John Dean

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/9020036/publications.pdf

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23 papers	1,934 citations	706676 14 h-index	721071 23 g-index
23 all docs	23 docs citations	23 times ranked	4445 citing authors

#	Article	IF	Citations
1	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	0.7	12
2	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. Archives of Disease in Childhood, 2020, 105, 384-389.	1.0	3
3	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	3.7	34
4	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
5	Deletion of Exon 1 in AMER1 in Osteopathia Striata with Cranial Sclerosis. Genes, 2020, 11, 1439.	1.0	7
6	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	2.6	33
7	Parentsâ€™ views of genetic testing and treatment of familial hypercholesterolemia in children: a qualitative study. Journal of Community Genetics, 2019, 10, 129-141.	0.5	9
8	An actionable KCNH2 Long QT Syndrome variant detected by sequence and haplotype analysis in a population research cohort. Scientific Reports, 2019, 9, 10964.	1.6	17
9	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
10	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	6.0	158
11	Elevated plasma levels of cardiac troponin-I predict left ventricular systolic dysfunction in patients with myotonic dystrophy type 1: A multicentre cohort follow-up study. PLoS ONE, 2017, 12, e0174166.	1.1	13
12	Structural and electrical cardiac abnormalities are prevalent in asymptomatic adults with myotonic dystrophy. Heart, 2016, 102, 1472-1478.	1.2	32
13	A New, Atypical Case of Cobalamin F Disorder Diagnosed by Whole Exome Sequencing. Molecular Syndromology, 2015, 6, 254-258.	0.3	8
14	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	1.1	178
15	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	0.7	17
16	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359.	9.4	219
17	Heterozygous Mutations in TREX1 Cause Familial Chilblain Lupus and Dominant Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 80, 811-815.	2.6	339
18	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375

#	Article	IF	Citations
19	Fetal anticonvulsant syndromes and polymorphisms in MTHFR, MTR, and MTRR. American Journal of Medical Genetics, Part A, 2007, 143A, 2303-2311.	0.7	25
20	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. Human Genetics, 2002, 111, 421-427.	1.8	45
21	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	1.4	147
22	Antenatal cystic fibrosis carrier screeningâ€"whether, when and how?. Paediatric and Perinatal Epidemiology, 1993, 7, 368-375.	0.8	6
23	Screening for cystic fibrosis. Lancet, The, 1991, 338, 1524-1525.	6.3	10