Julien H Park

List of Publications by Year in Descending Order

21 papers	431 citations	9 h-index	20 g-index
23 ext. papers	574 ext. citations	5.6 avg, IF	2.87 L-index

#	Paper	IF	Citations
21	TMEM16A deficiency: a potentially fatal neonatal disease resulting from impaired chloride currents. *Journal of Medical Genetics*, **2021**, 58, 247-253	5.8	4
20	Mannose supplementation in PMM2-CDG. *Orphanet Journal of Rare Diseases*, **2021**, 16, 359	4.2	0
19	Treatment Options in Congenital Disorders of Glycosylation. *Frontiers in Genetics*, **2021**, 12, 735348	4.5	1
18	Reply: Not every excessive startle is hyperekplexia, the curious case of SOD1. *Brain*, **2020**, 143, e12	11.2	
17	Cystinosis: Therapy adherence and metabolic monitoring in patients treated with immediate-release cysteamine. *Molecular Genetics and Metabolism Reports*, **2020**, 24, 100620	1.8	2
16	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation. *Scientific Reports*, **2020**, 10, 13162	4.9	18
15	N-glycome analysis detects dysglycosylation missed by conventional methods in SLC39A8 deficiency. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1370-1381	5.4	4
14	L-Fucose treatment of FUT8-CDG. *Molecular Genetics and Metabolism Reports*, **2020**, 25, 100680	1.8	5
13	SOD1 deficiency: a novel syndrome distinct from amyotrophic lateral sclerosis. *Brain*, **2019**, 142, 2230-2237	11.2	33
12	Transferrin glycosylation analysis from dried blood spot cards and capillary blood samples. *Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences*, **2019**, 1106-1107, 64-70	3.2	4
11	SLC39A8 deficiency: biochemical correction and major clinical improvement by manganese therapy. *Genetics in Medicine*, **2018**, 20, 259-268	8.1	47
10	Limitations of galactose therapy in phosphoglucomutase 1 deficiency. *Molecular Genetics and Metabolism Reports*, **2017**, 13, 33-40	1.8	23
9	Mutations in the X-linked cause a glycosylation disorder with autophagic defects. *Journal of Experimental Medicine*, **2017**, 214, 3707-3729	16.6	43
8	TMEM165 Deficiency: Postnatal Changes in Glycosylation. *JIMD Reports*, **2016**, 26, 21-9	1.9	19
7	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Answers. *Pediatric Nephrology*, **2016**, 31, 1283-6	3.2	0
6	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Questions. *Pediatric Nephrology*, **2016**, 31, 1281	3.2	0
5	It Is Not Always Alcohol Abuse--A Transferrin Variant Impairing the CDT Test. *Alcohol and Alcoholism*, **2016**, 51, 148-53	3.5	9

List of Publications

4	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. *American Journal of Human Genetics*, **2015**, 97, 894-903	11	180
3	Transferrin variants: pitfalls in the diagnostics of Congenital disorders of glycosylation. *Clinical Biochemistry*, **2015**, 48, 11-3	3.5	23
2	The novel transferrin E592A variant impairs the diagnostics of congenital disorders of glycosylation. *Clinica Chimica Acta*, **2014**, 436, 135-9	6.2	9
1	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation		6