William T. Gibson

List of Publications by Year
in descending order

83 papers	3,916 citations	159358 30 h-index	138251 58 g-index
88 all docs	88 docs citations	88 times ranked	7405 citing authors

#	Article	IF	Citations
1	SETD1B-associated neurodevelopmental disorder. Journal of Medical Genetics, 2021, 58, 196-204.	1.5	22
2	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
3	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
4	Searching for Monogenic Diabetes in a High-risk Autoimmune Diabetes Cohort: Needles in a Paperclip Stack. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e3266-e3268.	1.8	0
5	Somatic mosaicism detected by genome-wide sequencing in 500 parentâ€“child trios with suspected genetic disease: clinical and genetic counseling implications. Journal of Physical Education and Sports Management, 2021, 7, a006125.	0.5	8
6	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
7	T regâ€“specific insulin receptor deletion prevents diet-induced and age-associated metabolic syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	32
8	Reciprocal skeletal phenotypes of PRC2-related overgrowth and Rubinsteinâ€“Taybi syndromes: potential role of H3K27 modifications. Journal of Physical Education and Sports Management, 2020, 6, a005058.	0.5	3
9	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	2.6	59
10	Complexity in unclassified auto-inflammatory disease: a case report illustrating the potential for disease arising from the allelic burden of multiple variants. Pediatric Rheumatology, 2019, 17, 70.	0.9	6
11	PRC2â€complex related dysfunction in overgrowth syndromes: A review of EZH2, EED, and SUZ12 and their syndromic phenotypes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 519-531.	0.7	47
12	A distinct neurodevelopmental syndrome with intellectual disability, autism spectrum disorder, characteristic facies, and macrocephaly is caused by defects in CHD8. Journal of Human Genetics, 2019, 64, 271-280.	1.1	35
13	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€”successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	1.3	59
14	Mutations in ILK, encoding integrin-linked kinase, are associated with arrhythmogenic cardiomyopathy. Translational Research, 2019, 208, 15-29.	2.2	33
15	Rare SUZ12 variants commonly cause an overgrowth phenotype. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 532-547.	0.7	23
16	Practice guideline: joint CCMG-SOGC recommendations for the use of chromosomal microarray analysis for prenatal diagnosis and assessment of fetal loss in Canada. Journal of Medical Genetics, 2018, 55, 215-221.	1.5	84
17	A Clinical Review of Generalized Overgrowth Syndromes in the Era of Massively Parallel Sequencing. Molecular Syndromology, 2018, 9, 70-82.	0.3	36
18	Neuronal PAS Domain Protein 4 Suppression of Oxygen Sensing Optimizes Metabolism during Excitation of Neuroendocrine Cells. Cell Reports, 2018, 22, 163-174.	2.9	19

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19	Genetic ablation ofCyp8b1preserves host metabolic function by repressing steatohepatitis and altering gut microbiota composition. American Journal of Physiology - Endocrinology and Metabolism, 2018, 314, E418-E432.	1.8	22
20	The p300 and CBP Transcriptional Coactivators Are Required for Î²-Cell and Î±-Cell Proliferation. Diabetes, 2018, 67, 412-422.	0.3	24
21	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	5.8	70
22	Loss of maternal EED results in postnatal overgrowth. Clinical Epigenetics, 2018, 10, 95.	1.8	34
23	ROHHAD and Prader-Willi syndrome (PWS): clinical and genetic comparison. Orphanet Journal of Rare Diseases, 2018, 13, 124.	1.2	27
24	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo OPA3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a001156.	0.5	11
25	TSC2 c.1864C>T variant associated with mild cases of tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2017, 173, 771-775.	0.7	15
26	A systematic review of genetic syndromes with obesity. Obesity Reviews, 2017, 18, 603-634.	3.1	138
27	Compound heterozygous TRPV4 mutations in two siblings with a complex phenotype including severe intellectual disability and neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 3087-3092.	0.7	13
28	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	2.6	99
29	A Review of the Genetics of Intracranial Berry Aneurysms and Implications for Genetic Counseling. Journal of Genetic Counseling, 2017, 26, 21-31.	0.9	37
30	Ghrelin, Ghrelin O-Acyltransferase, and Carbohydrate Metabolism During Pregnancy in Calorie-Restricted Mice. Hormone and Metabolic Research, 2017, 49, 64-72.	0.7	7
31	A maternal high-fat, high-sucrose diet has sex-specific effects on fetal glucocorticoids with little consequence for offspring metabolism and voluntary locomotor activity in mice. PLoS ONE, 2017, 12, e0174030.	1.1	21
32	Mutations inFLNCare Associated with Familial Restrictive Cardiomyopathy. Human Mutation, 2016, 37, 269-279.	1.1	138
33	Somatic overgrowth associated with homozygous mutations in bothMAN1B1andSEC23A. Journal of Physical Education and Sports Management, 2016, 2, a000737.	0.5	18
34	EED-associated overgrowth in a second male patient. Journal of Human Genetics, 2016, 61, 831-834.	1.1	52
35	Weaver Syndromeâ€Associated EZH2 Protein Variants Show Impaired Histone Methyltransferase Function In Vitro. Human Mutation, 2016, 37, 301-307.	1.1	68
36	Episodic ataxia associated with a de novo SCN2A mutation. European Journal of Paediatric Neurology, 2016, 20, 772-776.	0.7	26

