in Oocytes. PLoS Genetics, 2008, 4, e1000033.	3.5	146
13	Genetic Analysis of Variation in Human Meiotic Recombination. PLoS Genetics, 2009, 5, e1000648.	3.5	142
14	Down syndrome: genetic recombination and the origin of the extra chromosome 21. Clinical Genetics, 2000, 57, 95-100.	2.0	125
15	The relation of the dopamine transporter gene (DAT1) to symptoms of internalizing disorders in children. Behavior Genetics, 1998, 28, 215-225.	2.1	119
16	A study of the distributional characteristics of FMR1 transcript levels in 238 individuals. Human Genetics, 2004, 114, 439-447.	3.8	119
17	Associated features in females with an FMR1 premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 30.	3.1	116
18	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

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19	Association between Maternal Age and Meiotic Recombination for Trisomy 21. American Journal of Human Genetics, 2005, 76, 91-99.	6.2	107
20	Fragile X analysis of 1112 prenatal samples from 1991 to 2010. Prenatal Diagnosis, 2011, 31, 925-931.	2.3	86
21	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
22	Coâ€œoccurring diagnoses among FMR1 premutation allele carriers. Clinical Genetics, 2010, 77, 374-381.	2.0	80
23	The cognitive neuropsychological phenotype of carriers of the FMR1 premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 28.	3.1	74
24	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
25	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
26	The National down Syndrome Project: Design and Implementation. Public Health Reports, 2007, 122, 62-72.	2.5	67
27	New clinical findings in the fragile X-associated tremor ataxia syndrome (FXTAS). Neurogenetics, 2011, 12, 123-135.	1.4	67
28	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
29	Premature Ovarian Failure among Fragile X Premutation Carriers: Parent-of-Origin Effect?. American Journal of Human Genetics, 2000, 67, 11-13.	6.2	61
30	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
31	Survey of the fragile X syndrome and the fragile X E syndrome in a special education needs population. , 1996, 64, 428-433.		56
32	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
33	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
34	Reproductive and gynecologic care of women with fragile X primary ovarian insufficiency (FXPOI). Menopause, 2016, 23, 993-999.	2.0	45
35	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
36	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

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37	The FMR1 Premutation and Attention-Deficit Hyperactivity Disorder (ADHD): Evidence for a Complex Inheritance. Behavior Genetics, 2012, 42, 415-422.	2.1	41
38	Altered patterns of multiple recombinant events are associated with nondisjunction of chromosome 21. Human Genetics, 2012, 131, 1039-1046.	3.8	39
39	FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. Pediatrics, 2017, 139, S183-S193.	2.1	39
40	Inherited genetic susceptibility to acute lymphoblastic leukemia in Down syndrome. Blood, 2019, 134, 1227-1237.	1.4	37
41	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
42	Opportunities, barriers, and recommendations in Down syndrome research. Translational Science of Rare Diseases, 2021, 5, 99-129.	1.5	33
43	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
44	Cognitive aspects of Fragile X syndrome. Wiley Interdisciplinary Reviews: Cognitive Science, 2014, 5, 501-508.	2.8	32
45	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
46	Clustering of comorbid conditions among women who carry an FMR1 premutation. Genetics in Medicine, 2020, 22, 758-766.	2.4	31
47	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
48	Genome-Wide Association Study of Down Syndrome-Associated Atrioventricular Septal Defects. G3: Genes, Genomes, Genetics, 2015, 5, 1961-1971.	1.8	28
49	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
50	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
51	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
52	A candidate gene analysis and GWAS for genes associated with maternal nondisjunction of chromosome 21. PLoS Genetics, 2019, 15, e1008414.	3.5	25
53	Contribution of copy-number variation to Down syndromeâ€“associated atrioventricular septal defects. Genetics in Medicine, 2015, 17, 554-560.	2.4	24
54	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

