

Jean McGowan-Jordan

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8977566/publications.pdf>

Version: 2024-02-01

15
papers

1,349
citations

1307594

7
h-index

1199594

12
g-index

16
all docs

16
docs citations

16
times ranked

3989
citing authors

#	ARTICLE	IF	CITATIONS
1	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. <i>Human Mutation</i> , 2016, 37, 564-569.	2.5	1,194
2	Update on molecular findings in rhabdomyosarcoma. <i>Pathology</i> , 2017, 49, 238-246.	0.6	40
3	Molecular Analysis of Cystinosis: Probable Irish Origin of the Most Common French Canadian Mutation. <i>European Journal of Human Genetics</i> , 1999, 7, 671-678.	2.8	35
4	Reinterpretation of sequence variants: one diagnostic laboratory's experience, and the need for standard guidelines. <i>Genetics in Medicine</i> , 2018, 20, 365-368.	2.4	28
5	Genotype-phenotype characterization in 13 individuals with chromosome Xp11.22 duplications. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 967-977.	1.2	11
6	Use of multicolor fluorescence in situ hybridization to detect deletions in clinical tissue sections. <i>Laboratory Investigation</i> , 2018, 98, 403-413.	3.7	10
7	Variable developmental delays and characteristic facial features: A novel 7p22.3p22.2 microdeletion syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1593-1600.	1.2	9
8	A rare case of pediatric lipoma with t(9;12)(p22;q14) and evidence of HMGA2-NFIB gene fusion. <i>Cancer Genetics</i> , 2017, 216-217, 100-104.	0.4	7
9	Genetic Diagnostic Testing for Inherited Cardiomyopathies. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 437-448.	2.8	7
10	Atypical Hepatic Mesenchymal Hamartoma: Histologic Appearance, Immunophenotype, and Molecular Findings. <i>Pediatric and Developmental Pathology</i> , 2019, 22, 365-369.	1.0	3
11	Leveraging the power of new molecular technologies in the clinical setting requires unprecedented awareness of limitations and drawbacks: experience of one diagnostic laboratory. <i>Journal of Medical Genetics</i> , 2019, 56, 408-412.	3.2	3
12	CML with complex chromosome rearrangements and dysplastic megakaryocytes. <i>Blood</i> , 2016, 128, 604-604.	1.4	0
13	MDS with isolated del(5q) and internuclear bridging. <i>Blood</i> , 2017, 129, 2333-2333.	1.4	0
14	Adopting High-Resolution Allele Frequencies Substantially Expedites Variant Interpretation in Genetic Diagnostic Laboratories. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 602-611.	2.8	0
15	Porokeratotic eccrine ostial and dermal duct nevus associated with an 11 megabase 3p deletion. <i>Pediatric Dermatology</i> , 2022, 39, 107-111.	0.9	0