# Dylan A Mordaunt

List of Publications by Year
in descending order

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

Version: 2024-02-01

| | | 1163117 | 794594 |
|---|---|---|---|
| 24 | 567 | 8 | 19 |
| papers | citations | h-index | g-index |

| | | | |
|---|---|---|---|
| 29 | 29 | 29 | 1524 |
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | 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 |
| 2 | 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 |
| 3 | Phenotypic variation of TTC19â€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 |
| 4 | Metabolomics to Improve the Diagnostic Efficiency of Inborn Errors of Metabolism. International Journal of Molecular Sciences, 2020, 21, 1195. | 4.1 | 30 |
| 5 | Healthcare pathway discovery and probabilistic machine learning. International Journal of Medical Informatics, 2020, 137, 104087. | 3.3 | 29 |
| 6 | Pachyonychia Congenita: A Spectrum of <scp>KRT</scp>6a Mutations in Australian Patients. Pediatric Dermatology, 2016, 33, 337-342. | 0.9 | 20 |
| 7 | Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. Npj Genomic Medicine, 2019, 4, 28. | 3.8 | 16 |
| 8 | <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 |
| 9 | See How They Grow: Testing the feasibility of a mobile app to support parentsâ€™ understanding of child growth charts. PLoS ONE, 2021, 16, e0246045. | 2.5 | 10 |
| 10 | Uptake and Diagnostic Yield of Chromosomal Microarray in an Australian Child Development Clinic. Children, 2014, 1, 21-30. | 1.5 | 6 |
| 11 | 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 |
| 12 | HDAC8â€deficiency causes an Xâ€linked dominant disorder with a wide range of severity. Clinical Genetics, 2015, 88, 98-98. | 2.0 | 4 |
| 13 | Presentation of m.3243A>G (MTâ€TL1; tRNALeu) variant with focal neurology in infancy. American Journal of Medical Genetics, Part A, 2015, 167, 2697-2701. | 1.2 | 3 |
| 14 | 8q13.1-q13.2 Deletion Associated With Inferior Cerebellar Vermian Hypoplasia and Digital Anomalies: A New Syndrome?. Pediatric Neurology, 2015, 52, 230-234.e1. | 2.1 | 3 |
| 15 | Congenital coronary anomalies are not uncommon and consideration should be given to screening first degree relatives. Pathology, 2014, 46, 669-670. | 0.6 | 1 |
| 16 | Protein-Losing Enteropathy in a Patient on Ketogenic Diet for Limbic Encephalitisâ€”Treatment Effect or Underlying Pathology?. Pediatric Neurology, 2015, 52, e11. | 2.1 | 1 |
| 17 | Sharing (data) is caring for patients with pachyonychia congenita. British Journal of Dermatology, 2020, 182, 537-537. | 1.5 | 1 |
| 18 | Re: â€œRemote Patient Monitoring: A Systematic Reviewâ€•by Farias et al.. Telemedicine Journal and E-Health, 2021, 27, 374-377. | 2.8 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Translating the Digital Health Conceptual Framework into population health practice. Journal of the International Society for Telemedicine and EHealth, 0, 8, . | 0.0 | 1 |
| 20 | Diagnostic yield and cost-utility analysis of genetic investigations for assessing children with autism in an australian metropolitan child development service. Pathology, 2014, 46, S91. | 0.6 | 0 |
| 21 | Case 2: A Late Preterm Newborn with Cyanosis. NeoReviews, 2016, 17, e166-e169. | 0.8 | 0 |
| 22 | On Clinical Utility and Systematic Reporting in Case Studies of Healthcare Process Mining. Comment on: 10.3390/ijerph17041348 â€œTowards the Use of Standardised Terms in Clinical Case Studies for Process Mining in Healthcareâ€• International Journal of Environmental Research and Public Health, 2020, 17, 8298. | 2.6 | 0 |
| 23 | Clinical implications of discordant massarray and sanger sequencing results in cystic fibrosis newborn screening. Pathology, 2020, 52, S60. | 0.6 | 0 |
| 24 | Review of 12 months of copy number variant calling on a clinical next generation sequencing pipeline. Pathology, 2020, 52, S108. | 0.6 | 0 |