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	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
2	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
3	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
4	Tattonâ€Brownâ€Rahman syndrome: cognitive and behavioural phenotypes. Developmental Medicine and Child Neurology, 2020, 62, 993-998.	2.1	8
5	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
6	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
7	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
8	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
9	Genetics of Growth Disordersâ€”Which Patients Require Genetic Testing?. Frontiers in Endocrinology, 2019, 10, 602.	3.5	33
10	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	5.5	38
11	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35
12	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
13	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
14	Unusual association of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser and Sotos syndromes: a case report. Clinical Dysmorphology, 2019, 28, 155-157.	0.3	0
15	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
16	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
17	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
18	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

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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	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
21	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
22	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
23	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 2015, 24, 2733-2745.	2.9	54
24	Mutations in the PP2A regulatory subunit B family genesPPP2R5B,PPP2R5CandPPP2R5Dcause human overgrowth. Human Molecular Genetics, 2015, 24, 4775-4779.	2.9	85
25	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
26	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	21.4	280
27	Molecular Mechanisms of Childhood Overgrowth. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 71-75.	1.6	43
28	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	1.2	119
29	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
30	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133.	1.8	145
31	Sotos syndrome. European Journal of Human Genetics, 2007, 15, 264-271.	2.8	146
32	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
33	Clinical features of NSD1-positive Sotos syndrome. Clinical Dysmorphology, 2004, 13, 199-204.	0.3	27