Mirian Janssen

List of Publications by Year
in descending order

21 papers	551 citations	840585 11 h-index	713332 21 g-index
22 all docs	22 docs citations	22 times ranked	849 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.	1.7	7
2	Urine-Derived Kidney Progenitor Cells in Cystinosis. Cells, 2022, 11, 1245.	1.8	2
3	High childhood serum triglyceride concentrations associate with hepatocellular adenoma development in patients with glycogen storage disease type Ia. JHEP Reports, 2022, 4, 100512.	2.6	7
4	High protein prescription in methylmalonic and propionic acidemia patients and its negative association with long-term outcome. Clinical Nutrition, 2021, 40, 3622-3630.	2.3	9
5	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	1.4	7
6	An international cohort study spanning five decades assessed outcomes of nephropathic cystinosis. Kidney International, 2021, 100, 1112-1123.	2.6	31
7	Six-year prospective follow-up study in 151 carriers of the mitochondrial DNA 3243 A>G variant. Journal of Medical Genetics, 2021, 58, 48-55.	1.5	17
8	Mitochondrial disease and COVID-19: An international cohort study confirms risks and long-term outcomes. Journal of the Neurological Sciences, 2021, 429, 119358.	0.3	0
9	Mitochondrial migraine; a prevalence, impact and treatment efficacy cohort study. Mitochondrion, 2020, 53, 128-132.	1.6	15
10	The KHENERGY Study: Safety and Efficacy of KH 176 in Mitochondrial m.3243A>G Spectrum Disorders. Clinical Pharmacology and Therapeutics, 2019, 105, 101-111.	2.3	41
11	Enhanced Intrinsic Skin Aging in Nephropathic Cystinosis Assessed by High-Definition Optical Coherence Tomography. Journal of Investigative Dermatology, 2019, 139, 2242-2245.e5.	0.3	5
12	Swallowing dysfunction in patients with nephropathic cystinosis. Molecular Genetics and Metabolism, 2019, 126, 413-415.	0.5	8
13	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. Translational Research, 2018, 199, 62-76.	2.2	22
14	Hyperammonemia due to Adult-Onset N-Acetylglutamate Synthase Deficiency. JIMD Reports, 2016, 31, 95-99.	0.7	13
15	Quality of life, fatigue and mental health in patients with the m.3243Aâ€‰>â€‰G mutation and its correlates with genetic characteristics and disease manifestation. Orphanet Journal of Rare Diseases, 2016, 11, 25.	1.2	14
16	Successful Liver Transplantation and Long-Term Follow-up in a Patient With MPI-CDG. Pediatrics, 2014, 134, e279-e283.	1.0	48
17	Screening of a healthy newborn identifies three adult family members with symptomatic glutaric aciduria type I. BBA Clinical, 2014, 1, 30-32.	4.1	4
18	Biochemical monitoring of pregnancy and breast feeding in five patients with classical galactosaemia â€“ and review of the literature. European Journal of Pediatrics, 2009, 168, 721-729.	1.3	24

#	Article	IF	Citations
19	Therapeutic Erythrocytapheresis (TE) versus Phlebotomy (P) in the treatment of Hereditary Hemochromatosis (HH) patients: Preliminary results from an ongoing randomized clinical trial (NCT) Tj ETQq1 1 0.784314 rgBT /Over		
20	Retinal vein occlusion: A form of venous thrombosis or a complication of atherosclerosis?. Thrombosis and Haemostasis, 2005, 93, 1021-1026.	1.8	222
21	Rapid D-Dimer Assays to Exclude Deep Venous Thrombosis and Pulmonary Embolism: Current Status and New Developments. Seminars in Thrombosis and Hemostasis, 1998, 24, 393-400.	1.5	43