

# Julien H Park

## List of Publications by Citations

**Source:** <https://exaly.com/author-pdf/8285077/julien-h-park-publications-by-citations.pdf>

**Version:** 2024-04-28

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 894-903	11	180
20	SLC39A8 deficiency: biochemical correction and major clinical improvement by manganese therapy. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 259-268	8.1	47
19	Mutations in the X-linked cause a glycosylation disorder with autophagic defects. <i>Journal of Experimental Medicine</i> , <b>2017</b> , 214, 3707-3729	16.6	43
18	SOD1 deficiency: a novel syndrome distinct from amyotrophic lateral sclerosis. <i>Brain</i> , <b>2019</b> , 142, 2230-2237	17.2	33
17	Limitations of galactose therapy in phosphoglucomutase 1 deficiency. <i>Molecular Genetics and Metabolism Reports</i> , <b>2017</b> , 13, 33-40	1.8	23
16	Transferrin variants: pitfalls in the diagnostics of Congenital disorders of glycosylation. <i>Clinical Biochemistry</i> , <b>2015</b> , 48, 11-3	3.5	23
15	TMEM165 Deficiency: Postnatal Changes in Glycosylation. <i>JIMD Reports</i> , <b>2016</b> , 26, 21-9	1.9	19
14	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation. <i>Scientific Reports</i> , <b>2020</b> , 10, 13162	4.9	18
13	The novel transferrin E592A variant impairs the diagnostics of congenital disorders of glycosylation. <i>Clinica Chimica Acta</i> , <b>2014</b> , 436, 135-9	6.2	9
12	It Is Not Always Alcohol Abuse--A Transferrin Variant Impairing the CDT Test. <i>Alcohol and Alcoholism</i> , <b>2016</b> , 51, 148-53	3.5	9
11	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation		6
10	L-Fucose treatment of FUT8-CDG. <i>Molecular Genetics and Metabolism Reports</i> , <b>2020</b> , 25, 100680	1.8	5
9	N-glycome analysis detects dysglycosylation missed by conventional methods in SLC39A8 deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2020</b> , 43, 1370-1381	5.4	4
8	Transferrin glycosylation analysis from dried blood spot cards and capillary blood samples. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , <b>2019</b> , 1106-1107, 64-70	3.2	4
7	TMEM16A deficiency: a potentially fatal neonatal disease resulting from impaired chloride currents. <i>Journal of Medical Genetics</i> , <b>2021</b> , 58, 247-253	5.8	4
6	Cystinosis: Therapy adherence and metabolic monitoring in patients treated with immediate-release cysteamine. <i>Molecular Genetics and Metabolism Reports</i> , <b>2020</b> , 24, 100620	1.8	2
5	Treatment Options in Congenital Disorders of Glycosylation. <i>Frontiers in Genetics</i> , <b>2021</b> , 12, 735348	4.5	1

4	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Answers. <i>Pediatric Nephrology</i> , <b>2016</b> , 31, 1283-6	3.2	○
3	Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Questions. <i>Pediatric Nephrology</i> , <b>2016</b> , 31, 1281	3.2	○
2	Mannose supplementation in PMM2-CDG. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 359	4.2	○
1	Reply: Not every excessive startle is hyperekplexia, the curious case of SOD1. <i>Brain</i> , <b>2020</b> , 143, e12	11.2	