Kyle Retterer

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/120719/publications.pdf

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42 papers	4,491 citations	182225 30 h-index	325983 40 g-index
44 all docs	44 docs citations	44 times ranked	10072 citing authors

#	Article	IF	Citations
1	eP281: SeqFirst-neo: Improving access equity for a precise genetic diagnosis in the NICU. Genetics in Medicine, 2022, 24, S178.	1.1	0
2	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
3	Molecular Diagnostic Yield of Exome Sequencing in Patients With Cerebral Palsy. JAMA - Journal of the American Medical Association, 2021, 325, 467.	3.8	64
4	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	2.6	42
5	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
6	Uniparental disomy in a population of 32,067 clinical exome trios. Genetics in Medicine, 2021, 23, 1101-1107.	1.1	31
7	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	3.9	38
8	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
9	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
10	Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease. New England Journal of Medicine, 2020, 383, 2628-2638.	13.9	580
11	The tale of two genes: from next-generation sequencing to phenotype. Journal of Physical Education and Sports Management, 2020, 6, a004846.	0.5	3
12	Mobile element insertion detection in 89,874 clinical exomes. Genetics in Medicine, 2020, 22, 974-978.	1.1	42
13	Age-adjusted association of homologous recombination genes with ovarian cancer using clinical exomes as controls. Hereditary Cancer in Clinical Practice, 2019, 17, 19.	0.6	7
14	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
15	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	1.4	19
16	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	2.6	84
17	Diagnostic outcomes for genetic testing of 70 genes in 8565 patients with epilepsy and neurodevelopmental disorders. Epilepsia, 2018, 59, 1062-1071.	2.6	218
18	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51

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19	High frequency of mosaic pathogenic variants in genes causing epilepsy-related neurodevelopmental disorders. Genetics in Medicine, 2018, 20, 403-410.	1.1	131
20	De novo missense variants in MEIS2 recapitulate the microdeletion phenotype of cardiac and palate abnormalities, developmental delay, intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2018, 176, 1845-1851.	0.7	21
21	Holoprosencephaly: A clinical genomics perspective. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 194-197.	0.7	5
22	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. American Journal of Human Genetics, 2018, 102, 858-873.	2.6	65
23	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
24	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46
25	Whole-exome sequencing on deceased fetuses with ultrasound anomalies: expanding our knowledge of genetic disease during fetal development. Genetics in Medicine, 2017, 19, 1171-1178.	1.1	121
26	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
27	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
28	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	2.6	83
29	Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164.	0.7	31
30	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	0.7	32
31	De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. Human Genetics, 2016, 135, 699-705.	1.8	47
32	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	1.8	40
33	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	2.6	75
34	Mutations in TKT Are the Cause of a Syndrome Including Short Stature, Developmental Delay, and Congenital Heart Defects. American Journal of Human Genetics, 2016, 98, 1235-1242.	2.6	31
35	Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704.	1.1	780
36	De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49.	0.7	61

#	Article	IF	Citations
37	De novo POGZ mutations are associated with neurodevelopmental disorders and microcephaly. Journal of Physical Education and Sports Management, 2015, 1, a000455.	0.5	51
38	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
39	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	1.5	68
40	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	0.7	54
41	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	2.6	134
42	Assessing copy number from exome sequencing and exome array CGH based on CNV spectrum in a large clinical cohort. Genetics in Medicine, 2015, 17, 623-629.	1.1	106