Gert Matthijs

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/818246/publications.pdf

Version: 2024-02-01

124 papers	6,837 citations	93792 39 h-index	73587 79 g-index
129 all docs	129 docs citations	129 times ranked	8969 citing authors

#	Article	IF	Citations
1	<scp>COG6â€CDG</scp>: Novel variants and novel malformation. Birth Defects Research, 2022, 114, 165-174.	0.8	4
2	Lack of NKG2D in MAGT1-deficient patients is caused by hypoglycosylation. Human Genetics, 2022, 141, 1279-1286.	1.8	6
3	CAMLG-CDG: a novel congenital disorder of glycosylation linked to defective membrane trafficking. Human Molecular Genetics, 2022, , .	1.4	7
4	Knowledge, attitudes and preferences regarding reproductive genetic carrier screening among reproductive-aged men and women in Flanders (Belgium). European Journal of Human Genetics, 2022, , .	1.4	5
5	Reasons affecting the uptake of reproductive genetic carrier screening among nonpregnant reproductiveâ€aged women in Flanders (Belgium). Journal of Genetic Counseling, 2022, 31, 1043-1053.	0.9	5
6	CDG or not CDG. Journal of Inherited Metabolic Disease, 2022, 45, 383-385.	1.7	8
7	DNA testing for sickle cell anemia in Africa: Implementation choices for the Democratic Republic of Congo. Journal of Clinical Laboratory Analysis, 2022, , e24398.	0.9	2
8	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
9	Congenital Disorders of Glycosylation in Portugalâ€"Two Decades of Experience. Journal of Pediatrics, 2021, 231, 148-156.	0.9	9
10	SLC37A4â€CDG : Second patient. JIMD Reports, 2021, 58, 122-128.	0.7	5
11	Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). Cerebellum, 2021, 20, 596-605.	1.4	8
12	SLC35A2-CDG: Novel variant and review. Molecular Genetics and Metabolism Reports, 2021, 26, 100717.	0.4	15
13	Aberrant sialylation in a patient with a HNF1Î± variant and liver adenomatosis. IScience, 2021, 24, 102323.	1.9	4
14	MAN1B1-CDG: novel patients and novel variant. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1207-1209.	0.4	3
15	Expanded carrierÂscreening in Flanders (Belgium): an online survey on the perspectives of nonpregnant reproductive-aged women. Personalized Medicine, 2021, 18, 361-373.	0.8	6
16	The evolving genetic landscape of congenital disorders of glycosylation. Biochimica Et Biophysica Acta - General Subjects, 2021, 1865, 129976.	1.1	24
17	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	2.6	5
18	SRD5A3 defective congenital disorder of glycosylation: clinical utility gene card. European Journal of Human Genetics, 2020, 28, 1297-1300.	1.4	12