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37	Core Concepts in Human Genetics: Understanding the Complex Phenotype of Sport Performance and Susceptibility to Sport Injury. Medicine and Sport Science, 2016, 61, 1-14.	1.4	4
38	Absence of mutations in HCRT , HCRTR1 and HCRTR2 in patients with ROHHAD. Respiratory Physiology and Neurobiology, 2016, 221, 59-63.	0.7	19
39	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	1.0	323
40	Case Report: Direct Access Genetic Testing and A Falseâ€Positive Result For Long QT Syndrome. Journal of Genetic Counseling, 2016, 25, 25-31.	0.9	3
41	Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): exome sequencing of trios, monozygotic twins and tumours. Orphanet Journal of Rare Diseases, 2015, 10, 103.	1.2	45
42	Vasoactive Intestinal Polypeptide Promotes Intestinal Barrier Homeostasis and Protection Against Colitis in Mice. PLoS ONE, 2015, 10, e0125225.	1.1	43
43	Endocrine Aspects of 4H Leukodystrophy: A Case Report and Review of the Literature. Case Reports in Endocrinology, 2015, 2015, 1-6.	0.2	10
44	Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 345-51.	0.4	10
45	Beneficial metabolic phenotypes caused by lossâ€ofâ€function <scp>APOC3</scp> mutations. Clinical Genetics, 2015, 87, 31-32.	1.0	3
46	A high-fat diet rich in corn oil reduces spontaneous locomotor activity and induces insulin resistance in mice. Journal of Nutritional Biochemistry, 2015, 26, 319-326.	1.9	36
47	Acylated ghrelin is not required for the surge in pituitary growth hormone observed in pregnant mice. Peptides, 2015, 65, 29-33.	1.2	6
48	A novel mutation in EED associated with overgrowth. Journal of Human Genetics, 2015, 60, 339-342.	1.1	75
49	Somatic mosaicism for the p.His1047Arg mutation in PIK3CA in a girl with mesenteric lipomatosis. American Journal of Medical Genetics, Part A, 2014, 164, 2360-2364.	0.7	13
50	Complex genomic rearrangements in the dystrophin gene due to replicationâ€based mechanisms. Molecular Genetics & Genomic Medicine, 2014, 2, 539-547.	0.6	16
51	Duplication of AKT3 is associated with macrocephaly and speech delay. American Journal of Medical Genetics, Part A, 2014, 164, 1868-1869.	0.7	10
52	Exome sequencing identifies mutations in <scp>KIF14</scp> as a novel cause of an autosomal recessive lethal fetal ciliopathy phenotype. Clinical Genetics, 2014, 86, 220-228.	1.0	92
53	Genetic Counseling in Directâ€toâ€Consumer Exome Sequencing: A Case Report. Journal of Genetic Counseling, 2014, 23, 742-753.	0.9	5
54	Biallelic mutations at PPARG cause a congenital, generalized lipodystrophy similar to the Berardinelliâ€“Seip syndrome. European Journal of Medical Genetics, 2014, 57, 524-526.	0.7	30

