PÃ¥l R NjÃ¸lstad

List of Publications by Year
in descending order

119 papers	10,430 citations	71097 41 h-index	37202 96 g-index
137 all docs	137 docs citations	137 times ranked	13997 citing authors

#	Article	IF	Citations
1	Activating Mutations in the Gene Encoding the ATP-Sensitive Potassium-Channel Subunit Kir6.2 and Permanent Neonatal Diabetes. New England Journal of Medicine, 2004, 350, 1838-1849.	27.0	1,077
2	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	27.8	952
3	Switching from Insulin to Oral Sulfonylureas in Patients with Diabetes Due to Kir6.2 Mutations. New England Journal of Medicine, 2006, 355, 467-477.	27.0	878
4	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
5	Physical Activity Attenuates the Influence of FTO Variants on Obesity Risk: A Meta-Analysis of 218,166 Adults and 19,268 Children. PLoS Medicine, 2011, 8, e1001116.	8.4	446
6	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. Nature Genetics, 2014, 46, 357-363.	21.4	428
7	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. Nature Genetics, 2019, 51, 804-814.	21.4	402
8	Neonatal Diabetes Mellitus Due to Complete Glucokinase Deficiency. New England Journal of Medicine, 2001, 344, 1588-1592.	27.0	386
9	Permanent Neonatal Diabetes due to Mutations in KCNJ11 Encoding Kir6.2. Diabetes, 2004, 53, 2713-2718.	0.6	350
10	Mutations in the CEL VNTR cause a syndrome of diabetes and pancreatic exocrine dysfunction. Nature Genetics, 2006, 38, 54-62.	21.4	296
11	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	21.4	286
12	Association of a Low-Frequency Variant inHNF1AWith Type 2 Diabetes in a Latino Population. JAMA - Journal of the American Medical Association, 2014, 311, 2305.	7.4	230
13	Mutations in the Insulin Gene Can Cause MODY and Autoantibody-Negative Type 1 Diabetes. Diabetes, 2008, 57, 1131-1135.	0.6	184
14	Permanent Neonatal Diabetes Caused by Glucokinase Deficiency. Diabetes, 2003, 52, 2854-2860.	0.6	173
15	A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. Nature Genetics, 2015, 47, 518-522.	21.4	157
16	Genome-wide association study of offspring birth weight in 86â€‰577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. Human Molecular Genetics, 2018, 27, 742-756.	2.9	156
17	Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects. Nature Genetics, 2022, 54, 581-592.	21.4	142
18	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. Nature Genetics, 2013, 45, 1380-1385.	21.4	129

#	Article	IF	Citations
19	Assessing the Causal Relationship of Maternal Height on Birth Size and Gestational Age at Birth: A Mendelian Randomization Analysis. PLoS Medicine, 2015, 12, e1001865.	8.4	121
20	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. Lancet Diabetes and Endocrinology,the, 2018, 6, 637-646.	11.4	120
21	Common and rare forms of diabetes mellitus: towards a continuum of diabetes subtypes. Nature Reviews Endocrinology, 2016, 12, 394-406.	9.6	112
22	Targeted next-generation sequencing reveals MODY in up to 6.5% of antibody-negative diabetes cases listed in the Norwegian Childhood Diabetes Registry. Diabetologia, 2017, 60, 625-635.	6.3	106
23	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. PLoS Genetics, 2020, 16, e1008718.	3.5	95
24	Pancreatic Lipomatosis Is a Structural Marker in Nondiabetic Children With Mutations in Carboxyl-Ester Lipase. Diabetes, 2007, 56, 444-449.	0.6	91
25	Exome Sequencing and Genetic Testing for MODY. PLoS ONE, 2012, 7, e38050.	2.5	91
26	Hepatocyte Nuclear Factor-1Î± Gene Mutations and Diabetes in Norway. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 920-931.	3.6	82
27	Diabetes and Pancreatic Exocrine Dysfunction Due to Mutations in the Carboxyl Ester Lipase Gene-Maturity Onset Diabetes of the Young (CEL-MODY). Journal of Biological Chemistry, 2011, 286, 34593-34605.	3.4	80
28	Infant Feeding and Risk of Type 1 Diabetes in Two Large Scandinavian Birth Cohorts. Diabetes Care, 2017, 40, 920-927.	8.6	78
29	A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. Human Molecular Genetics, 2019, 28, 3327-3338.	2.9	76
30	From Clinicogenetic Studies of Maturity-Onset Diabetes of the Young to Unraveling Complex Mechanisms of Glucokinase Regulation. Diabetes, 2006, 55, 1713-1722.	0.6	72
31	Prospective Associations of Systemic and Urinary Choline Metabolites with Incident Type 2 Diabetes. Clinical Chemistry, 2016, 62, 755-765.	3.2	70
32	Kir6.2 mutations causing neonatal diabetes provide new insights into Kir6.2â€“SUR1 interactions. EMBO Journal, 2005, 24, 2318-2330.	7.8	63
33	A zebrafish engrailed-like homeobox sequence expressed during embryogenesis. FEBS Letters, 1988, 231, 355-360.	2.8	62
34	Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. Human Genetics, 2010, 127, 55-64.	3.8	61
35	Genome-wide association study reveals dynamic role of genetic variation in infant and early childhood growth. Nature Communications, 2019, 10, 4448.	12.8	61
36	Role of molecular genetics in transforming diagnosis of diabetes mellitus. Expert Review of Molecular Diagnostics, 2011, 11, 313-320.	3.1	60