#	Article	IF	Citations
19	Interest in expanded carrier screening among individuals and couples in the general population: systematic review of the literature. Human Reproduction Update, 2020, 26, 335-355.	5.2	36
20	Clinical Utility Gene Card for: PGM3 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2019, 27, 1757-1760.	1.4	5
21	Mutations in MAGT1 lead to a glycosylation disorder with a variable phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9865-9870.	3.3	66
22	Long-term follow-up in PMM2-CDG: are we ready to start treatment trials?. Genetics in Medicine, 2019, 21, 1181-1188.	1.1	36
23	Clinical Utility Gene Card For: GALNT3 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2018, 26, 1230-1233.	1.4	4
24	Documento de consenso sobre la implementaciÃ³n de la secuenciaciÃ³n masiva de nueva generaciÃ³n en el diagnÃ³stico genÃ©tico de la predisposiciÃ³n hereditaria al cÃ¡ncer. Medicina ClÃnica, 2018, 151, 80.e1-80.e10.	0.3	7
25	Protective <scp>BCL</scp>11A and <scp>HBS</scp>1Lâ€<scp>MYB</scp> polymorphisms in a cohort of 102 Congolese patients suffering from sickle cell anemia. Journal of Clinical Laboratory Analysis, 2018, 32, .	0.9	6
26	Association between sickle cell anemia and alpha thalassemia reveals a high prevalence of the Î±^{3.7} triplication in congolese patients than in worldwide series. Journal of Clinical Laboratory Analysis, 2018, 32, .	0.9	10
27	Congenital disorders of glycosylation (CDG): Quo vadis?. European Journal of Medical Genetics, 2018, 61, 643-663.	0.7	191
28	Investigating the function of Gdt1p in yeast Golgi glycosylation. Biochimica Et Biophysica Acta - General Subjects, 2018, 1862, 394-402.	1.1	29
29	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. Frontiers in Genetics, 2018, 9, 150.	1.1	26
30	Clinical utility gene card for: B4GALT7-defective congenital disorder of glycosylation. European Journal of Human Genetics, 2017, 25, 271-271.	1.4	0
31	Manganese-induced turnover of TMEM165. Biochemical Journal, 2017, 474, 1481-1493.	1.7	44
32	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. Genetics in Medicine, 2017, 19, 104-111.	1.1	71
33	Galactose Epimerase Deficiency: Expanding the Phenotype. JIMD Reports, 2017, 37, 19-25.	0.7	8
34	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
35	Repeat genetic testing with targeted capture sequencing in primary arrhythmia syndrome and cardiomyopathy. European Journal of Human Genetics, 2017, 25, 1313-1323.	1.4	9
36	PMM2â€CDG and sensorineural hearing loss. Journal of Inherited Metabolic Disease, 2017, 40, 629-630.	1.7	6

#	Article	IF	Citations
37	Galactose Supplementation in Patients With TMEM165-CDG Rescues the Glycosylation Defects. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1375-1386.	1.8	61
38	Mutations inTRAPPC11are associated with a congenital disorder of glycosylation. Human Mutation, 2017, 38, 148-151.	1.1	34
39	Detecting AGG Interruptions in Male and Female FMR1 Premutation Carriers by Single-Molecule Sequencing. Human Mutation, 2017, 38, 324-331.	1.1	37
40	Mutations in the X-linked ATP6AP2 cause a glycosylation disorder with autophagic defects. Journal of Experimental Medicine, 2017, 214, 3707-3729.	4.2	62
41	Intratumoral heterogeneity in colorectal cancer: Can histology be used as a guidance for molecular testing?. Journal of Clinical Oncology, 2017, 35, 611-611.	0.8	3
42	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	1.1	40
43	Clinical utility gene card for: Peters plus syndrome. European Journal of Human Genetics, 2016, 24, 1-3.	1.4	5
44	Clinical utility gene card for: MAN1B1 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2016, 24, 1-3.	1.4	2
45	ALG6â€CDG: a recognizable phenotype with epilepsy, proximal muscle weakness, ataxia and behavioral and limb anomalies. Journal of Inherited Metabolic Disease, 2016, 39, 713-723.	1.7	36
46	Glycosylation abnormalities in Gdt1p/TMEM165 deficient cells result from a defect in Golgi manganese homeostasis. Human Molecular Genetics, 2016, 25, 1489-1500.	1.4	92
47	New EuroGentest/ESHG guidelines and a new clinical utility gene card format for NGS-based testing. European Journal of Human Genetics, 2016, 24, 1-1.	1.4	37
48	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	1.4	389
49	Serum transferrin carrying the xenoâ€tetrasaccharide NeuAcâ€Galâ€GlcNAc₂ is a biomarker of ALG1â€CDG. Journal of Inherited Metabolic Disease, 2016, 39, 107-114.	1.7	21
50	Biochemical and molecular predictors for prognosis in nonketotic hyperglycinemia. Annals of Neurology, 2015, 78, 606-618.	2.8	68
51	ALG3â€CDG: Report of two siblings with antenatal features carrying homozygous p.Gly96Arg mutation. American Journal of Medical Genetics, Part A, 2015, 167, 2748-2754.	0.7	21
52	Global implementation of genomic medicine: We are not alone. Science Translational Medicine, 2015, 7, 290ps13.	5.8	146
53	Abnormal cartilage development and altered N-glycosylation in Tmem165-deficient zebrafish mirrors the phenotypes associated with TMEM165-CDG. Glycobiology, 2015, 25, 669-682.	1.3	29
54	Key features and clinical variability of COG6-CDG. Molecular Genetics and Metabolism, 2015, 116, 163-170.	0.5	49

