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List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704.	2.4	780
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
3	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.	6.2	230
4	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
5	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	6.2	134
6	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
7	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.	3.2	68
8	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	1.4	54
9	Mutations in <i>COQ4</i> , an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	3.2	48
10	De novo missense variants in <i>HECW2</i> are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	3.2	46
11	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
12	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	1.4	32
13	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
14	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28
15	<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.	1.2	21
16	A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586.	1.6	16