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List of Publications by Year in descending order

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13 papers

3,654 citations

932766 10 h-index 1199166 12 g-index

13 all docs

13 docs citations

13 times ranked 4163 citing authors

#	Article	lF	CITATIONS
1	Congenital hypopituitarism in two brothers with a duplication of the â€acrogigantism gene†GPR101: clinical findings and review of the literature. Pituitary, 2021, 24, 229-241.	1.6	2
2	The Genetics of Atypical Femur Fracturesâ€"a Systematic Review. Current Osteoporosis Reports, 2021, 19, 123-130.	1.5	15
3	A comparison of genotyping arrays. European Journal of Human Genetics, 2021, 29, 1611-1624.	1.4	43
4	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.	1.1	24
5	Linkage analysis and whole exome sequencing identify a novel candidate gene in a Dutch multiple sclerosis family. Multiple Sclerosis Journal, 2019, 25, 909-917.	1.4	19
6	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.	1.4	12
7	Reply: PRKAR1B mutations are a rare cause of FUS negative neuronal intermediate filament inclusion disease. Brain, 2015, 138, e358-e358.	3.7	O
8	Complex craniosynostosis is associated with the 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 244-253.	0.7	16
9	Boston type craniosynostosis: Report of a second mutation in <i>MSX2</i> . American Journal of Medical Genetics, Part A, 2013, 161, 2626-2633.	0.7	23
10	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.	1.1	53
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.	0.7	6
12	Mutation in the AP4M1 Gene Provides a Model for Neuroaxonal Injury in Cerebral Palsy. American Journal of Human Genetics, 2009, 85, 40-52.	2.6	156
13	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.	13.5	3,285