

# Nathalie S Seta

## List of Publications by Year in descending order

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148  
papers

5,891  
citations

71102

41  
h-index

95266

68  
g-index

164  
all docs

164  
docs citations

164  
times ranked

6032  
citing authors

#	ARTICLE	IF	CITATIONS
1	Effects of mechanical ventilation on diaphragmatic contractile properties in rats.. American Journal of Respiratory and Critical Care Medicine, 1994, 149, 1539-1544.	5.6	254
2	Compartmentalized cytokine production within the human lung in unilateral pneumonia.. American Journal of Respiratory and Critical Care Medicine, 1994, 150, 710-716.	5.6	233
3	Protein Glycosylation and Diseases: Blood and Urinary Oligosaccharides as Markers for Diagnosis and Therapeutic Monitoring. Clinical Chemistry, 2000, 46, 795-805.	3.2	218
4	A broad spectrum of clinical presentations in congenital disorders of glycosylation I: a series of 26 cases. Journal of Medical Genetics, 2001, 38, 14-19.	3.2	204
5	Congenital disorders of glycosylation (CDG): Quo vadis?. European Journal of Medical Genetics, 2018, 61, 643-663.	1.3	191
6	Compartmentalized IL-8 and elastase release within the human lung in unilateral pneumonia.. American Journal of Respiratory and Critical Care Medicine, 1996, 153, 336-342.	5.6	151
7	Cobblestone lissencephaly: neuropathological subtypes and correlations with genes of dystroglycanopathies. Brain, 2012, 135, 469-482.	7.6	151
8	Mutations in PMM2 that cause congenital disorders of glycosylation, type Ia (CDG-Ia). Human Mutation, 2000, 16, 386-394.	2.5	136
9	Hyperinsulinemic hypoglycemia as a presenting sign in phosphomannose isomerase deficiency: A new manifestation of carbohydrate-deficient glycoprotein syndrome treatable with mannose. Journal of Pediatrics, 1999, 135, 379-383.	1.8	127
10	Identification of Mutations in TMEM5 and ISPD as a Cause of Severe Cobblestone Lissencephaly. American Journal of Human Genetics, 2012, 91, 1135-1143.	6.2	126
11	Indoor aldehydes: measurement of contamination levels and identification of their determinants in Paris dwellings. Environmental Research, 2003, 92, 245-253.	7.5	125
12	ISPD produces CDP-ribitol used by FKTN and FKRK to transfer ribitol phosphate onto Î±-dystroglycan. Nature Communications, 2016, 7, 11534.	12.8	113
13	The clinical spectrum of phosphomannose isomerase deficiency, with an evaluation of mannose treatment for CDG-Ib. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 841-843.	3.8	103
14	Carbohydrate-deficient glycoprotein syndromes become congenital disorders of glycosylation: an updated nomenclature for CDG. First International Workshop on CDGS. Glycoconjugate Journal, 1999, 16, 669-671.	2.7	93
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	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. Journal of Medical Genetics, 2017, 54, 843-851.	3.2	88
17	Congenital Disorders of Glycosylation Type Ig Is Defined by a Deficiency in Dolichyl-P-mannose:Man7GlcNAc2-PP-dolichyl Mannosyltransferase. Journal of Biological Chemistry, 2002, 277, 25815-25822.	3.4	87
18	Nasal inflammation and personal exposure to fine particles PM2.5 in asthmatic children. Journal of Allergy and Clinical Immunology, 2006, 117, 1382-1388.	2.9	83

