Stephanie Grunewald

List of Publications by Year
in descending order

32	1,860	361413 20	377865 34
papers	citations	h-index	g-index

35	35	35	2187
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Synergistic use of glycomics and singleâ€molecule molecular inversion probes for <scp>identification</scp> of congenital disorders of glycosylation typeâ€1. Journal of Inherited Metabolic Disease, 2022, 45, 769-781.	3.6	7
2	International consensus guidelines for phosphoglucomutase 1 deficiency (<scp>PGM1â€CDG</scp>): Diagnosis, followâ€up, and management. Journal of Inherited Metabolic Disease, 2021, 44, 148-163.	3.6	27
3	Effects of triheptanoin (<scp>UX007</scp>) in patients with longâ€chain fatty acid oxidation disorders: Results from an <scp>openâ€label</scp>, <scp>longâ€term</scp> extension study. Journal of Inherited Metabolic Disease, 2021, 44, 253-263.	3.6	36
4	Congenital Disorders of Glycosylation from a Neurological Perspective. Brain Sciences, 2021, 11, 88.	2.3	53
5	Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: FirstÂrevision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	3.6	118
6	New insights into carnitineâ€acylcarnitine translocase deficiency from 23 cases: Management challenges and potential therapeutic approaches. Journal of Inherited Metabolic Disease, 2021, 44, 903-915.	3.6	8
7	Efficacy and safety of arimoclomol in <scp>Niemannâ€Pick</scp> disease type C: Results from a doubleâ€blind, randomised, placeboâ€controlled, multinational phase 2/3 trial of a novel treatment. Journal of Inherited Metabolic Disease, 2021, 44, 1463-1480.	3.6	26
8	26â€…Clinical trials and tribulations: the ordeals of setting up a high intensity clinical trial during COVID19 times. , 2021, , .		0
9	Inborn errors of metabolism leading to neuronal migration defects. Journal of Inherited Metabolic Disease, 2020, 43, 145-155.	3.6	16
10	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	3.6	24
11	Clinical disease progression and biomarkers in Niemannâ€“Pick disease type C: a prospective cohort study. Orphanet Journal of Rare Diseases, 2020, 15, 328.	2.7	12
12	Criss-cross gait. Neurology, 2020, 95, 500-501.	1.1	5
13	An expanding spectrum of complications in isolated methylmalonic aciduria. Medycyna Wieku Rozwojowego, 2020, 24, 9-13.	0.2	1
14	Liver neoplasms in methylmalonic aciduria: An emerging complication. Journal of Inherited Metabolic Disease, 2019, 42, 793-802.	3.6	25
15	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	3.6	91
16	Movement disorders and nonmotor neuropsychological symptoms in children and adults with classical galactosemia. Journal of Inherited Metabolic Disease, 2019, 42, 451-458.	3.6	27
17	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	3.6	53
18	Phase I/II Trial of Liverâ€“derived Mesenchymal Stem Cells in Pediatric Liverâ€“based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liverâ€“derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. Transplantation, 2019, 103, 1903-1915.	1.0	47

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19	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	3.6	32
20	Liver transplantation for neonatalâ€onset citrullinemia. Pediatric Transplantation, 2018, 22, e13191.	1.0	10
21	FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia. JCI Insight, 2018, 3, .	5.0	50
22	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
23	Systematic Review and Meta-analysis of Intelligence Quotient in Early-Treated Individuals with Classical Galactosemia. JIMD Reports, 2017, 37, 115-123.	1.5	15
24	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	2.5	43
25	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
26	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and followâ€up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176.	3.6	132
27	Global serum glycoform profiling for the investigation of dystroglycanopathies & Congenital Disorders of Glycosylation. Molecular Genetics and Metabolism Reports, 2016, 7, 55-62.	1.1	8
28	Proteomic Discovery and Development of a Multiplexed Targeted MRM-LC-MS/MS Assay for Urine Biomarkers of Extracellular Matrix Disruption in Mucopolysaccharidoses I, II, and VI. Analytical Chemistry, 2015, 87, 12238-12244.	6.5	20
29	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	3.6	186
30	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	3.6	175
31	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
32	Congenital disorders of glycosylation: Rapidly enlarging group of (neuro)metabolic disorders. Early Human Development, 2007, 83, 825-830.	1.8	41