

Matthew F Hunter

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

29
papers

1,136
citations

12
h-index

30
g-index

30
ext. papers

1,405
ext. citations

6
avg, IF

3.07
L-index

#	Paper	IF	Citations
29	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. <i>Nature Genetics</i> , 2009 , 41, 829-32	36.3	507
28	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016 , 17, 243	18.3	166
27	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. <i>Genetics in Medicine</i> , 2018 , 20, 1554-1563	8.1	84
26	Phenotypic insights into ADCY5-associated disease. <i>Movement Disorders</i> , 2016 , 31, 1033-40	7	78
25	FOXP1 mutations cause intellectual disability and a recognizable phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 3166-75	2.5	63
24	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. <i>American Journal of Human Genetics</i> , 2015 , 97, 302-10	11	34
23	Alpers syndrome with mutations in POLG: clinical and investigative features. <i>Pediatric Neurology</i> , 2011 , 45, 311-8	2.9	20
22	Maternal attitudes to newborn screening for fragile X syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 301-11	2.5	19
21	A genotype-first approach for the molecular and clinical characterization of uncommon de novo microdeletion of 20q13.33. <i>PLoS ONE</i> , 2010 , 5, e12462	3.7	18
20	Partially methylated alleles, microdeletion, and tissue mosaicism in a fragile X male with tremor and ataxia at 30 years of age: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 3327-3332 ¹⁶	2.5	16
19	Incomplete silencing of full mutation alleles in males with fragile X syndrome is associated with autistic features. <i>Molecular Autism</i> , 2019 , 10, 21	6.5	15
18	Intragenic DNA methylation in buccal epithelial cells and intellectual functioning in a paediatric cohort of males with fragile X. <i>Scientific Reports</i> , 2018 , 8, 3644	4.9	14
17	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. <i>European Journal of Human Genetics</i> , 2019 , 27, 1821-1826	5.3	12
16	Intellectual functioning and behavioural features associated with mosaicism in fragile X syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2019 , 11, 41	4.6	12
15	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 531-535	2.5	8
14	Outfoxed by RBFOX1-a caution about ascertainment bias. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1411-8	2.5	8
13	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. <i>Allergy, Asthma and Clinical Immunology</i> , 2018 , 14, 65	3.2	7

12	Prenatal Diagnosis of Fragile X Syndrome in a Twin Pregnancy Complicated by a Complete Retraction. <i>Genes</i> , 2018 , 9,	4.2	6
11	A familial 7q36.3 duplication associated with agenesis of the corpus callosum. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2201-8	2.5	6
10	DNA Methylation at Birth Predicts Intellectual Functioning and Autism Features in Children with Fragile X Syndrome. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	6
9	The changing face of clinical genetics service delivery in the era of genomics: a framework for monitoring service delivery and data from a comprehensive metropolitan general genetics service. <i>Genetics in Medicine</i> , 2020 , 22, 210-218	8.1	6
8	Wilms tumor in patients with osteopathia striata with cranial sclerosis. <i>European Journal of Human Genetics</i> , 2021 , 29, 396-401	5.3	6
7	FMR1 mRNA from full mutation alleles is associated with ABC-C scores in males with fragile X syndrome. <i>Scientific Reports</i> , 2020 , 10, 11701	4.9	5
6	Functional disomy of proximal Xp causes a distinct phenotype comprising early hypotonia, hypertelorism, small hands and feet, ear abnormalities, myopia and cognitive impairment. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1763-7	2.5	4
5	An Activating Variant in CTNNB1 is Associated with a Sclerosing Bone Dysplasia and Adrenocortical Neoplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	4
4	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. <i>Human Mutation</i> , 2020 , 41, 1884-1891	4.7	4
3	Childhood Hearing Australasian Medical Professionals network: Consensus guidelines on investigation and clinical management of childhood hearing loss. <i>Journal of Paediatrics and Child Health</i> , 2019 , 55, 1013-1022	1.3	3
2	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants.. <i>Genetics in Medicine</i> , 2021 ,	8.1	3
1	Interstitial deletion of chromosome 1 (1p21.1p12) in an infant with congenital diaphragmatic hernia, hydrops fetalis, and interrupted aortic arch. <i>Clinical Case Reports (discontinued)</i> , 2017 , 5, 164-169 ^{0.7}		2