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19	Protein glycosylation and diseases: blood and urinary oligosaccharides as markers for diagnosis and therapeutic monitoring. <i>Clinical Chemistry</i> , 2000, 46, 795-805.	3.2	82
20	A Deficiency in Dolichyl-P-glucose:Glc1Man9GlcNAc2-PP-dolichyl $\beta$ -3-Glucosyltransferase Defines a New Subtype of Congenital Disorders of Glycosylation. <i>Journal of Biological Chemistry</i> , 2003, 278, 9962-9971.	3.4	78
21	Pesticide exposure of non-occupationally exposed subjects compared to some occupational exposure: A French pilot study. <i>Science of the Total Environment</i> , 2006, 366, 74-91.	8.0	78
22	Secretion of $\alpha$ -antitrypsin by alveolar epithelial cells. <i>FEBS Letters</i> , 1994, 346, 171-174.	2.8	76
23	New <i>POMT2</i> mutations causing congenital muscular dystrophy. <i>Neurology</i> , 2007, 69, 1254-1260.	1.1	62
24	Diagnostic value of Western blotting in carbohydrate-deficient glycoprotein syndrome. <i>Clinica Chimica Acta</i> , 1996, 254, 131-140.	1.1	59
25	Risk assessment of acute vascular events in congenital disorder of glycosylation type Ia. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 444-449.	1.1	59
26	Molecular heterogeneity in fetal forms of type II lissencephaly. <i>Human Mutation</i> , 2007, 28, 1020-1027.	2.5	58
27	Assessment and predictor determination of indoor aldehyde levels in Paris newborn babies' homes. <i>Indoor Air</i> , 2009, 19, 314-323.	4.3	58
28	Oncostatin M Is a Potent Stimulator of $\alpha$ -1-Antitrypsin Secretion in Lung Epithelial Cells: Modulation by Transforming Growth Factor- $\beta$ 2 and Interferon- $\gamma$ 3. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 1998, 18, 511-520.	2.9	54
29	Insecticide Urinary Metabolites in Nonoccupationally Exposed Populations. <i>Journal of Toxicology and Environmental Health - Part B: Critical Reviews</i> , 2005, 8, 485-512.	6.5	54
30	29 French adult patients with PMM2-congenital disorder of glycosylation: outcome of the classical pediatric phenotype and depiction of a late-onset phenotype. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 207.	2.7	52
31	From splitting GLUT1 deficiency syndromes to overlapping phenotypes. <i>European Journal of Medical Genetics</i> , 2015, 58, 443-454.	1.3	52
32	An in vitro model to evaluate the inflammatory response after gaseous formaldehyde exposure of lung epithelial cells. <i>Toxicology Letters</i> , 2010, 195, 99-105.	0.8	51
33	A case of fatal Type I congenital disorders of glycosylation (CDG I) associated with low dehydrololichol diphosphate synthase (DHDDS) activity. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 84.	2.7	50
34	Neurological Presentation in Pediatric Patients with Congenital Disorders of Glycosylation Type Ia. <i>Neuropediatrics</i> , 2003, 34, 1-6.	0.6	49
35	Alterations in relative proportions of microheterogenous forms of human $\alpha$ -1-acid glycoprotein in liver disease. <i>Journal of Hepatology</i> , 1986, 2, 245-252.	3.7	47
36	Interleukin 6 secretion by monocytes and alveolar macrophages in systemic sclerosis with lung involvement.. <i>American Journal of Respiratory and Critical Care Medicine</i> , 1994, 149, 1260-1265.	5.6	47

