Eric Vilain

List of Publications by Year
in descending order

72 papers	5,350 citations	136950 32 h-index	88630 70 g-index
84 all docs	84 docs citations	84 times ranked	8621 citing authors

#	Article	IF	Citations
1	Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care. Hormone Research in Paediatrics, 2016, 85, 158-180.	1.8	852
2	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
3	Sexual Orientation, Controversy, and Science. Psychological Science in the Public Interest: A Journal of the American Psychological Society, 2016, 17, 45-101.	10.7	401
4	Mutant Cohesin in Premature Ovarian Failure. New England Journal of Medicine, 2014, 370, 943-949.	27.0	244
5	Identification of SOX3 as an XX male sex reversal gene in mice and humans. Journal of Clinical Investigation, 2011, 121, 328-341.	8.2	234
6	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
7	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E333-E344.	3.6	172
8	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. Nature Genetics, 2012, 44, 788-792.	21.4	169
9	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. Genetics in Medicine, 2014, 16, 510-515.	2.4	121
10	Copy Number Variation in Patients with Disorders of Sex Development Due to 46,XY Gonadal Dysgenesis. PLoS ONE, 2011, 6, e17793.	2.5	116
11	De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay. American Journal of Human Genetics, 2015, 96, 498-506.	6.2	115
12	Acceleration of Age-Associated Methylation Patterns in HIV-1-Infected Adults. PLoS ONE, 2015, 10, e0119201.	2.5	101
13	DSDs: genetics, underlying pathologies and psychosexual differentiation. Nature Reviews Endocrinology, 2014, 10, 603-615.	9.6	93
14	A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. Human Molecular Genetics, 2016, 25, 3446-3453.	2.9	90
15	The importance of having two X chromosomes. Philosophical Transactions of the Royal Society B: Biological Sciences, 2016, 371, 20150113.	4.0	89
16	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. Genome Medicine, 2017, 9, 90.	8.2	86
17	The Genetics of Disorders of Sex Development in Humans. Sexual Development, 2014, 8, 262-272.	2.0	83
18	Disorders of sex development (DSD): Clinical service delivery in the United States. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 268-278.	1.6	67

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19	Human SRY inhibits Î²-catenin-mediated transcription. International Journal of Biochemistry and Cell Biology, 2008, 40, 2889-2900.	2.8	63
20	XY Sex reversal associated with a nonsense mutation in SRY. Genomics, 1992, 13, 838-840.	2.9	61
21	Sex-specific neuroprotection by inhibition of the Y-chromosome gene, SRY , in experimental Parkinsonâ€™s disease. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 16577-16582.	7.1	60
22	Genetics of Disorders ofÂSex Development. Endocrinology and Metabolism Clinics of North America, 2017, 46, 519-537.	3.2	55
23	Parental Reports of Stigma Associated with Childâ€™s Disorder of Sex Development. International Journal of Endocrinology, 2015, 2015, 1-15.	1.5	49
24	Long-read single-molecule maps of the functional methylome. Genome Research, 2019, 29, 646-656.	5.5	48
25	In Addition to Stigma: Cognitive and Autism-Related Predictors of Mental Health in Transgender Adolescents. Journal of Clinical Child and Adolescent Psychology, 2023, 52, 212-229.	3.4	45
26	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
27	Sex-Specific Effects of Testosterone on the Sexually Dimorphic Transcriptome and Epigenome of Embryonic Neural Stem/Progenitor Cells. Scientific Reports, 2016, 6, 36916.	3.3	41
28	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
29	New technologies to uncover the molecular basis of disorders of sex development. Molecular and Cellular Endocrinology, 2018, 468, 60-69.	3.2	40
30	Multisystem Inflammatory Syndrome of Children: Subphenotypes, Risk Factors, Biomarkers, Cytokine Profiles, and Viral Sequencing. Journal of Pediatrics, 2021, 237, 125-135.e18.	1.8	40
31	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
32	The Neuroanatomy of Transgender Identity: Mega-Analytic Findings From the ENIGMA Transgender Persons Working Group. Journal of Sexual Medicine, 2021, 18, 1122-1129.	0.6	36
33	MAP3K1â€related gonadal dysgenesis: Six new cases and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 253-259.	1.6	35
34	Towards improved genetic diagnosis of human differences of sex development. Nature Reviews Genetics, 2021, 22, 588-602.	16.3	35
35	The Sex Chromosome Trisomy mouse model of XXY and XYY: metabolism and motor performance. Biology of Sex Differences, 2013, 4, 15.	4.1	31
36	Loss-of-Function Variants in PPP1R12A: From Isolated Sex Reversal to Holoprosencephaly Spectrum and Urogenital Malformations. American Journal of Human Genetics, 2020, 106, 121-128.	6.2	30

