Matthew F Hunter

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3580990/publications.pdf

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30 papers	1,594 citations	566801 15 h-index	454577 30 g-index
30 all docs	30 docs citations	30 times ranked	3122 citing authors

#	Article	IF	Citations
1	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	610
2	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	3.8	241
3	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. Genetics in Medicine, 2018, 20, 1554-1563.	1.1	125
4	Phenotypic insights into ADCY5 â€associated disease. Movement Disorders, 2016, 31, 1033-1040.	2.2	106
5	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	2.6	82
6	FOXP1 mutations cause intellectual disability and a recognizable phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 3166-3175.	0.7	79
7	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
8	Intellectual functioning and behavioural features associated with mosaicism in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2019, 11, 41.	1.5	26
9	Alpers Syndrome With Mutations in POLG: Clinical and Investigative Features. Pediatric Neurology, 2011, 45, 311-318.	1.0	24
10	Maternal attitudes to newborn screening for fragile X syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 301-311.	0.7	24
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.	0.7	20
12	Incomplete silencing of full mutation alleles in males with fragile X syndrome is associated with autistic features. Molecular Autism, 2019, 10, 21.	2.6	20
13	A Genotype-First Approach for the Molecular and Clinical Characterization of Uncommon De Novo Microdeletion of 20q13.33. PLoS ONE, 2010, 5, e12462.	1.1	19
14	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.	1.4	19
15	Is faster better? An economic evaluation of rapid and ultra-rapid genomic testing in critically ill infants and children. Genetics in Medicine, 2022, 24, 1037-1044.	1.1	18
16	Intragenic DNA methylation in buccal epithelial cells and intellectual functioning in a paediatric cohort of males with fragile X. Scientific Reports, 2018, 8, 3644.	1.6	17
17	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.	0.4	12
18	FMR1 mRNA from full mutation alleles is associated with ABC-CFX scores in males with fragile X syndrome. Scientific Reports, 2020, 10, 11701.	1.6	11

#	Article	IF	Citations
19	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. Allergy, Asthma and Clinical Immunology, 2018, 14, 65.	0.9	10
20	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.	1.1	10
21	DNA Methylation at Birth Predicts Intellectual Functioning and Autism Features in Children with Fragile X Syndrome. International Journal of Molecular Sciences, 2020, 21, 7735.	1.8	10
22	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	1.4	10
23	Outfoxed by RBFOX1â€A caution about ascertainment bias. American Journal of Medical Genetics, Part A, 2014, 164, 1411-1418.	0.7	9
24	A familial 7q36.3 duplication associated with agenesis of the corpus callosum. American Journal of Medical Genetics, Part A, 2015, 167, 2201-2208.	0.7	9
25	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535.	0.7	9
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.	1.1	8
27	Prenatal Diagnosis of Fragile X Syndrome in a Twin Pregnancy Complicated by a Complete Retraction. Genes, 2018, 9, 287.	1.0	7
28	An Activating Variant inCTNNB1is Associated with a Sclerosing Bone Dysplasia and Adrenocortical Neoplasia. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 688-695.	1.8	7
29	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, 1763-1767.	0.7	4
30	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.	0.2	3