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37	Characterization of the 415G>A (E139K) PMM2 mutation in carbohydrate-deficient glycoprotein syndrome type Ia disrupting a splicing enhancer resulting in exon 5 skipping. <i>Human Mutation</i> , 1999, 14, 543-544.	2.5	47
38	A rapid mass spectrometric strategy for the characterization of N- and O-glycan chains in the diagnosis of defects in glycan biosynthesis. <i>Proteomics</i> , 2007, 7, 1800-1813.	2.2	47
39	Early polysensitization is associated with allergic multimorbidity in PARIS birth cohort infants. <i>Pediatric Allergy and Immunology</i> , 2016, 27, 831-837.	2.6	46
40	Development of liver disease despite mannose treatment in two patients with CDG-Ib. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 40-43.	1.1	44
41	Urinary arsenic concentrations and speciation in residents living in an area with naturally contaminated soils. <i>Science of the Total Environment</i> , 2010, 408, 1190-1194.	8.0	43
42	Neurological presentation of a congenital disorder of glycosylation CDG-Ia: Implications for diagnosis and genetic counseling. <i>American Journal of Medical Genetics Part A</i> , 2001, 101, 46-49.	2.4	40
43	IL6 and acute phase plasma proteins in peritoneal fluid of women with endometriosis. <i>Clinica Chimica Acta</i> , 1992, 210, 187-195.	1.1	39
44	Guanosine diphosphate-mannose:GlcNAc2-PP-dolichol mannosyltransferase deficiency (congenital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 <i>Genetics</i> , 2010, 47, 729-735.	3.2	39
45	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. <i>Nature Communications</i> , 2018, 9, 3087.	12.8	39
46	Inflammatory response modulation of airway epithelial cells exposed to formaldehyde. <i>Toxicology Letters</i> , 2012, 211, 159-163.	0.8	38
47	A new insight into PMM2 mutations in the French population. <i>Human Mutation</i> , 2005, 25, 504-505.	2.5	37
48	Asialoglycoprotein receptor in human isolated hepatocytes from normal liver and its apparent increase in liver with histological alterations. <i>Journal of Hepatology</i> , 1991, 13, 305-309.	3.7	36
49	Four Caucasian patients with mutations in the fukutin gene and variable clinical phenotype. <i>Neuromuscular Disorders</i> , 2009, 19, 182-188.	0.6	36
50	ALG6â€CDG: a recognizable phenotype with epilepsy, proximal muscle weakness, ataxia and behavioral and limb anomalies. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 713-723.	3.6	36
51	Long-term follow-up in PMM2-CDG: are we ready to start treatment trials?. <i>Genetics in Medicine</i> , 2019, 21, 1181-1188.	2.4	36
52	Endothelin-1 Secretion by Alveolar Macrophages in Systemic Sclerosis. <i>American Journal of Respiratory and Critical Care Medicine</i> , 1997, 156, 1429-1435.	5.6	35
53	Modifications of Concanavalin A patterns of Î±1-acid glycoprotein and Î±2-HS glycoprotein in alcoholic liver disease. <i>Clinica Chimica Acta</i> , 1988, 176, 49-57.	1.1	34
54	Microheterogeneity of the carbohydrate moiety of human alpha 1-acid glycoprotein in two benign liver diseases: Alcoholic cirrhosis and acute hepatitis. <i>Clinica Chimica Acta</i> , 1989, 186, 59-66.	1.1	34

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55	Interleukin-8 and neutrophils in systemic sclerosis with lung involvement.. American Journal of Respiratory and Critical Care Medicine, 1994, 150, 1363-1367.	5.6	34
56	Environmental and biological monitoring of exposure to organophosphorus pesticides: Application to occupationally and non-occupationally exposed adult populations. Journal of Exposure Science and Environmental Epidemiology, 2006, 16, 417-426.	3.9	34
57	Protein O-mannosyltransferase activities in lymphoblasts from patients with $\hat{\pm}$ -dystroglycanopathies. Neuromuscular Disorders, 2008, 18, 45-51.	0.6	33
58	Cardiomyopathy in the congenital disorders of glycosylation (CDG): a case of late presentation and literature review. Journal of Inherited Metabolic Disease, 2009, 32, 313-319.	3.6	32
59	Should PMM2-deficiency (CDG Ia) be searched in every case of unexplained hydrops fetalis?. Molecular Genetics and Metabolism, 2010, 101, 253-257.	1.1	32
60	Dystroglycanopathies: About Numerous Genes Involved in Glycosylation of One Single Glycoprotein. Journal of Neuromuscular Diseases, 2015, 2, 27-38.	2.6	32
61	Congenital Disorder of Glycosylation Ia with Deficient Phosphomannomutase Activity but Normal Plasma Glycoprotein Pattern. Clinical Chemistry, 2001, 47, 132-134.	3.2	31
62	A New Intronic Mutation in the DPM1 Gene Is Associated With a Milder Form of CDG Ie in Two French Siblings. Pediatric Research, 2006, 59, 835-839.	2.3	31
63	Allergic sensitisation in early childhood: Patterns and related factors in PARIS birth cohort. International Journal of Hygiene and Environmental Health, 2016, 219, 792-800.	4.3	31
64	Manganese Superoxide Dismutase (SOD2) Polymorphisms, Plasma Advanced Oxidation Protein Products (AOPP) Concentration and Risk of Kidney Complications in Subjects with Type 1 Diabetes. PLoS ONE, 2014, 9, e96916.	2.5	31
65	Congenital disorders of glycosylation IIa cause growth retardation, mental retardation, and facial dysmorphism. Journal of Medical Genetics, 2000, 37, 875-877.	3.2	30
66	In vitro model adapted to the study of skin ageing induced by air pollution. Toxicology Letters, 2016, 259, 60-68.	0.8	30
67	Asthma and allergic rhinitis risk depends on house dust mite specific IgE levels in PARIS birth cohort children. World Allergy Organization Journal, 2019, 12, 100057.	3.5	30
68	A Genome-Wide CRISPR-Cas9 Screen Identifies the Dolichol-Phosphate Mannose Synthase Complex as a Host Dependency Factor for Dengue Virus Infection. Journal of Virology, 2020, 94, .	3.4	30
69	Complementarity of electrophoretic, mass spectrometric, and gene sequencing techniques for the diagnosis and characterization of congenital disorders of glycosylation. Electrophoresis, 2018, 39, 3123-3132.	2.4	29
70	Underdiagnosis of mild congenital disorders of glycosylation type Ia. Pediatric Neurology, 2005, 32, 121-123.	2.1	28
71	Congenital disorders of glycosylation type I: a rare but new cause of hyperechoic kidneys in infants and children due to early microcystic changes. Pediatric Radiology, 2006, 36, 108-114.	2.0	28
72	Indoor airborne endotoxin assessment in homes of Paris newborn babies. Indoor Air, 2008, 18, 480-487.	4.3	28

