Victoria Mok Siu

List of Publications by Year
in descending order

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7 papers	200 citations	1683934 5 h-index	1719901 7 g-index
7 all docs	7 docs citations	7 times ranked	451 citing authors

#	Article	IF	Citations
1	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	1.4	115
2	MECP2 Mutation Interrupts Nucleolinâ€“mTORâ€“P70S6K Signaling in Rett Syndrome Patients. Frontiers in Genetics, 2018, 9, 635.	1.1	37
3	Differential brain regionâ€specific expression of MeCP2 and BDNF in Rett Syndrome patients: a distinct greyâ€white matter variation. Neuropathology and Applied Neurobiology, 2020, 46, 735-750.	1.8	20
4	Homozygous/compound heterozygote RYR1 gene variants: Expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 386-396.	0.7	19
5	Transient hyponatremia of prematurity caused by mild Bartter syndrome type II: a case report. BMC Pediatrics, 2020, 20, 311.	0.7	5
6	A novel homozygous variant in REN in a family presenting with classic features of disorders involving the reninâ€“angiotensin pathway, without renal tubular dysgenesis. American Journal of Medical Genetics, Part A, 2020, 182, 2284-2290.	0.7	3
7	Potential Pitfalls in Pre-implantation Genetic Diagnosis in a Patient with Tuberous Sclerosis and Isolated Mosaicism for a <i>TSC2</i> Variant in Renal Tissue. Molecular Syndromology, 2021, 12, 154-158.	0.3	1