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37	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
38	Mutations in STAG2 cause an Xâ€linked cohesinopathy associated with undergrowth, developmental delay, and dysmorphia: Expanding the phenotype in males. Molecular Genetics & Genomic Medicine, 2019, 7, e00501.	1.2	29
39	Sex steroid hormone modulation of neural stem cells: a critical review. Biology of Sex Differences, 2019, 10, 28.	4.1	28
40	Genetic Evidence of the Association of DEAH-Box Helicase 37 Defects With 46,XY Gonadal Dysgenesis Spectrum. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5923-5934.	3.6	26
41	Translating genomics to the clinical diagnosis of disorders/differences of sex development. Current Topics in Developmental Biology, 2019, 134, 317-375.	2.2	25
42	Mutations in the PCNA-binding site of CDKN1C inhibit cell proliferation by impairing the entry into S phase. Cell Division, 2015, 10, 2.	2.4	23
43	Effects of chromosomal sex and hormonal influences on shaping sex differences in brain and behavior: Lessons from cases of disorders of sex development. Journal of Neuroscience Research, 2017, 95, 65-74.	2.9	23
44	Consensus Parameter: Research Methodologies to Evaluate Neurodevelopmental Effects of Pubertal Suppression in Transgender Youth. Transgender Health, 2020, 5, 246-257.	2.5	22
45	Newborn Screening Protocols and Positive Predictive Value for Congenital Adrenal Hyperplasia Vary across the United States. International Journal of Neonatal Screening, 2020, 6, 37.	3.2	19
46	Psychosocial Screening in Disorders/Differences of Sex Development: Psychometric Evaluation of the Psychosocial Assessment Tool. Hormone Research in Paediatrics, 2018, 90, 368-380.	1.8	17
47	Transwomen and the Metabolic Syndrome: Is Orchiectomy Protective?. Transgender Health, 2016, 1, 165-171.	2.5	16
48	Pan-Filovirus Serum Neutralizing Antibodies in a Subset of Congolese Ebolavirus Infection Survivors. Journal of Infectious Diseases, 2018, 218, 1929-1936.	4.0	16
49	Hurdling Over Sex? Sport, Science, and Equity. Archives of Sexual Behavior, 2014, 43, 1035-1042.	1.9	15
50	Is it time to retire fragile X testing as a first-tier test for developmental delay, intellectual disability, and autism spectrum disorder?. Genetics in Medicine, 2017, 19, 1380-1381.	2.4	15
51	Anomalies of Human Sexual Development: Clinical Aspects and Genetic Analysis. Novartis Foundation Symposium, 2008, , 43-56.	1.1	14
52	Regulation of Sex Determination in Mice by a Non-coding Genomic Region. Genetics, 2014, 197, 885-897.	2.9	14
53	The Genetics of Ovotesticular Disorders of Sex Development. Advances in Experimental Medicine and Biology, 2011, 707, 105-106.	1.6	14
54	Gender destinies: assigning gender in Disorders of Sex Developmentâ€Intersex clinics. Sociology of Health and Illness, 2019, 41, 1520-1534.	2.1	13

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55	Transgender Youth Executive Functioning: Relationships with Anxiety Symptoms, Autism Spectrum Disorder, and Gender-Affirming Medical Treatment Status. Child Psychiatry and Human Development, 2022, 53, 1252-1265.	1.9	12
56	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
57	Emerging issues in disorders/differences of sex development (DSD). American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 249-252.	1.6	9
58	Early Infantile Epileptic Encephalopathy with a de novo variant in ZEB2 identified by exome sequencing. European Journal of Medical Genetics, 2016, 59, 70-74.	1.3	8
59	Long reads capture simultaneous enhancerâ€“promoter methylation status for cell-type deconvolution. Bioinformatics, 2021, 37, i327-i333.	4.1	8
60	The gut microbiome in konzo. Nature Communications, 2021, 12, 5371.	12.8	8
61	The unfinished race: 30 years of gender verification in sport. Lancet, The, 2016, 388, 541-543.	13.7	6
62	nanotatoR: a tool for enhanced annotation of genomic structural variants. BMC Genomics, 2021, 22, 10.	2.8	6
63	Motor control and cognition deficits associated with protein carbamoylation in food (cassava) cyanogenic poisoning: Neurodegeneration and genomic perspectives. Food and Chemical Toxicology, 2021, 148, 111917.	3.6	4
64	Facial analysis technology for the detection of Down syndrome in the Democratic Republic of the Congo. European Journal of Medical Genetics, 2021, 64, 104267.	1.3	4
65	An infant with <scp>MLH</scp>3 variants, <scp>FOXG</scp>1â€duplication and multiple, benign cranial and spinal tumors: A clinical exome sequencing study. Genes Chromosomes and Cancer, 2016, 55, 131-142.	2.8	3
66	Science's place in shaping gender-based policies in athletics. Lancet, The, 2019, 393, 1504.	13.7	3
67	Identification of differential hypothalamic DNA methylation and gene expression associated with sexual partner preferences in rams. PLoS ONE, 2022, 17, e0263319.	2.5	3
68	Decision making in differences of sex development/intersex care in the USA: bridging advocacy and family-centred care. Lancet Diabetes and Endocrinology,the, 2022, 10, 381-383.	11.4	2
69	Interpreting Whole-Genome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 296.	7.4	1
70	The End of Compulsory Gender Verification: Is It Progress for Inclusion of Women in Sports?. Archives of Sexual Behavior, 2021, 50, 2799-2807.	1.9	1
71	Disorders of Sex Development. , 2016, , 259-278.		0
72	COVID-19 and the International Academy of Sex Research: We Will Be Back. Archives of Sexual Behavior, 2020, 49, 1401-1401.	1.9	0