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55	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
56	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
57	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
58	Smarter clustering methods for SNP genotype calling. Bioinformatics, 2008, 24, 2665-2671.	4.1	18
59	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
60	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
61	Evidence for dysregulation of genome-wide recombination in oocytes with nondisjoined chromosomes 21. Human Molecular Genetics, 2014, 23, 408-417.	2.9	17
62	Concordance and recessive inheritance of Leber congenital amaurosis. American Journal of Medical Genetics Part A, 1993, 46, 275-277.	2.4	16
63	Olfactory dysfunction in fragile X tremor ataxia syndrome. Movement Disorders, 2012, 27, 1556-1559.	3.9	16
64	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
65	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
66	An Examination of the Relationship between Hotspots and Recombination Associated with Chromosome 21 Nondisjunction. PLoS ONE, 2014, 9, e99560.	2.5	15
67	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
68	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
69	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
70	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
71	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
72	Genome-Wide Association Study of Meiotic Recombination Phenotypes. G3: Genes, Genomes, Genetics, 2016, 6, 3995-4007.	1.8	9

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73	Symptoms of Autism Spectrum Disorder in Individuals with Down Syndrome. Brain Sciences, 2021, 11, 1278.	2.3	9
74	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
75	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
76	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
77	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
78	Comparison of COVID-19 and Non-COVID-19 Pneumonia in Down Syndrome. Journal of Clinical Medicine, 2021, 10, 3748.	2.4	7
79	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
80	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
81	Regionally Smoothed Meta-Analysis Methods for GWAS Datasets. Genetic Epidemiology, 2016, 40, 154-160.	1.3	6
82	Cliniciansâ€™ experiences with the fragile X clinical and research consortium. American Journal of Medical Genetics, Part A, 2016, 170, 3138-3143.	1.2	6
83	Improving Health Education for Women Who Carry an FMR1 Premutation. Journal of Genetic Counseling, 2016, 25, 228-238.	1.6	6
84	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
85	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
86	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
87	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
88	Testing for contributions of mitochondrial DNA mutations to complex diseases. , 1998, 15, 451-469.		4
89	Preventive care services and health behaviors in children with fragile X syndrome. Disability and Health Journal, 2019, 12, 564-573.	2.8	4
90	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

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91	Predictors of Comorbid Conditions in Women Who Carry an FMR1 Premutation. Frontiers in Psychiatry, 2021, 12, 715922.	2.6	4
92	Modeling the natural history of the fragile X gene. Mental Retardation and Developmental Disabilities Research Reviews, 1995, 1, 263-268.	3.6	3
93	The method of sib-pair linkage analysis in context of case-control design. Genetic Epidemiology, 1997, 14, 939-944.	1.3	3
94	Maternal Age and Oocyte Aneuploidy: Lessons Learned from Trisomy 21. , 2013, , 69-85.		3
95	Relationship between Apgar scores and long-term cognitive outcomes in individuals with Down syndrome. Scientific Reports, 2021, 11, 12707.	3.3	3
96	Stephen T. Warren, Ph.D. (1953â€“2021): A remembrance. American Journal of Human Genetics, 2022, 109, 3-11.	6.2	2
97	Sequential sib-pair and association studies to detect genes in quantitative traits. Genetic Epidemiology, 1997, 14, 885-890.	1.3	1
98	Genetic basis for primary ovarian insufficiency. , 2013, , 394-408.		1
99	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
100	Genotype/Phenotype Relationships in FXTAS. , 2010, , 95-122.		1
101	Genotype/Phenotype Relationships in FXTAS. , 2016, , 129-160.		1
102	Newton E. Morton (1929â€“2018). American Journal of Human Genetics, 2018, 102, 1011-1017.	6.2	0
103	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
104	Men with an FMR1 premutation and their health education needs. Journal of Genetic Counseling, 2021, 30, 1156-1167.	1.6	0
105	P2â€030: THE LIFEâ€DSR STUDY. Alzheimer's and Dementia, 2019, 15, .	0.8	0