#	Article	IF	Citations
37	The role of the carboxyl ester lipase (CEL) gene in pancreatic disease. Pancreatology, 2018, 18, 12-19.	1.1	60
38	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. Diabetes, 2017, 66, 335-346.	0.6	54
39	The genetic architecture of sporadic and multiple consecutive miscarriage. Nature Communications, 2020, 11, 5980.	12.8	52
40	Diagnostic screening of MODY2/GCKmutations in the Norwegian MODY Registry. Pediatric Diabetes, 2008, 9, 442-449.	2.9	49
41	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. Nature Communications, 2019, 10, 3927.	12.8	49
42	Structure and early embryonic expression of the zebrafish engrailed-2 gene. Mechanisms of Development, 1992, 39, 51-62.	1.7	46
43	Studies in 3,523 Norwegians and Meta-Analysis in 11,571 Subjects Indicate That Variants in the Hepatocyte Nuclear Factor 4Â (HNF4A) P2 Region Are Associated With Type 2 Diabetes in Scandinavians. Diabetes, 2007, 56, 3112-3117.	0.6	46
44	Maternal and Newborn Vitamin Dâ€“Binding Protein, Vitamin D Levels, Vitamin D Receptor Genotype, and Childhood Type 1 Diabetes. Diabetes Care, 2019, 42, 553-559.	8.6	42
45	Population prevalence and inheritance pattern of recurrent CNVs associated with neurodevelopmental disorders in 12,252 newborns and their parents. European Journal of Human Genetics, 2021, 29, 205-215.	2.8	40
46	Endocytosis of Secreted Carboxyl Ester Lipase in a Syndrome of Diabetes and Pancreatic Exocrine Dysfunction. Journal of Biological Chemistry, 2014, 289, 29097-29111.	3.4	39
47	The number of tandem repeats in the carboxyl-ester lipase (CEL) gene as a risk factor in alcoholic and idiopathic chronic pancreatitis. Pancreatology, 2013, 13, 29-32.	1.1	38
48	Carboxyl-Ester Lipase Maturity-Onset Diabetes of the Young Is Associated With Development of Pancreatic Cysts and Upregulated MAPK Signaling in Secretin-Stimulated Duodenal Fluid. Diabetes, 2014, 63, 259-269.	0.6	38
49	Primary structure, developmentally regulated expression and potential duplication of the zebrafish homeobox gene ZF-21. Nucleic Acids Research, 1988, 16, 9097-9111.	14.5	33
50	A Hepatocyte Nuclear Factor-4Â Gene (HNF4A) P2 Promoter Haplotype Linked With Late-Onset Diabetes: Studies of HNF4A Variants in the Norwegian MODY Registry. Diabetes, 2006, 55, 1899-1903.	0.6	33
51	Copy number variants and VNTR length polymorphisms of the carboxyl-ester lipase (CEL) gene as risk factors in pancreatic cancer. Pancreatology, 2017, 17, 83-88.	1.1	33
52	Allosteric Activation of Human Glucokinase by Free Polyubiquitin Chains and Its Ubiquitin-dependent Cotranslational Proteasomal Degradation. Journal of Biological Chemistry, 2007, 282, 22757-22764.	3.4	32
53	The role of pancreatic imaging in monogenic diabetes mellitus. Nature Reviews Endocrinology, 2012, 8, 148-159.	9.6	32
54	High Incidence of HeterozygousABCC8andHNF1AMutations in Czech Patients With Congenital Hyperinsulinism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1540-E1549.	3.6	32

