Matthew F Hunter

List of Publications by Year in Descending Order

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	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants.. *Genetics in Medicine*, **2021**,	8.1	3
28	Wilms tumor in patients with osteopathia striata with cranial sclerosis. *European Journal of Human Genetics*, **2021**, 29, 396-401	5.3	6
27	An Activating Variant in CTNNB1 is Associated with a Sclerosing Bone Dysplasia and Adrenocortical Neoplasia. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	4
26	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. *Human Mutation*, **2020**, 41, 1884-1891	4.7	4
25	FMR1 mRNA from full mutation alleles is associated with ABC-C scores in males with fragile X syndrome. *Scientific Reports*, **2020**, 10, 11701	4.9	5
24	DNA Methylation at Birth Predicts Intellectual Functioning and Autism Features in Children with Fragile X Syndrome. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	6
23	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. *Genetics in Medicine*, **2020**, 22, 210-218	8.1	6
22	Incomplete silencing of full mutation alleles in males with fragile X syndrome is associated with autistic features. *Molecular Autism*, **2019**, 10, 21	6.5	15
21	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. *European Journal of Human Genetics*, **2019**, 27, 1821-1826	5.3	12
20	Childhood Hearing Australasian Medical Professionals network: Consensus guidelines on investigation and clinical management of childhood hearing loss. *Journal of Paediatrics and Child Health*, **2019**, 55, 1013-1022	1.3	3
19	Intellectual functioning and behavioural features associated with mosaicism in fragile X syndrome. *Journal of Neurodevelopmental Disorders*, **2019**, 11, 41	4.6	12
18	Intragenic DNA methylation in buccal epithelial cells and intellectual functioning in a paediatric cohort of males with fragile X. *Scientific Reports*, **2018**, 8, 3644	4.9	14
17	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. *Genetics in Medicine*, **2018**, 20, 1554-1563	8.1	84
16	Prenatal Diagnosis of Fragile X Syndrome in a Twin Pregnancy Complicated by a Complete Retraction. *Genes*, **2018**, 9,	4.2	6
15	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. *Allergy, Asthma and Clinical Immunology*, **2018**, 14, 65	3.2	7
14	Interstitial deletion of chromosome 1 (1p21.1p12) in an infant with congenital diaphragmatic hernia, hydrops fetalis, and interrupted aortic arch. *Clinical Case Reports (discontinued)*, **2017**, 5, 164-169	0.7	2
13	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 531-535	2.5	8

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12	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. *Genome Biology*, **2016**, 17, 243	18.3	166
11	Partially methylated alleles, microdeletion, and tissue mosaicism in a fragile X male with tremor and ataxia at 30 years of age: A case report. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3327-3332	2.5	16
10	Phenotypic insights into ADCY5-associated disease. *Movement Disorders*, **2016**, 31, 1033-40	7	78
9	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. *American Journal of Human Genetics*, **2015**, 97, 302-10	11	34
8	A familial 7q36.3 duplication associated with agenesis of the corpus callosum. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2201-8	2.5	6
7	Outfoxed by RBFOX1-a caution about ascertainment bias. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1411-8	2.5	8
6	Maternal attitudes to newborn screening for fragile X syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 301-11	2.5	19
5	FOXP1 mutations cause intellectual disability and a recognizable phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3166-75	2.5	63
4	Alpers syndrome with mutations in POLG: clinical and investigative features. *Pediatric Neurology*, **2011**, 45, 311-8	2.9	20
3	A genotype-first approach for the molecular and clinical characterization of uncommon de novo microdeletion of 20q13.33. *PLoS ONE*, **2010**, 5, e12462	3.7	18
2	Functional disomy of proximal Xp causes a distinct phenotype comprising early hypotonia, hypertelorism, small hands and feet, ear abnormalities, myopia and cognitive impairment. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1763-7	2.5	4
1	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32	36.3	507