#	Article	IF	Citations
55	Clinical utility gene card for: ALG6 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2015, 23, 1-3.	1.4	9
56	Clinical utility gene card for: DPAGT1 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2015, 23, 1-3.	1.4	8
57	Clinical utility gene card for: ALG1 defective congenital disorder of glycosylation. European Journal of Human Genetics, 2015, 23, 1431-1431.	1.4	27
58	Correlation between the Lactate Dehydrogenase Levels with Laboratory Variables in the Clinical Severity of Sickle Cell Anemia in Congolese Patients. PLoS ONE, 2015, 10, e0123568.	1.1	19
59	Clinical utility gene card for: Phosphomannose isomerase deficiency. European Journal of Human Genetics, 2014, 22, 1153-1153.	1.4	10
60	Congenital disorders of glycosylation: other causes of ichthyosis. European Journal of Human Genetics, 2014, 22, 444-444.	1.4	8
61	Clinical utility gene card for: Phosphomannomutase 2 deficiency. European Journal of Human Genetics, 2014, 22, 1054-1054.	1.4	2
62	Asymptomatic phosphomannose isomerase deficiency (MPI-CDG) initially mistaken for excessive alcohol consumption. Clinica Chimica Acta, 2014, 431, 15-18.	0.5	29
63	Exome sequencing reveals HINT1 mutations as a cause of distal hereditary motor neuropathy. European Journal of Human Genetics, 2014, 22, 847-850.	1.4	33
64	Mismatch repair deficiency endows tumors with a unique mutation signature and sensitivity to DNA double-strand breaks. ELife, 2014, 3, e02725.	2.8	71
65	The European BRCA patent oppositions and appeals: coloring inside the lines. Nature Biotechnology, 2013, 31, 704-710.	9.4	12
66	Approaches to homozygosity mapping and exome sequencing for the identification of novel types of CDG. Glycoconjugate Journal, 2013, 30, 67-76.	1.4	16
67	MAN1B1 Deficiency: An Unexpected CDG-II. PLoS Genetics, 2013, 9, e1003989.	1.5	63
68	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	2.8	121
69	TMEM165 Deficiency Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2012, 91, 15-26.	2.6	162
70	Severe ALG8-CDG (CDG-Ih) associated with homozygosity for two novel missense mutations detected by exome sequencing of candidate genes. European Journal of Medical Genetics, 2012, 55, 196-202.	0.7	14
71	COG5-CDG: expanding the clinical spectrum. Orphanet Journal of Rare Diseases, 2012, 7, 94.	1.2	36
72	The fate and future of patents on human genes and genetic diagnostic methods. Nature Reviews Genetics, 2012, 13, 441-448.	7.7	27

