Gail E Graham

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4143258/publications.pdf

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19	455	840776 11	839539 18
papers	citations	h-index	g-index

20	20	20	1190
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	2.0	86
2	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71
3	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474.	3.2	64
4	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	3.4	35
5	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. American Journal of Medical Genetics, Part A, 2018, 176, 1688-1691.	1.2	28
6	Phenotypic spectrum of Auâ€“Kline syndrome: a report of six new cases and review of the literature. European Journal of Human Genetics, 2018, 26, 1272-1281.	2.8	26
7	Benchmarking outcomes in the Neonatal Intensive Care Unit: Cytogenetic and molecular diagnostic rates in a retrospective cohort. American Journal of Medical Genetics, Part A, 2017, 173, 1839-1847.	1.2	25
8	A retrospective review of 48 individuals, including 12 families, molecularly diagnosed with hereditary leiomyomatosis and renal cell cancer (HLRCC). Familial Cancer, 2018, 17, 615-620.	1.9	25
9	An attenuated phenotype of Costello syndrome in three unrelated individuals with a HRAS c.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences. American Journal of Medical Genetics, Part A, 2015, 167, 2085-2097.	1.2	20
10	Pathogenic variants in CDC45 on the remaining allele in patients with a chromosome 22q11.2 deletion result in a novel autosomal recessive condition. Genetics in Medicine, 2020, 22, 326-335.	2.4	17
11	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	1.2	11
12	Tattonâ€Brownâ€Rahman syndrome: Six individuals with novel features. American Journal of Medical Genetics, Part A, 2020, 182, 673-680.	1.2	11
13	A pilot eConsultation service in Eastern Ontario: bridging clinical genetics and primary care. European Journal of Human Genetics, 2019, 27, 1026-1032.	2.8	8
14	Myotonic Myopathy With Secondary Joint and Skeletal Anomalies From the c.2386C>G, p.L796V Mutation in SCN4A. Frontiers in Neurology, 2020, 11, 77.	2.4	8
15	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
16	Severe craniosynostosis in an infant with deletion 22q11.2 syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 153-157.	1.2	5
17	Knowledge of genetic testing for hereditary kidney cancer in Canada is lacking: The results of the Canadian national hereditary kidney cancer needs assessment survey. Canadian Urological Association Journal, 2014, 8, 832.	0.6	5
18	Application of Canadian hereditary renal cell carcinoma risk criteria to a population database.. Journal of Clinical Oncology, 2018, 36, 621-621.	1.6	0

#	Article	IF	Citations
19	A 79â€‰kb paternally inherited 7q32.2 microdeletion involving <scp> MEST </scp> in a patient with a <scp>Silverâ€‰Russell</scp> syndromeâ€‰like phenotype. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0