#	Article	IF	Citations
55	Paternal and maternal obesity but not gestational weight gain is associated with type 1 diabetes. International Journal of Epidemiology, 2018, 47, 417-426.	1.9	31
56	Lack of Association Between Maternal or Neonatal Vitamin D Status and Risk of Childhood Type 1 Diabetes: A Scandinavian Case-Cohort Study. American Journal of Epidemiology, 2018, 187, 1174-1181.	3.4	31
57	Genetic association study of childhood aggression across raters, instruments, and age. Translational Psychiatry, 2021, 11, 413.	4.8	31
58	SUMOylation of Pancreatic Glucokinase Regulates Its Cellular Stability and Activity*. Journal of Biological Chemistry, 2013, 288, 5951-5962.	3.4	30
59	MODY Associated with Two Novel Hepatocyte Nuclear Factor-1Î± Loss-of-Function Mutations (P112L and) Tj ETQq1 1 0.784314 rgBT /O	2.1	29
60	Parental Smoking and Risk of Childhood-onset Type 1 Diabetes. Epidemiology, 2018, 29, 848-856.	2.7	28
61	Insitu hybridization patterns of zebrafish homeobox genes homologous to Hox-2.1 and En-2 of mouse. Biochemical and Biophysical Research Communications, 1988, 157, 426-432.	2.1	26
62	Pancreatic Function in Carboxyl-Ester Lipase Knockout Mice. Pancreatology, 2010, 10, 467-476.	1.1	26
63	Characterization of the genetic architecture of infant and early childhood body mass index. Nature Metabolism, 2022, 4, 344-358.	11.9	26
64	Genome-wide Association Meta-analysis of Childhood and Adolescent Internalizing Symptoms. Journal of the American Academy of Child and Adolescent Psychiatry, 2022, 61, 934-945.	0.5	26
65	Functional Dissection of the HNF-1alpha Transcription Factor: A Study on Nuclear Localization and Transcriptional Activation. DNA and Cell Biology, 2005, 24, 661-669.	1.9	25
66	Prenatal iron exposure and childhood type 1 diabetes. Scientific Reports, 2018, 8, 9067.	3.3	25
67	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. American Journal of Human Genetics, 2020, 107, 670-682.	6.2	25
68	Modeling assortative mating and genetic similarities between partners, siblings, and in-laws. Nature Communications, 2022, 13, 1108.	12.8	23
69	A zebrafish homologue of the murineHox-2.1 gene. FEBS Letters, 1988, 230, 25-30.	2.8	22
70	Roadmap for a precision-medicine initiative in the Nordic region. Nature Genetics, 2019, 51, 924-930.	21.4	22
71	Enlarged nephrons and severe nondiabetic nephropathy in hepatocyte nuclear factor-1Î² (HNF-1Î²) mutation carriers. Kidney International, 2003, 64, 793-800.	5.2	21
72	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. European Journal of Pediatrics, 2010, 169, 207-213.	2.7	21