#	Article	IF	Citations
73	Gene and genetic diagnostic method patent claims: a comparison under current European and US patent law. European Journal of Human Genetics, 2011, 19, 1104-1107.	1.4	18
74	Golgi function and dysfunction in the first COG4-deficient CDG type II patient. Human Molecular Genetics, 2009, 18, 3244-3256.	1.4	129
75	RFT1 deficiency in three novel CDG patients. Human Mutation, 2009, 30, 1428-1434.	1.1	25
76	Legal uncertainty in the area of genetic diagnostic testing. Nature Biotechnology, 2009, 27, 903-909.	9.4	45
77	CDG nomenclature: Time for a change!. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 825-826.	1.8	123
78	Screening for OST deficiencies in unsolved CDG-I patients. Biochemical and Biophysical Research Communications, 2009, 390, 769-774.	1.0	15
79	Introduction. European Journal of Human Genetics, 2008, 16, S1-S2.	1.4	3
80	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	9.4	330
81	Oligosaccharyltransferase-Subunit Mutations in Nonsyndromic Mental Retardation. American Journal of Human Genetics, 2008, 82, 1150-1157.	2.6	130
82	Deficiencies in subunits of the Conserved Oligomeric Golgi (COG) complex define a novel group of Congenital Disorders of Glycosylation. Molecular Genetics and Metabolism, 2008, 93, 15-21.	0.5	97
83	The impact of patenting on DNA diagnostic practice. Clinical Medicine, 2008, 8, 58-60.	0.8	8
84	A new inborn error of glycosylation due to a Cog8 deficiency reveals a critical role for the Cog1â€“Cog8 interaction in COG complex formation. Human Molecular Genetics, 2007, 16, 717-730.	1.4	114
85	Congenital Disorders of Glycosylation: A Rapidly Expanding Disease Family. Annual Review of Genomics and Human Genetics, 2007, 8, 261-278.	2.5	250
86	The European Opposition Against the BRCA Gene Patents*. Familial Cancer, 2006, 5, 95-102.	0.9	28
87	Congenital Disorders of Glycosylation (CDG): Update and Perspectives. Current Pediatric Reviews, 2006, 2, 323-330.	0.4	3
88	Conserved oligomeric Golgi complex subunit 1 deficiency reveals a previously uncharacterized congenital disorder of glycosylation type II. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3764-3769.	3.3	175
89	Research Network: EUROGLYCANET: a European network focused on congenital disorders of glycosylation. European Journal of Human Genetics, 2005, 13, 395-397.	1.4	11
90	Deficiency of the first mannosylation step in the N-glycosylation pathway causes congenital disorder of glycosylation type Ik. Human Molecular Genetics, 2004, 13, 535-542.	1.4	78

#	Article	IF	Citations
91	The prenatal diagnosis of congenital disorders of glycosylation(CDG). Prenatal Diagnosis, 2004, 24, 114-116.	1.1	24
92	European-wide opposition against the breast cancer gene patents. European Journal of Human Genetics, 2002, 10, 783-785.	1.4	16
93	CONGENITALDISORDERS OFGLYCOSYLATION. Annual Review of Genomics and Human Genetics, 2001, 2, 129-151.	2.5	167
94	High Residual Activity of PMM2 in Patientsâ€™ Fibroblasts: Possible Pitfall in the Diagnosis of CDG-Ia (Phosphomannomutase Deficiency). American Journal of Human Genetics, 2001, 68, 347-354.	2.6	93
95	Detection of mutations in theCOL4A5gene by SSCP in X-linked Alport syndrome. Human Mutation, 2001, 18, 141-148.	1.1	38
96	Neurological presentation of a congenital disorder of glycosylation CDG-Ia: Implications for diagnosis and genetic counseling. American Journal of Medical Genetics Part A, 2001, 101, 46-49.	2.4	40
97	Best practice guidelines for molecular analysis in spinal muscular atrophy. European Journal of Human Genetics, 2001, 9, 484-491.	1.4	63
98	Lack of Hardy-Weinberg equilibrium for the most prevalent PMM2 mutation in CDG-Ia (congenital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.4	106
99	Preaxial polydactyly type 1 and severe language deficit in maternal uniparental disomy of chromosome 7. European Journal of Pediatrics, 2000, 159, 929-929.	1.3	4
100	Multi-allelic origin of congenital disorder of glycosylation (CDG)-Ic. Human Genetics, 2000, 106, 538-545.	1.8	25
101	Genotype-phenotype correlations in families with deletions in the von Hippel-Lindau (VHL) gene. Human Genetics, 2000, 106, 425-431.	1.8	75
102	Multi-allelic origin of congenital disorder of glycosylation (CDG)-Ic. Human Genetics, 2000, 106, 538-545.	1.8	62
103	Mutation analysis in Belgian familial colorectal cancer kindreds: High proportion of novel mutations in the mismatch repair genes. Gastroenterology, 2000, 118, A1413.	0.6	0
104	Deficiency of dolichol-phosphate-mannose synthase-1 causes congenital disorder of glycosylation type Ie. Journal of Clinical Investigation, 2000, 105, 233-239.	3.9	146
105	Mutational analysis of the Cu/Zn superoxide dismutase gene in 23 familial and 69 sporadic cases of amyotrophic lateral sclerosis in Belgium. European Journal of Human Genetics, 1999, 7, 599-602.	1.4	26
106	Synthesis and radical polymerization of pyrocarbonate-functionalized monomers: application to positive-tone photoresists. Macromolecular Rapid Communications, 1999, 20, 333-336.	2.0	1
107	Effect of mutations found in carbohydrate-deficient glycoprotein syndrome type IA on the activity of phosphomannomutase 2. FEBS Letters, 1999, 452, 319-322.	1.3	64
108	Fine mapping of Noonan/cardio-facio cutaneous syndrome in a large family. European Journal of Human Genetics, 1998, 6, 32-37.	1.4	77

