Monique van Scherpenzeel

List of Publications by Year
in descending order

35 papers	1,771 citations	304743 22 h-index	377865 34 g-index
36 all docs	36 docs citations	36 times ranked	2820 citing authors

#	Article	IF	Citations
1	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236
2	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	6.2	197
3	Galectinâ€3â€Binding Glycomimetics that Strongly Reduce Bleomycinâ€Induced Lung Fibrosis and Modulate Intracellular Glycan Recognition. ChemBioChem, 2016, 17, 1759-1770.	2.6	145
4	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
5	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
6	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
7	High-resolution mass spectrometry glycoprofiling of intact transferrin for diagnosis and subtype identification in the congenital disorders of glycosylation. Translational Research, 2015, 166, 639-649.e1.	5.0	73
8	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. American Journal of Human Genetics, 2016, 98, 322-330.	6.2	73
9	Human ISPD Is a Cytidyltransferase Required for Dystroglycan O-Mannosylation. Chemistry and Biology, 2015, 22, 1643-1652.	6.0	67
10	Clinical diagnostics and therapy monitoring in the congenital disorders of glycosylation. Glycoconjugate Journal, 2016, 33, 345-358.	2.7	56
11	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	2.4	55
12	A new chemical probe for the detection of the cancer-linked galectin-3. Organic and Biomolecular Chemistry, 2006, 4, 4387.	2.8	52
13	Successful Liver Transplantation and Long-Term Follow-up in a Patient With MPI-CDG. Pediatrics, 2014, 134, e279-e283.	2.1	48
14	Clinical glycomics for the diagnosis of congenital disorders of glycosylation. Journal of Inherited Metabolic Disease, 2018, 41, 499-513.	3.6	44
15	Unraveling the unknown areas of the human metabolome: the role of infrared ion spectroscopy. Journal of Inherited Metabolic Disease, 2018, 41, 367-377.	3.6	44
16	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
17	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	2.9	37
18	Synthesis and Evaluation of New Thiodigalactosideâ€Based Chemical Probes to Label Galectinâ€3. ChemBioChem, 2009, 10, 1724-1733.	2.6	36

#	Article	IF	Citations
19	Sialic acid catabolism by N-acetylneuraminate pyruvate lyase is essential for muscle function. JCI Insight, 2018, 3, .	5.0	36
20	A common sugarâ€nucleotideâ€mediated mechanism of inhibition of (glycosamino)glycan biosynthesis, as evidenced by 6Fâ€GalNAc (Ac ₃). FASEB Journal, 2015, 29, 2993-3002.	0.5	31
21	A compound heterozygous mutation in DPAGT1 results in a congenital disorder of glycosylation with a relatively mild phenotype. European Journal of Human Genetics, 2013, 21, 844-849.	2.8	25
22	Nanomolar affinity, iminosugar-based chemical probes for specific labeling of lysosomal glucocerebrosidase. Bioorganic and Medicinal Chemistry, 2010, 18, 267-273.	3.0	24
23	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. Translational Research, 2018, 199, 62-76.	5.0	22
24	Fast, robust and high-resolution glycosylation profiling of intact monoclonal IgG antibodies using nanoLC-chip-QTOF. Clinica Chimica Acta, 2016, 461, 90-97.	1.1	20
25	Combined sialic acid and histone deacetylase (HDAC) inhibitor treatment up-regulates the neuroblastoma antigen GD2. Journal of Biological Chemistry, 2019, 294, 4437-4449.	3.4	20
26	Detection of galectin-3 by novel peptidic photoprobes. Bioorganic and Medicinal Chemistry Letters, 2007, 17, 376-378.	2.2	19
27	Activity of N-acylneuraminate-9-phosphatase (NANP) is not essential for de novo sialic acid biosynthesis. Biochimica Et Biophysica Acta - General Subjects, 2019, 1863, 1471-1479.	2.4	18
28	Dynamic tracing of sugar metabolism reveals the mechanisms of action of synthetic sugar analogs. Glycobiology, 2022, 32, 239-250.	2.5	15
29	Phosphoglucomutase-1 deficiency: Early presentation, metabolic management and detection in neonatal blood spots. Molecular Genetics and Metabolism, 2020, 131, 135-146.	1.1	14
30	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306.	3.2	11
31	Protein enrichment by captureâ€“release based on strain-promoted cycloaddition of azide with bicyclononyne (BCN). Bioorganic and Medicinal Chemistry, 2012, 20, 655-661.	3.0	8
32	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
33	Screening for abnormal glycosylation in a cohort of adult liver disease patients. Journal of Inherited Metabolic Disease, 2020, 43, 1310-1320.	3.6	6
34	Structureâ€“Activity Relationship of Fluorinated Sialic Acid Inhibitors for Bacterial Sialylation. Bioconjugate Chemistry, 2021, 32, 1047-1051.	3.6	5
35	NAFLD Phenotype in Patients With V-ATPase Proton Pump Assembly Defects. Cellular and Molecular Gastroenterology and Hepatology, 2018, 5, 415-417.e1.	4.5	0