

Parneet Kaur

List of Publications by Year in descending order

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Version: 2024-02-01

18
papers

135
citations

1163117

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1372567

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docs citations

18
times ranked

222
citing authors

#	ARTICLE	IF	CITATIONS
1	Growth and neurodevelopmental disorder with arthrogryposis, microcephaly and structural brain anomalies caused by Bi-allelic partial deletion of SMPD4 gene. <i>Journal of Human Genetics</i> , 2022, 67, 133-136.	2.3	5
2	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures. <i>Human Genetics</i> , 2022, , 1.	3.8	0
3	Bi-allelic variants in <i>CHKA</i> cause a neurodevelopmental disorder with epilepsy and microcephaly. <i>Brain</i> , 2022, 145, 1916-1923.	7.6	3
4	Further evidence of muscle involvement in neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy. <i>Annals of Human Genetics</i> , 2022, 86, 94-101.	0.8	3
5	Homozygous variant p.(Arg163Trp) in PIGH causes glycosylphosphatidylinositol biosynthesis defect with epileptic encephalopathy and delayed myelination. <i>Clinical Dysmorphology</i> , 2022, Publish Ahead of Print, .	0.3	0
6	Genetic disorders with central nervous system white matter abnormalities: An update. <i>Clinical Genetics</i> , 2021, 99, 119-132.	2.0	7
7	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	6.2	17
8	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. <i>Clinical Genetics</i> , 2021, 100, 542-550.	2.0	12
9	Multilocus disease-causing genomic variations for Mendelian disorders: role of systematic phenotyping and implications on genetic counselling. <i>European Journal of Human Genetics</i> , 2021, 29, 1774-1780.	2.8	7
10	Bi-allelic missense variant, p.<sc>Ser35Leu</sc> in <sc><i>EXOSC1</i></sc> is associated with pontocerebellar hypoplasia. <i>Clinical Genetics</i> , 2021, 99, 594-600.	2.0	16
11	Recurrent bi-allelic splicing variant c.454+3A>G in TRAPPC4 is associated with progressive encephalopathy and muscle involvement. <i>Brain</i> , 2020, 143, e29.	7.6	8
12	Novel variant p.(Ala102Thr) in <i>SDHB</i> causes mitochondrial complex II deficiency: Case report and review of the literature. <i>Annals of Human Genetics</i> , 2020, 84, 345-351.	0.8	9
13	Spastic Paraplegia Type 56 in a Young Child. <i>Indian Journal of Pediatrics</i> , 2020, 87, 650-651.	0.8	3
14	Homozygous variant, p.(Arg643Trp) in VAC14 causes striatonigral degeneration: report of a novel variant and review of VAC14-related disorders. <i>Journal of Human Genetics</i> , 2019, 64, 1237-1242.	2.3	8
15	Confirmation of a Rare Genetic Leukoencephalopathy due to a Novel Bi-allelic Variant in RPIA. <i>European Journal of Medical Genetics</i> , 2019, 62, 103708.	1.3	10
16	Identification of a novel homozygous variant confirms <i>ITPA</i> as a developmental and epileptic encephalopathy gene. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 857-861.	1.2	14
17	GATAD2B-related intellectual disability due to parental mosaicism and review of literature. <i>Clinical Dysmorphology</i> , 2019, 28, 190-194.	0.3	5
18	Report of the Third Family with Multiple Mitochondrial Dysfunctions Syndrome 5 Caused by the Founder Variant p.(Glu87Lys) in ISCA1. <i>Journal of Pediatric Genetics</i> , 2018, 07, 130-133.	0.7	8