Kyle Retterer

List of Publications by Year in descending order

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#	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	Ο
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 <i>UBA1</i> 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	<i>De novo</i> missense variants in <i>MEIS2</i> 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 in <i>HECW2</i> are 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

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37	De novo <i>POGZ</i> 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 in <i>SLC1A4</i> , 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