Susan M White

List of Publications by Year
in descending order

77 papers	3,759 citations	186265 28 h-index	144013 57 g-index
83 all docs	83 docs citations	83 times ranked	6473 citing authors

#	Article	IF	Citations
1	Exome sequencing for patients with developmental and epileptic encephalopathies in clinical practice. Developmental Medicine and Child Neurology, 2023, 65, 50-57.	2.1	11
2	Lessons learnt from multifaceted diagnostic approaches to the first 150 families in Victoriaâ€™s Undiagnosed Diseases Program. Journal of Medical Genetics, 2022, 59, 748-758.	3.2	9
3	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
4	A systematic review of geographical inequities for accessing clinical genomic and genetic services for non-cancer related rare disease. European Journal of Human Genetics, 2022, 30, 645-652.	2.8	13
5	Is faster better? An economic evaluation of rapid and ultra-rapid genomic testing in critically ill infants and children. Genetics in Medicine, 2022, 24, 1037-1044.	2.4	18
6	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
7	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
8	De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 3, 100111.	1.7	7
9	Monoallelic and biallelic variants in LEF1 are associated with a new syndrome combining ectodermal dysplasia and limb malformations caused by altered WNT signaling. Genetics in Medicine, 2022, 24, 1708-1721.	2.4	4
10	Pathogenic variants causing ABL1 malformation syndrome cluster in a myristoyl-binding pocket and increase tyrosine kinase activity. European Journal of Human Genetics, 2021, 29, 593-603.	2.8	7
11	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.7	10
12	DOORS syndrome and a recurrentÂtruncating ATP6V1B2 variant. Genetics in Medicine, 2021, 23, 149-154.	2.4	11
13	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. Genetics in Medicine, 2021, 23, 183-191.	2.4	70
14	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
15	Paediatric genomic testing: Navigating medicare rebatable genomic testing. Journal of Paediatrics and Child Health, 2021, 57, 477-483.	0.8	8
16	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	6.2	6
17	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
18	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13

#	Article	IF	Citations
19	Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. American Journal of Human Genetics, 2021, 108, 1126-1137.	6.2	14
20	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. European Journal of Medical Genetics, 2021, 64, 104259.	1.3	2
21	Paediatric genomic testing: Navigating genomic reports for the general paediatrician. Journal of Paediatrics and Child Health, 2021, , .	0.8	3
22	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
23	ERCC1mutations impede DNA damage repair and cause liver and kidney dysfunction in patients. Journal of Experimental Medicine, 2021, 218, .	8.5	18
24	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	6.2	5
25	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	2.5	6
26	Response to Ferket et al.. Genetics in Medicine, 2020, 22, 1910.	2.4	0
27	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A (KAT6A). Journal of Genetics and Genomics, 2020, 47, 650-654.	3.9	2
28	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. Genetics in Medicine, 2020, 22, 1986-1993.	2.4	25
29	Microarray diagnosis of autoimmune polyendocrinopathyâ€candidiasisâ€ectodermal dystrophy caused by a novel homozygous intragenic AIRE deletion. Journal of Paediatrics and Child Health, 2020, 57, 1109-1112.	0.8	0
30	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1508.	1.2	44
31	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
32	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a <scp>Threeâ€Generation</scp> Family Using <scp>Shortâ€Read Wholeâ€Genome</scp> Sequencing Data. Movement Disorders, 2020, 35, 1675-1679.	3.9	12
33	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. JAMA - Journal of the American Medical Association, 2020, 323, 2503.	7.4	160
34	Bi-allelic LoF NRROS Variants Impairing Active TGF-Î²1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. American Journal of Human Genetics, 2020, 106, 559-569.	6.2	18
35	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
36	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59

