

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	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. <i>Molecular Syndromology</i> , 2021, 12, 154-158.	0.3	1
2	A novel homozygous variant in <i>REN</i> in a family presenting with classic features of disorders involving the renin-angiotensin pathway, without renal tubular dysgenesis. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2284-2290.	0.7	3
3	Transient hyponatremia of prematurity caused by mild Bartter syndrome type II: a case report. <i>BMC Pediatrics</i> , 2020, 20, 311.	0.7	5
4	Differential brain region-specific expression of <i>MeCP2</i> and <i>BDNF</i> in Rett Syndrome patients: a distinct grey-white matter variation. <i>Neuropathology and Applied Neurobiology</i> , 2020, 46, 735-750.	1.8	20
5	Homozygous/compound heterozygote <i>RYR1</i> gene variants: Expanding the clinical spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 386-396.	0.7	19
6	<i>MECP2</i> Mutation Interrupts Nucleolin-mTOR-P70S6K Signaling in Rett Syndrome Patients. <i>Frontiers in Genetics</i> , 2018, 9, 635.	1.1	37
7	Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. <i>European Journal of Human Genetics</i> , 2015, 23, 292-301.	1.4	115