Parneet Kaur

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10669062/publications.pdf

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18	135	1163117 8	1372567 10
papers	citations	h-index	g-index

18	18	18	222
all docs	docs citations	times ranked	citing authors

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