Julien H Park

List of Publications by Year
in descending order

21	654	933410 10	752679 20
papers	citations	h-index	g-index

23	23	23	1211
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. American Journal of Human Genetics, 2015, 97, 894-903.	6.2	242
2	Mutations in the X-linked ATP6AP2 cause a glycosylation disorder with autophagic defects. Journal of Experimental Medicine, 2017, 214, 3707-3729.	8.5	62
3	SLC39A8 deficiency: biochemical correction and major clinical improvement by manganese therapy. Genetics in Medicine, 2018, 20, 259-268.	2.4	62
4	SOD1 deficiency: a novel syndrome distinct from amyotrophic lateral sclerosis. Brain, 2019, 142, 2230-2237.	7.6	59
5	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation. Scientific Reports, 2020, 10, 13162.	3.3	43
6	Limitations of galactose therapy in phosphoglucomutase 1 deficiency. Molecular Genetics and Metabolism Reports, 2017, 13, 33-40.	1.1	34
7	Transferrin variants: Pitfalls in the diagnostics of Congenital disorders of glycosylation. Clinical Biochemistry, 2015, 48, 11-13.	1.9	30
8	TMEM165 Deficiency: Postnatal Changes in Glycosylation. JIMD Reports, 2015, 26, 21-29.	1.5	24
9	Treatment Options in Congenital Disorders of Glycosylation. Frontiers in Genetics, 2021, 12, 735348.	2.3	13
10	It Is Not Always Alcohol Abuseâ€”A Transferrin Variant Impairing the CDT Test. Alcohol and Alcoholism, 2016, 51, 148-153.	1.6	12
11	L-Fucose treatment of FUT8-CDG. Molecular Genetics and Metabolism Reports, 2020, 25, 100680.	1.1	11
12	The novel transferrin E592A variant impairs the diagnostics of congenital disorders of glycosylation. Clinica Chimica Acta, 2014, 436, 135-139.	1.1	10
13	TMEM16A deficiency: a potentially fatal neonatal disease resulting from impaired chloride currents. Journal of Medical Genetics, 2021, 58, 247-253.	3.2	10
14	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
15	N â€glycome analysis detects dysglycosylation missed by conventional methods in SLC39A8 deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 1370-1381.	3.6	8
16	Cystinosis: Therapy adherence and metabolic monitoring in patients treated with immediate-release cysteamine. Molecular Genetics and Metabolism Reports, 2020, 24, 100620.	1.1	7
17	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.	2.3	6
18	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Answers. Pediatric Nephrology, 2016, 31, 1283-1286.	1.7	2

#	Article	IF	Citations
19	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Questions. Pediatric Nephrology, 2016, 31, 1281-1281.	1.7	1
20	Mannose supplementation in PMM2-CDG. Orphanet Journal of Rare Diseases, 2021, 16, 359.	2.7	1
21	Reply: Not every excessive startle is hyperekplexia, the curious case of SOD1. Brain, 2020, 143, e12-e12.	7.6	0