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73	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 349-352.	2.8	27
74	Tolerance of once-daily dosing of netilmicin and teicoplanin, alone or in combination, in healthy volunteers. <i>Clinical Pharmacology and Therapeutics</i> , 1988, 44, 458-466.	4.7	26
75	Defect in N-glycosylation of proteins is tissue-dependent in Congenital Disorders of Glycosylation Ia. <i>Glycobiology</i> , 2000, 10, 1277-1281.	2.5	26
76	An in vitro model to evaluate the impact of environmental fine particles (PM0.3-2.5) on skin damage. <i>Toxicology Letters</i> , 2019, 305, 94-102.	0.8	25
77	Limited protection by small unilamellar liposomes against the renal tubular toxicity induced by repeated amphotericin B infusions in rats. <i>Antimicrobial Agents and Chemotherapy</i> , 1991, 35, 1303-1308.	3.2	24
78	Identification of four novel PMM2 mutations in congenital disorders of glycosylation (CDG) Ia French patients. <i>Journal of Medical Genetics</i> , 2000, 37, 579-580.	3.2	24
79	Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib. <i>Journal of Medical Genetics</i> , 2002, 39, 849-851.	3.2	24
80	Detection of an Alu insertion in the POMT1 gene from three French Walker Warburg syndrome families. <i>Molecular Genetics and Metabolism</i> , 2007, 90, 93-96.	1.1	24
81	Conotruncal heart defects in three patients with congenital disorder of glycosylation type Ia (CDG) Tj ETQq1 1 0.784314 rgBT / Overlo	3.2	24
82	POMT2 intragenic deletions and splicing abnormalities causing congenital muscular dystrophy with mental retardation. <i>European Journal of Medical Genetics</i> , 2009, 52, 201-206.	1.3	24
83	Nasal Epithelial and Inflammatory Response to Ozone Exposure: A Review of Laboratory-Based Studies Published Since 1985. <i>Journal of Toxicology and Environmental Health - Part B: Critical Reviews</i> , 2003, 6, 521-568.	6.5	23
84	Nasal lavage as a tool for the assessment of upper-airway inflammation in adults and children. <i>Translational Research</i> , 2002, 139, 173-180.	2.3	22
85	Sequential air-liquid exposure of human respiratory cells to chemical and biological pollutants. <i>Toxicology Letters</i> , 2011, 207, 53-59.	0.8	22
86	Changes in $\alpha$ 1-acid glycoprotein serum concentrations and glycoforms in the developing human fetus. <i>Clinica Chimica Acta</i> , 1991, 203, 167-175.	1.1	21
87	Increased recurrence risk in congenital disorders of glycosylation type Ia (CDG-Ia) due to a transmission ratio distortion. <i>Journal of Medical Genetics</i> , 2004, 41, 877-880.	3.2	21
88	A model of human nasal epithelial cells adapted for direct and repeated exposure to airborne pollutants. <i>Toxicology Letters</i> , 2014, 229, 144-149.	0.8	21
89	MALDI-TOF MS applied to apoB glycoforms of patients with congenital disorders affecting O-glycosylation. Comparison with two-dimensional electrophoresis. <i>Proteomics - Clinical Applications</i> , 2015, 9, 787-793.	1.6	20
90	Impact of Mycotoxins Secreted by Aspergillus Molds on the Inflammatory Response of Human Corneal Epithelial Cells. <i>Toxins</i> , 2017, 9, 197.	3.4	20