#	Article	IF	Citations
109	Prenatal diagnosis in CDG1 families: beware of heterogeneity. European Journal of Human Genetics, 1998, 6, 99-104.	1.4	37
110	Detailed mapping of the phosphomannomutase 2 (PMM2) gene and mutation detection enable improved analysis for Scandinavian CDG type I families. European Journal of Human Genetics, 1998, 6, 603-611.	1.4	31
111	Second trimester miscarriage of a male fetus with incontinentia pigmenti. , 1998, 80, 298-299.		9
112	Fluorescent chemical cleavage of mismatches for efficient screening of the factor VIII gene. Human Mutation, 1998, 11, 470-479.	1.1	20
113	Missense mutation and hexanucleotide duplication in the PAX2 gene in two unrelated families with renal-coloboma syndrome (MIM 120330). Human Genetics, 1998, 103, 149-153.	1.8	52
114	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	9.4	733
115	Fluorescent chemical cleavage of mismatches for efficient screening of the factor VIII gene. Human Mutation, 1998, 11, 470-479.	1.1	3
116	In SituSurface Plasmon Resonance Analysis of Dextran Monolayer Degradation by Dextranase. Langmuir, 1997, 13, 7115-7120.	1.6	25
117	High-Resolution Atomic Force Microscopy of Dextran Monolayer Hydration. Langmuir, 1997, 13, 4795-4798.	1.6	18
118	Comparison of PMM1 with the phosphomannomutases expressed in rat liver and in human cells. FEBS Letters, 1997, 411, 251-254.	1.3	37
119	Mutations in PMM2, a phosphomannomutase gene on chromosome 16p13 in carbohydrate-deficient glycoprotein type I syndrome (Jaeken syndrome). Nature Genetics, 1997, 16, 88-92.	9.4	333
120	Clinical and molecular genetic features of congenital spinal muscular atrophy. Annals of Neurology, 1996, 40, 731-738.	2.8	54
121	BINDING AND CONTRACTION-INDUCING ACTIVITY OF MOTILIN ANALOGUES . Biomedical Research, 1988, 9, 361-366.	0.3	10
122	Comparison of the biological activity of canine and porcine motilin in rabbit. Regulatory Peptides, 1986, 15, 333-339.	1.9	26
123	Gene patents: from discovery to invention. A geneticist's view. , 0, , 311-330.		1
124	Dealing with patent fragmentation in ICT and genetics: Patent pools and clearing houses. First Monday, 0, , .	0.6	2