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55	The c.7409G>A (p.Cys2470Tyr) Variant of <i>FBN1</i>: Phenotypic Variability across Three Generations. Molecular Syndromology, 2013, 4, 125-135.	0.3	5
56	Mutations inPOLR3AandPOLR3Bare a major cause of hypomyelinating leukodystrophies with or without dental abnormalities and/or hypogonadotropic hypogonadism. Journal of Medical Genetics, 2013, 50, 194-197.	1.5	109
57	Mutations in EZH2 Cause Weaver Syndrome. American Journal of Human Genetics, 2012, 90, 110-118.	2.6	253
58	Duchenne muscular dystrophy caused by a complex rearrangement between intron 43 of the DMD gene and chromosome 4. Neuromuscular Disorders, 2011, 21, 178-182.	0.3	19
59	Acute Disruption of Leptin Signaling in Vivo Leads to Increased Insulin Levels and Insulin Resistance. Endocrinology, 2011, 152, 3385-3395.	1.4	37
60	Effects of glucose and insulin on acyl ghrelin and desacyl ghrelin, leptin, and adiponectin in pregnant women with diabetes. Metabolism: Clinical and Experimental, 2010, 59, 841-847.	1.5	15
61	Pseudohypoparathyroidism type 1a and the GNAS p.R231H mutation: Somatic mosaicism in a mother with two affected sons. American Journal of Medical Genetics, Part A, 2010, 152A, 2784-2790.	0.7	17
62	Ldlrâ^'/â^' Mice Display Decreased Susceptibility to Western-Type Diet-Induced Obesity Due to Increased Thermogenesis. Endocrinology, 2010, 151, 5226-5236.	1.4	26
63	The metabolic phenotype of SCD1-deficient mice is independent of melanin-concentrating hormone. Peptides, 2010, 31, 123-129.	1.2	2
64	Genetic association studies for complex traits: relevance for the sports medicine practitioner. British Journal of Sports Medicine, 2009, 43, 314-316.	3.1	13
65	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics, 2009, 10, 526.	1.2	30
66	Clinical study of two brothers with a novel 33 bp duplication in the ARX gene. American Journal of Medical Genetics, Part A, 2009, 149A, 1482-1486.	0.7	17
67	Key Concepts in Human Genetics: Understanding the Complex Phenotype. Medicine and Sport Science, 2009, 54, 1-10.	1.4	5
68	Phenotypeâ€“genotype characterization of alphaâ€thalassemia mental retardation syndrome due to isolated monosomy of 16p13.3. American Journal of Medical Genetics, Part A, 2008, 146A, 225-232.	0.7	21
69	Schinzelâ€“Giedion syndrome: Report of splenopancreatic fusion and proposed diagnostic criteria. American Journal of Medical Genetics, Part A, 2008, 146A, 1299-1306.	0.7	45
70	Absence of stearoyl-CoA desaturase-1 ameliorates features of the metabolic syndrome in LDLR-deficient mice. Journal of Lipid Research, 2008, 49, 217-229.	2.0	59
71	Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children. Journal of Medical Genetics, 2007, 44, 556-561.	1.5	68
72	Mycophenolate Mofetil and Atherosclerosis: Results of Animal and Human Studies. Annals of the New York Academy of Sciences, 2007, 1110, 209-221.	1.8	23

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73	Oligonucleotide Microarray Analysis of Genomic Imbalance in Children with Mental Retardation. American Journal of Human Genetics, 2006, 79, 500-513.	2.6	261
74	The beat goes on: ciliary proteins are defective in Meckel syndrome.. Clinical Genetics, 2006, 69, 400-401.	1.0	3
75	Body weight is modulated by levels of full-length Huntingtin. Human Molecular Genetics, 2006, 15, 1513-1523.	1.4	101
76	A new Marfan-like syndrome caused by perturbed transforming growth factor-Î² signaling. Clinical Genetics, 2005, 68, 330-331.	1.0	1
77	Noonan syndrome in a premature infant with hypertrophic cardiomyopathy and death in infancy. Journal of the National Medical Association, 2005, 97, 805-7.	0.6	5
78	Mutational analysis of the serotonin receptor 5HT2c in severe early-onset human obesity. Canadian Journal of Physiology and Pharmacology, 2004, 82, 426-429.	0.7	22
79	So many asthma loci, so little time. Clinical Genetics, 2004, 66, 107-108.	1.0	0
80	Another four bite the dust: mutations in a ubiquitously expressed filamin protein cause several skeletal dysplasias. Clinical Genetics, 2004, 66, 110-111.	1.0	0
81	Congenital Leptin Deficiency Due to Homozygosity for the Î”133G Mutation: Report of Another Case and Evaluation of Response to Four Years of Leptin Therapy. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 4821-4826.	1.8	245
82	Partial leptin deficiency and human adiposity. Nature, 2001, 414, 34-35.	13.7	356
83	The immunoglobulin heavy chain and disease association: application to pemphigus vulgaris. Human Genetics, 1994, 94, 675-83.	1.8	14