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91	CCDC115-CDG: A new rare and misleading inherited cause of liver disease. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 228-235.	1.1	20
92	Dystroglycanopathies: About Numerous Genes Involved in Glycosylation of One Single Glycoprotein. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 27-38.	2.6	19
93	Elevated thrombin generation in patients with congenital disorder of glycosylation and combined coagulation factor deficiencies. <i>Journal of Thrombosis and Haemostasis</i> , 2019, 17, 1798-1807.	3.8	18
94	Incorporation of amphotericin B (AMB) into liposomes alters AMB-induced acute nephrotoxicity in rabbits. <i>Journal of Pharmacology and Experimental Therapeutics</i> , 1989, 251, 311-6.	2.5	18
95	Abnormal Glycosylation of Red Cell Membrane Band 3 in the Congenital Disorder of Glycosylation Ig. <i>Pediatric Research</i> , 2003, 54, 224-229.	2.3	17
96	Dioxins in adipose tissue of non-occupationally exposed persons in France: correlation with individual food exposure. <i>Chemosphere</i> , 2001, 44, 1347-1352.	8.2	16
97	Long term outcome of <scp>MPIâ€CDG</scp> patients on Dâ€mannose therapy. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1360-1369.	3.6	16
98	The T911C (F304S) substitution in the human ALG6 gene is a common polymorphism and not a causal mutation of CDG-Ic. <i>Journal of Human Genetics</i> , 2001, 46, 547-548.	2.3	15
99	Two-dimensional gel electrophoresis of apolipoprotein C-III and other serum glycoproteins for the combined screening of human congenital disorders of O- and N-glycosylation. <i>Proteomics - Clinical Applications</i> , 2007, 1, 321-324.	1.6	15
100	Catalase activity, allelic variations in the catalase gene and risk of kidney complications in patients with type 1 diabetes. <i>Diabetologia</i> , 2013, 56, 2733-2742.	6.3	14
101	Comparison of fluconazole and amphotericin B for treatment of experimental <i>Candida albicans</i> endocarditis in rabbits. <i>Antimicrobial Agents and Chemotherapy</i> , 1996, 40, 263-266.	3.2	13
102	Sequential study of serum glycoprotein fucosylation in acute hepatitis. <i>Journal of Hepatology</i> , 1997, 26, 265-271.	3.7	13
103	Absence of Mutation in the <i>SLC2A1</i> Gene in a Cohort of Patients with Alternating Hemiplegia of Childhood (AHC). <i>Neuropediatrics</i> , 2010, 41, 267-269.	0.6	13
104	Intragenic rearrangements in LARGE and POMGNT1 genes in severe dystroglycanopathies. <i>Neuromuscular Disorders</i> , 2011, 21, 782-790.	0.6	13
105	Expanding the Spectrum of PMM2-CDG Phenotype. <i>JIMD Reports</i> , 2011, 5, 123-125.	1.5	13
106	Congenital disorder of glycosylation Ia with deficient phosphomannomutase activity but normal plasma glycoprotein pattern. <i>Clinical Chemistry</i> , 2001, 47, 132-4.	3.2	13
107	Increased Biosynthesis of Glycosphingolipids in Congenital Disorder of Glycosylation Ia (CDG-Ia) Fibroblasts. <i>Pediatric Research</i> , 2002, 52, 645-651.	2.3	12
108	PMM2 intronic branch-site mutations in CDG-Ia. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 337-340.	1.1	12