#	Article	IF	Citations
37	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. American Journal of Human Genetics, 2020, 106, 779-792.	6.2	25
38	Does genomic sequencing early in the diagnostic trajectory make a difference? AÂfollow-up study of clinical outcomes and cost-effectiveness. Genetics in Medicine, 2019, 21, 173-180.	2.4	118
39	A head-to-head evaluation of the diagnostic efficacy and costs of trio versus singleton exome sequencing analysis. European Journal of Human Genetics, 2019, 27, 1791-1799.	2.8	37
40	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
41	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca2+-Activated K+ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	6.2	45
42	Long-term economic impacts of exome sequencing for suspected monogenic disorders: diagnosis, management, and reproductive outcomes. Genetics in Medicine, 2019, 21, 2586-2593.	2.4	43
43	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	1.2	20
44	EED and EZH2 constitutive variants: A study to expand the Cohenâ€Gibson syndrome phenotype and contrast it with Weaver syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 588-594.	1.2	24
45	Diagnostic and service impact of genomic testing technologies in a neonatal intensive care unit. Journal of Paediatrics and Child Health, 2019, 55, 1309-1314.	0.8	11
46	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	2.4	32
47	Smith-Lemli-Opitz syndrome: clinical and biochemical correlates. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 451-459.	0.9	29
48	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. Epilepsy Research, 2018, 140, 166-170.	1.6	12
49	Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. European Journal of Human Genetics, 2018, 26, 644-651.	2.8	102
50	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. Genetics in Medicine, 2018, 20, 1554-1563.	2.4	125
51	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. American Journal of Human Genetics, 2018, 102, 557-573.	6.2	69
52	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	2.0	24
53	Maternal inheritance of BDNF deletion, with phenotype of obesity and developmental delay in mother and child. American Journal of Medical Genetics, Part A, 2018, 176, 194-200.	1.2	8
54	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	6.2	17

#	Article	IF	Citations
55	Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. Npj Genomic Medicine, 2018, 3, 16.	3.8	420
56	Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. Genetics in Medicine, 2017, 19, 867-874.	2.4	194
57	A novel AMPD2 mutation outside the AMP deaminase domain causes pontocerebellar hypoplasia type 9. American Journal of Medical Genetics, Part A, 2017, 173, 820-823.	1.2	11
58	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
59	Cohort study of Gorlin syndrome with emphasis on standardised phenotyping and quality of life assessment. Internal Medicine Journal, 2017, 47, 664-673.	0.8	14
60	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
61	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	2.8	24
62	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	6.2	252
63	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
64	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
65	â€œIt wasn't a disaster or anythingâ€: Parentsâ€™ experiences of their child's uncertain chromosomal microarray result. American Journal of Medical Genetics, Part A, 2016, 170, 2895-2904.	1.2	24
66	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	2.4	332
67	The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. Genetics in Medicine, 2016, 18, 483-493.	2.4	127
68	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
69	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
70	Characterization of core clinical phenotypes associated with recurrent proximal 15q25.2 microdeletions. American Journal of Medical Genetics, Part A, 2014, 164, 77-86.	1.2	14
71	Targeted Next-Generation Sequencing Identifies Pathogenic Variants in Familial Congenital Heart Disease. Journal of the American College of Cardiology, 2014, 64, 2498-2506.	2.8	85
72	A Mouse Splice-Site Mutant and Individuals with Atypical Chromosome 22q11.2 Deletions Demonstrate the Crucial Role for Crkl in Craniofacial and Pharyngeal Development. Molecular Syndromology, 2014, 5, 276-286.	0.8	11

#	Article	IF	Citations
73	Drawing attention to difference: Dilemmas in discussing dysmorphism with parents. Journal of Paediatrics and Child Health, 2011, 47, 763-765.	0.8	3
74	Report of a further family with dominant deafnessâ€onychodystrophy (DDOD) syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2512-2515.	1.2	10
75	The phenotype of Floatingâ€“Harbor syndrome in 10 patients. American Journal of Medical Genetics, Part A, 2010, 152A, 821-829.	1.2	43
76	Talking genes. International Journal of Speech-Language Pathology, 2006, 8, 2-6.	0.5	1
77	The adult phenotype in Costello syndrome. American Journal of Medical Genetics, Part A, 2005, 136A, 128-135.	1.2	72