Mathew J Wallis

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5000867/publications.pdf

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30	964	687363 13	501196 28
papers	citations	h-index	g-index

30	30	30	2246
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
2	Any symptom, in any organ, at any age: A case report of multiple genetic diagnoses mimicking mitochondrial disease in an adult with kidney disease. Nephrology, 2022, , .	1.6	1
3	Population DNA screening for medically actionable disease risk in adults. Medical Journal of Australia, 2022, 216, 278-280.	1.7	10
4	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
5	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	5.3	8
6	Lymphedema distichiasis syndrome may be caused by <scp>FOXC2</scp> promoterâ€enhancer dissociation and disruption of a topological associated domain. American Journal of Medical Genetics, Part A, 2021, 185, 150-156.	1.2	4
7	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. Genetics in Medicine, 2021, 23, 183-191.	2.4	70
8	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.6	16
9	Paediatric genomic testing: Navigating medicare rebatable genomic testing. Journal of Paediatrics and Child Health, 2021, 57, 477-483.	0.8	8
10	Cryopyrin-associated periodic syndrome: a treatable genetic inflammatory condition. Practical Neurology, 2021, 21, 424-426.	1.1	1
11	Small interstitial 9p24.3 deletions principally involving KANK1 are likely benign copy number variants. European Journal of Medical Genetics, 2020, 63, 103618.	1.3	2
12	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
13	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
14	Genetic Kidney Disease in Southern Tasmania. Kidney International Reports, 2020, 5, 534-537.	0.8	1
15	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a <scp>Threeâ€Generation</scp> Family Using <scp>Shortâ€Read Wholeâ€Genome</scp> Sequencing Data. Movement Disorders, 2020, 35, 1675-1679.	3.9	12
16	Prospective Evaluation of the Utility of Whole Exome Sequencing in Dilated Cardiomyopathy. Journal of the American Heart Association, 2020, 9, e013346.	3.7	28
17	Homologous recombination DNA repair defects in PALB2-associated breast cancers. Npj Breast Cancer, 2019, 5, 23.	5.2	39
18	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31

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19	A novel approach to offering additional genomic findingsâ€"A protocol to test a twoâ€step approach in the healthcare system. Journal of Genetic Counseling, 2019, 28, 388-397.	1.6	14
20	Loss of TOP3B leads to increased R-loop formation and genome instability. Open Biology, 2019, 9, 190222.	3.6	46
21	Heritable DNA methylation marks associated with susceptibility to breast cancer. Nature Communications, 2018, 9, 867.	12.8	76
22	Surprisingly good outcome in antenatal diagnosis of severe hydrocephalus related to CCDC88C deficiency. European Journal of Medical Genetics, 2018, 61, 189-196.	1.3	9
23	A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. Genetics in Medicine, 2018, 20, 1061-1068.	2.4	37
24	Blake's pouch cyst in 13q deletion syndrome: Posterior fossa malformations may occur due to disruption of multiple genes. American Journal of Medical Genetics, Part A, 2017, 173, 2442-2445.	1.2	8
25	A balanced paternal interchromosomal reciprocal insertion between 5q12.1q13.2 and 20p12.3p12.1 resulting in separate genetic conditions in two siblings. American Journal of Medical Genetics, Part A, 2016, 170, 1930-1933.	1.2	2
26	Fanconi anemia in 55â€yearâ€old identical twins first presenting as fatal postâ€chemotherapy pancytopenia. American Journal of Hematology, 2016, 91, 1273-1276.	4.1	2
27	Dual genetic diagnoses: Atypical handâ€footâ€genital syndrome and developmental delay due to de novo mutations in HOXA13 and NRXN1. American Journal of Medical Genetics, Part A, 2016, 170, 717-724.	1.2	11
28	COMMD1 is linked to the WASH complex and regulates endosomal trafficking of the copper transporter ATP7A. Molecular Biology of the Cell, 2015, 26, 91-103.	2.1	200
29	A severe phenotype of Gitelman syndrome with increased prostaglandin excretion and favorable response to indomethacin. CKJ: Clinical Kidney Journal, 2014, 7, 306-310.	2.9	13
30	CCDC22 deficiency in humans blunts activation of proinflammatory NF-ÎºB signaling. Journal of Clinical Investigation, 2013, 123, 2244-2256.	8.2	101