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109	Homozygous Truncating Intragenic Duplication in TUSC3 Responsible for Rare Autosomal Recessive Nonsyndromic Intellectual Disability with No Clinical or Biochemical Metabolic Markers. <i>JIMD Reports</i> , 2014, 20, 45-55.	1.5	12
110	Novel variants and clinical symptoms in four new ALG3-CDG patients, review of the literature, and identification of AAGRP-ALG3 as a novel ALG3 variant with alanine and glycine-rich N-terminus. <i>Human Mutation</i> , 2019, 40, 938-951.	2.5	12
111	Serum bikunin isoforms in congenital disorders of glycosylation and linkeropathies. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1349-1359.	3.6	12
112	Reduction in biliary excretion of ceftriaxone by diclofenac in rabbits. <i>Antimicrobial Agents and Chemotherapy</i> , 1989, 33, 1506-1510.	3.2	11
113	Two-dimensional electrophoresis highlights haptoglobin beta chain as an additional biomarker of congenital disorders of glycosylation. <i>Clinica Chimica Acta</i> , 2017, 470, 70-74.	1.1	11
114	Comparison of enhanced chemiluminescence and colorimetric techniques for the immuno-detection of $\alpha$ 1-antitrypsin. <i>Clinica Chimica Acta</i> , 1994, 227, 175-184.	1.1	10
115	Does arsenic in soil contribute to arsenic urinary concentrations in a French population living in a naturally arsenic contaminated area?. <i>Science of the Total Environment</i> , 2010, 408, 6011-6016.	8.0	10
116	Arsenic urinary concentrations in children living in a naturally arsenic contaminated area. <i>Journal of Exposure Science and Environmental Epidemiology</i> , 2013, 23, 145-150.	3.9	10
117	Leukocyte Phosphomannomutase Activity in Diagnosis of Congenital Disorder of Glycosylation Ia. <i>Clinical Chemistry</i> , 2002, 48, 934-936.	3.2	9
118	Partial effectiveness of acetazolamide in a mild form of GLUT1 deficiency: A pediatric observation. <i>Movement Disorders</i> , 2013, 28, 1749-1751.	3.9	9
119	Influence of the environmental relative humidity on the inflammatory response of skin model after exposure to various environmental pollutants. <i>Environmental Research</i> , 2021, 196, 110350.	7.5	9
120	Effects of diltiazem on netilmicin-induced nephrotoxicity in rabbits. <i>Antimicrobial Agents and Chemotherapy</i> , 1993, 37, 1790-1798.	3.2	8
121	Serum bikunin is a biomarker of linkeropathies. <i>Clinica Chimica Acta</i> , 2018, 485, 178-180.	1.1	8
122	Wide clinical spectrum in ALG8-CDG: clues from molecular findings suggest an explanation for a milder phenotype in the first-described patient. <i>Pediatric Research</i> , 2019, 85, 384-389.	2.3	8
123	Two dimensional gel electrophoresis of apolipoprotein C-II and MALDI-TOF MS are complementary techniques for the study of combined defects in <i>N</i> -and mucin type <i>O</i> -glycan biosynthesis. <i>Proteomics - Clinical Applications</i> , 2008, 2, 1670-1674.	1.6	7
124	A Cause of Permanent Ketosis: GLUT-1 Deficiency. <i>JIMD Reports</i> , 2014, 18, 79-83.	1.5	7
125	220th ENMC workshop: Dystroglycan and the dystroglycanopathies Naarden, The Netherlands, 27-29 May 2016. <i>Neuromuscular Disorders</i> , 2017, 27, 387-395.	0.6	7
126	Dilated cardiomyopathy and limb-girdle muscular dystrophy-dystroglycanopathy due to novel pathogenic variants in the DPM3 gene. <i>Neuromuscular Disorders</i> , 2019, 29, 497-502.	0.6	7



