Katrina Tatton-brown

List of Publications by Year
in descending order

33 papers	2,624 citations	279798 23 h-index	395702 33 g-index

35 all docs	35 docs citations	35 times ranked	4415 citing authors

#	Article	IF	Citations
1	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	9.6	388
2	Genotype-Phenotype Associations in Sotos Syndrome: An Analysis of 266 Individuals with NSD1 Aberrations. American Journal of Human Genetics, 2005, 77, 193-204.	6.2	298
3	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	21.4	280
4	Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. American Journal of Human Genetics, 2017, 100, 725-736.	6.2	168
5	Sotos syndrome. European Journal of Human Genetics, 2007, 15, 264-271.	2.8	146
6	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133.	1.8	145
7	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
8	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
9	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	1.2	119
10	Mutations in the PP2A regulatory subunit B family genesPPP2R5B,PPP2R5CandPPP2R5Dcause human overgrowth. Human Molecular Genetics, 2015, 24, 4775-4779.	2.9	85
11	The NSD1 and EZH2 Overgrowth Genes, Similarities and Differences. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 86-91.	1.6	75
12	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	75
13	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
14	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 2015, 24, 2733-2745.	2.9	54
15	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
16	Molecular Mechanisms of Childhood Overgrowth. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 71-75.	1.6	43
17	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	5.5	38
18	Systematic Profiling of DNMT3A Variants Reveals Protein Instability Mediated by the DCAF8 E3 Ubiquitin Ligase Adaptor. Cancer Discovery, 2022, 12, 220-235.	9.4	38

#	Article	IF	Citations
19	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
20	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35
21	Genetics of Growth Disordersâ€"Which Patients Require Genetic Testing?. Frontiers in Endocrinology, 2019, 10, 602.	3.5	33
22	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 502-508.	1.6	31
23	Extending the phenotype associated with the CSNK2A1â€related Okurâ€"Chung syndromeâ€"A clinical study of 11 individuals. American Journal of Medical Genetics, Part A, 2018, 176, 1108-1114.	1.2	28
24	Clinical features of NSD1-positive Sotos syndrome. Clinical Dysmorphology, 2004, 13, 199-204.	0.3	27
25	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
26	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
27	Refining the Primrose syndrome phenotype: A study of five patients with ZBTB20 de novo variants and a review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 344-349.	1.2	16
28	Approach to overgrowth syndromes in the genome era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 483-490.	1.6	15
29	Tattonâ€Brownâ€Rahman syndrome: cognitive and behavioural phenotypes. Developmental Medicine and Child Neurology, 2020, 62, 993-998.	2.1	8
30	Genomics: the power, potential and pitfalls of the new technologies and how they are transforming healthcare. Clinical Medicine, 2019, 19, 269-272.	1.9	6
31	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	2.4	5
32	Delineating the <scp>Smithâ€Kingsmore</scp> syndrome phenotype: Investigation of 16 patients with the <scp>MTOR</scp> c.<scp>5395G</scp>Â>â€‰A p.(<scp>Glu1799Lys</scp>) missense variant. American Journal of Medical Genetics, Part A, 2021, 185, 2445-2454.	1.2	4
33	Unusual association of Mayerâ€"Rokitanskyâ€"KÃ¼sterâ€"Hauser and Sotos syndromes: a case report. Clinical Dysmorphology, 2019, 28, 155-157.	0.3	0