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List of Publications by Year
in descending order

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13	3,654	933447 10	1199594 12
papers	citations	h-index	g-index

13	13	13	4163
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. Cell, 1991, 65, 905-914.	28.9	3,285
2	Mutation in the AP4M1 Gene Provides a Model for Neuroaxonal Injury in Cerebral Palsy. American Journal of Human Genetics, 2009, 85, 40-52.	6.2	156
3	Allelic imbalance of expression and epigenetic regulation within the alpha-synuclein wild-type and p.Ala53Thr alleles in Parkinson disease. Human Mutation, 2010, 31, 685-691.	2.5	53
4	A comparison of genotyping arrays. European Journal of Human Genetics, 2021, 29, 1611-1624.	2.8	43
5	Reduced penetrance of pathogenic ACMG variants in a deeply phenotyped cohort study and evaluation of ClinVar classification over time. Genetics in Medicine, 2020, 22, 1812-1820.	2.4	24
6	Boston type craniosynostosis: Report of a second mutation in MSX2. American Journal of Medical Genetics, Part A, 2013, 161, 2626-2633.	1.2	23
7	Linkage analysis and whole exome sequencing identify a novel candidate gene in a Dutch multiple sclerosis family. Multiple Sclerosis Journal, 2019, 25, 909-917.	3.0	19
8	Complex craniosynostosis is associated with the 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 244-253.	1.2	16
9	The Genetics of Atypical Femur Fracturesâ€"a Systematic Review. Current Osteoporosis Reports, 2021, 19, 123-130.	3.6	15
10	CXorf56, a dendritic neuronal protein, identified as a new candidate gene for X-linked intellectual disability. European Journal of Human Genetics, 2018, 26, 552-560.	2.8	12
11	Unbalanced der(5)t(5;20) translocation associated with megalencephaly, perisylvian polymicrogyria, polydactyly and hydrocephalus. American Journal of Medical Genetics, Part A, 2010, 152A, 1488-1497.	1.2	6
12	Congenital hypopituitarism in two brothers with a duplication of the â€˜acrogigantism geneâ€™ GPR101: clinical findings and review of the literature. Pituitary, 2021, 24, 229-241.	2.9	2
13	Reply: PRKAR1B mutations are a rare cause of FUS negative neuronal intermediate filament inclusion disease. Brain, 2015, 138, e358-e358.	7.6	0