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127	Expanding the phenotype of X-linked SSR4 CDG: Connective tissue implications. <i>Human Mutation</i> , 2021, 42, 142-149.	2.5	7
128	A mutation in SLC37A4 causes a dominantly inherited congenital disorder of glycosylation characterized by liver dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 1040-1052.	6.2	7
129	Fluorimetric Measurement of Plasma $\alpha$ -L-Fucosidase Activity with a Centrifugal Analyzer: Reference Values in a Healthy French Adult Population. <i>Clinical Chemistry</i> , 2000, 46, 560-576.	3.2	6
130	Two Novel Homozygous Mutations in Phosphoglucomutase 3 Leading to Severe Combined Immunodeficiency, Skeletal Dysplasia, and Malformations. <i>Journal of Clinical Immunology</i> , 2021, 41, 958-966.	3.8	6
131	Polyclonal antibody-based enzyme-linked immunosorbent assay of alpha 1-acid glycoprotein. <i>Clinical Chemistry</i> , 1990, 36, 666-668.	3.2	5
132	Effect of the Platelet Activating Factor Antagonist BN52021 in Rabbits: Role in Gentamicin Nephrotoxicity. <i>Toxicology and Applied Pharmacology</i> , 1994, 128, 111-115.	2.8	5
133	Alteration of mannose transport in fibroblasts from type I carbohydrate deficient glycoprotein syndrome patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1999, 1453, 369-377.	3.8	5
134	POMGnT1, POMT1, and POMT2 Mutations in Congenital Muscular Dystrophies. <i>Methods in Enzymology</i> , 2010, 479, 343-352.	1.0	5
135	Abnormal Glycosylation Profile and High Alpha-Fetoprotein in a Patient with Twinkle Variants. <i>JIMD Reports</i> , 2016, 29, 109-113.	1.5	5
136	Cell Surface Carbohydrates of Rat Alveolar Type II Cells in Primary Culture. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 1993, 8, 145-152.	2.9	4
137	No Mutation in the SLC2A3 Gene in Cohorts of GLUT1 Deficiency Syndrome-Like Patients Negative for SLC2A1 and in Patients with AHC Negative for ATP1A3. <i>JIMD Reports</i> , 2013, 12, 115-120.	1.5	4
138	Human Reconstituted Nasal Epithelium, a promising in vitro model to assess impacts of environmental complex mixtures. <i>Toxicology in Vitro</i> , 2016, 32, 55-62.	2.4	4
139	Nasal inflammation induced by a common cold: comparison between controls and patients with nasal polyposis under topical steroid therapy. <i>Acta Otorhinolaryngologica Italica</i> , 2007, 27, 78-82.	1.5	4
140	Polyclonal antibody-based enzyme-linked immunosorbent assay of alpha 1-acid glycoprotein. <i>Clinical Chemistry</i> , 1990, 36, 666-9.	3.2	2
141	Leukocyte phosphomannomutase activity in diagnosis of congenital disorder of glycosylation Ia. <i>Clinical Chemistry</i> , 2002, 48, 934-6.	3.2	2
142	Le carbohydrate-deficient glycoprotein syndrome typel : un nouvel $\alpha$ -clairage sur le $\alpha$ -tabolisme du mannose.. <i>Medecine/Sciences</i> , 1999, 15, 1202.	0.2	1
143	Factors influencing the reaction of alpha 1-fetoprotein with concanavalin A and Lens culinaris agglutinin in crossed affinoimmunoelectrophoresis. <i>Clinical Chemistry</i> , 1992, 38, 1418-24.	3.2	1
144	An in vitro model to assess the impact on respiratory cells of air pollutants. <i>Toxicology Letters</i> , 2009, 189, S86.	0.8	0

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145	Effect of formaldehyde on corneal epithelial cells in an air-liquid culture model. Toxicology Letters, 2011, 205, S120.	0.8	0
146	Experimental elements towards induction of premature skin aging related with tobacco smoke exposure. Toxicology Letters, 2014, 229, S129.	0.8	0
147	Assesment of Effects of Formaldehyde Exposure on Respiratory Health: An Innovative in vitro Model. Epidemiology, 2009, 20, S116.	2.7	0
148	Comparison of glycan microheterogeneities of alpha 1-acid glycoprotein between mothers and their newborns. Progress in Clinical and Biological Research, 1989, 300, 139-42.	0.2	0