

Dylan A Mordaunt

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9579127/publications.pdf>

Version: 2024-02-01

24
papers

567
citations

1163117

8
h-index

794594

19
g-index

29
all docs

29
docs citations

29
times ranked

1524
citing authors

#	ARTICLE	IF	CITATIONS
1	Re: "Remote Patient Monitoring: A Systematic Review" by Farias et al.. Telemedicine Journal and E-Health, 2021, 27, 374-377.	2.8	1
2	See How They Grow: Testing the feasibility of a mobile app to support parents'™ understanding of child growth charts. PLoS ONE, 2021, 16, e0246045.	2.5	10
3	On Clinical Utility and Systematic Reporting in Case Studies of Healthcare Process Mining. Comment on: 10.3390/jerph17041348 "Towards the Use of Standardised Terms in Clinical Case Studies for Process Mining in Healthcare" International Journal of Environmental Research and Public Health, 2020, 17, 8298.	2.6	0
4	<p>Interventions for Post-Stroke Shoulder Pain: An Overview of Systematic Reviews</p>. International Journal of General Medicine, 2020, Volume 13, 1411-1426.	1.8	16
5	Healthcare pathway discovery and probabilistic machine learning. International Journal of Medical Informatics, 2020, 137, 104087.	3.3	29
6	Sharing (data) is caring for patients with pachyonychia congenita. British Journal of Dermatology, 2020, 182, 537-537.	1.5	1
7	Metabolomics to Improve the Diagnostic Efficiency of Inborn Errors of Metabolism. International Journal of Molecular Sciences, 2020, 21, 1195.	4.1	30
8	Clinical implications of discordant massarray and sanger sequencing results in cystic fibrosis newborn screening. Pathology, 2020, 52, S60.	0.6	0
9	Review of 12 months of copy number variant calling on a clinical next generation sequencing pipeline. Pathology, 2020, 52, S108.	0.6	0
10	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. Npj Genomic Medicine, 2019, 4, 28.	3.8	16
11	High Yield of Pathogenic Germline Mutations Causative or Likely Causative of the Cancer Phenotype in Selected Children with Cancer. Clinical Cancer Research, 2018, 24, 1594-1603.	7.0	52
12	Pachyonychia Congenita: A Spectrum of <sc>KRT</sc>6a Mutations in Australian Patients. Pediatric Dermatology, 2016, 33, 337-342.	0.9	20
13	Case 2: A Late Preterm Newborn with Cyanosis. NeoReviews, 2016, 17, e166-e169.	0.8	0
14	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	2.4	332
15	Does IARS2 Deficiency Cause an Intrinsic Disorder of Bone Development (Skeletal Dysplasia) or Are the Reported Skeletal Changes Secondary to Growth Hormone Deficiency and Neuromuscular Involvement?. Human Mutation, 2015, 36, 388-388.	2.5	4
16	Phenotypic variation of <i>TTC19</i>â€deficient mitochondrial complex III deficiency: A case report and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1330-1336.	1.2	31
17	Presentation of m.3243A>G (MTâ€L1; tRNA^{Leu}) variant with focal neurology in infancy. American Journal of Medical Genetics, Part A, 2015, 167, 2697-2701.	1.2	3
18	HDAC8â€deficiency causes an Xâ€linked dominant disorder with a wide range of severity. Clinical Genetics, 2015, 88, 98-98.	2.0	4

#	ARTICLE	IF	CITATIONS
19	Protein-Losing Enteropathy in a Patient on Ketogenic Diet for Limbic Encephalitis—Treatment Effect or Underlying Pathology?. <i>Pediatric Neurology</i> , 2015, 52, e11.	2.1	1
20	8q13.1-q13.2 Deletion Associated With Inferior Cerebellar Vermian Hypoplasia and Digital Anomalies: A New Syndrome?. <i>Pediatric Neurology</i> , 2015, 52, 230-234.e1.	2.1	3
21	Uptake and Diagnostic Yield of Chromosomal Microarray in an Australian Child Development Clinic. <i>Children</i> , 2014, 1, 21-30.	1.5	6
22	Congenital coronary anomalies are not uncommon and consideration should be given to screening first degree relatives. <i>Pathology</i> , 2014, 46, 669-670.	0.6	1
23	Diagnostic yield and cost-utility analysis of genetic investigations for assessing children with autism in an australian metropolitan child development service. <i>Pathology</i> , 2014, 46, S91.	0.6	0
24	Translating the Digital Health Conceptual Framework into population health practice. <i>Journal of the International Society for Telemedicine and EHealth</i> , 0, 8, .	0.0	1