Julie Mcgaughran

List of Publications by Year
in descending order

38 papers	2,111 citations	394421 19 h-index	315739 38 g-index
39 all docs	39 docs citations	39 times ranked	3594 citing authors

#	Article	IF	Citations
1	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
2	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
3	Recommendations for next generation sequencing data reanalysis of unsolved cases with suspected Mendelian disorders: A systematic review and meta-analysis. Genetics in Medicine, 2022, 24, 1618-1629.	2.4	20
4	Gene selection for the Australian Reproductive Genetic Carrier Screening Project (â€œMackenzieâ€™s) Tj ETQq0 0 0 rgBT /Overlock 10	2.8	60
5	Clinical delineation, sex differences, and genotypeâ€“phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. Genetics in Medicine, 2021, 23, 1202-1210.	2.4	30
6	â€œThis is my boyâ€™s health! Talk straight to me!â€•perspectives on accessible and culturally safe care among Aboriginal and Torres Strait Islander patients of clinical genetics services. International Journal for Equity in Health, 2021, 20, 103.	3.5	9
7	Investigation of current models of care for genetic heart disease in Australia: A national clinical audit. International Journal of Cardiology, 2021, 330, 128-134.	1.7	2
8	A novel ARX loss of function variant in female monozygotic twins is associated with chorea. European Journal of Medical Genetics, 2021, 64, 104315.	1.3	3
9	Constraint and conservation of pairedâ€type homeodomains predicts the clinical outcome of missense variants of uncertain significance. Human Mutation, 2020, 41, 1407-1424.	2.5	2
10	Hypertrophic Cardiomyopathy: Challenging the Status Quo?. Heart Lung and Circulation, 2020, 29, 556-565.	0.4	7
11	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
12	A tyrosine kinase-activating variant Asn666Ser in PDGFRB causes a progeria-like condition in the severe end of Penttinen syndrome. European Journal of Human Genetics, 2019, 27, 574-581.	2.8	20
13	Psychosocial Implications of Living with Catecholaminergic Polymorphic Ventricular Tachycardia in Adulthood. Journal of Genetic Counseling, 2018, 27, 549-557.	1.6	21
14	KBG syndrome: An Australian experience. American Journal of Medical Genetics, Part A, 2017, 173, 1866-1877.	1.2	25
15	Impact of the implantable cardioverter defibrillator on confidence to undertake physical activity in inherited heart disease: A cross-sectional study. European Journal of Cardiovascular Nursing, 2017, 16, 742-752.	0.9	10
16	Position Statement on the Diagnosis and Management of Familial Dilated Cardiomyopathy. Heart Lung and Circulation, 2017, 26, 1127-1132.	0.4	11
17	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
18	A multidisciplinary renal genetics clinic improves patient diagnosis. Medical Journal of Australia, 2016, 204, 58-59.	1.7	31

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19	Factors influencing uptake of familial long QT syndrome genetic testing. American Journal of Medical Genetics, Part A, 2016, 170, 418-425.	1.2	43
20	Expanding the genotypic spectrum of CCBE1 mutations in Hennekam syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2694-2697.	1.2	7
21	CCC- and WASH-mediated endosomal sorting of LDLR is required for normal clearance of circulating LDL. Nature Communications, 2016, 7, 10961.	12.8	165
22	A Prospective Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	27.0	619
23	Clinical and genetic features of Australian families with long QT syndrome: A registryâ€based study. Journal of Arrhythmia, 2016, 32, 456-461.	1.2	9
24	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
25	Antisenseâ€mediated exon skipping: a therapeutic strategy for titinâ€based dilated cardiomyopathy. EMBO Molecular Medicine, 2015, 7, 562-576.	6.9	94
26	A protocol for the identification and validation of novel genetic causes of kidney disease. BMC Nephrology, 2015, 16, 152.	1.8	8
27	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
28	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
29	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	21.4	130
30	Delay to diagnosis amongst patients with catecholaminergic polymorphic ventricular tachycardia. International Journal of Cardiology, 2014, 176, 1402-1404.	1.7	20
31	Clinical predictors of genetic testing outcomes in hypertrophic cardiomyopathy. Genetics in Medicine, 2013, 15, 972-977.	2.4	110
32	A cost-effectiveness model of genetic testing for the evaluation of families with hypertrophic cardiomyopathy. Heart, 2012, 98, 625-630.	2.9	119
33	Pleural malignancy in a 22â€yearâ€old female with a chromosome 22q13 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 2362-2363.	1.2	0
34	Progressive edema leading to pleural effusions in a female with a ring chromosome 22 leading to a 22q13 deletion. Clinical Dysmorphology, 2010, 19, 28-29.	0.3	8
35	Nasal encephalocele in a child with mosaic trisomy 14. Clinical Dysmorphology, 2009, 18, 164-165.	0.3	4
36	Establishment of an Australian National Genetic Heart Disease Registry. Heart Lung and Circulation, 2008, 17, 463-467.	0.4	13

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37	A case of Beareâ€“Stevenson syndrome with a broad spectrum of features and a review of the FGFR2 Y375C mutation phenotype. Clinical Dysmorphology, 2006, 15, 89-93.	0.3	23
38	Recurrence of Mowat-Wilson syndrome in siblings with the same proven mutation. American Journal of Medical Genetics, Part A, 2005, 137A, 302-304.	1.2	41