#	Article	IF	Citations
73	On the importance of parenting in externalizing disorders: an evaluation of indirect genetic effects in families. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2022, 63, 1186-1195.	5.2	20
74	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. Pediatric Diabetes, 2012, 13, 155-162.	2.9	19
75	Structure and neural expression of a zebrafish homeobox sequence. Gene, 1988, 73, 33-46.	2.2	18
76	Introducing M-GCTA a Software Package to Estimate Maternal (or Paternal) Genetic Effects on Offspring Phenotypes. Behavior Genetics, 2020, 50, 51-66.	2.1	18
77	An Egyptian family with H syndrome due toÂaÂnovel mutation inSLC29A3illustrating overlapping features with pigmented hypertrichotic dermatosis with insulin-dependent diabetes and Faisalabad histiocytosis. Pediatric Diabetes, 2013, 14, 466-472.	2.9	17
78	Intellectual Disability in KATP Channel Neonatal Diabetes. Diabetes Care, 2020, 43, 526-533.	8.6	16
79	Length of Variable Numbers of Tandem Repeats in the Carboxyl Ester Lipase (CEL) Gene May Confer Susceptibility to Alcoholic Liver Cirrhosis but Not Alcoholic Chronic Pancreatitis. PLoS ONE, 2016, 11, e0165567.	2.5	16
80	GCK-MODY diabetes as a protein misfolding disease: The mutation R275C promotes protein misfolding, self-association and cellular degradation. Molecular and Cellular Endocrinology, 2014, 382, 55-65.	3.2	15
81	GCK-MODY diabetes associated with protein misfolding, cellular self-association and degradation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1705-1715.	3.8	14
82	The mucinous domain of pancreatic carboxyl-ester lipase (CEL) contains core 1/core 2 O-glycans that can be modified by ABO blood group determinants. Journal of Biological Chemistry, 2018, 293, 19476-19491.	3.4	14
83	The E3 SUMO ligase PIASÎ³ is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1Î±. Scientific Reports, 2018, 8, 12780.	3.3	14
84	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study. PLoS Medicine, 2020, 17, e1003032.	8.4	14
85	Functional Analyses of HNF1A-MODY Variants Refine the Interpretation of Identified Sequence Variants. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1377-e1386.	3.6	14
86	Pathogenic Carboxyl Ester Lipase (CEL) Variants Interact with the Normal CEL Protein in Pancreatic Cells. Cells, 2020, 9, 244.	4.1	14
87	Plasma immunological markers in pregnancy and cord blood: AÂpossible link between macrophage chemoâ€attractants and risk of childhood type 1 diabetes. American Journal of Reproductive Immunology, 2018, 79, e12802.	1.2	13
88	The position of single-base deletions in the VNTR sequence of the carboxyl ester lipase (CEL) gene determines proteotoxicity. Journal of Biological Chemistry, 2021, 296, 100661.	3.4	13
89	Continuity of Genetic Risk for Aggressive Behavior Across the Life-Course. Behavior Genetics, 2021, 51, 592-606.	2.1	13
90	The homeobox factor Irx3 maintains adipogenic identity. Metabolism: Clinical and Experimental, 2020, 103, 154014.	3.4	12

#	Article	IF	Citations
91	Two New Mutations in the CEL Gene Causing Diabetes and Hereditary Pancreatitis: How to Correctly Identify MODY8 Cases. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1455-e1466.	3.6	12
92	The Hypoglycemic Phenotype Is Islet Cellâ€“Autonomous in Short-Chain Hydroxyacyl-CoA Dehydrogenaseâ€“Deficient Mice. Diabetes, 2016, 65, 1672-1678.	0.6	11
93	PeptideMapper: efficient and versatile amino acid sequence and tag mapping. Bioinformatics, 2017, 33, 2042-2044.	4.1	11
94	Iris Malformation and Anterior Segment Dysgenesis in Mice and Humans With a Mutation in PI 3-Kinase. , 2017, 58, 3100.		11
95	Does Current Diabetes Technology Improve Metabolic Control? A Cross-Sectional Study on the Use of Insulin Pumps and Continuous Glucose Monitoring Devices in a Nationwide Pediatric Population. Diabetes Therapy, 2021, 12, 2571-2583.	2.5	11
96	Smoking and infertility: multivariable regression and Mendelian randomization analyses in the Norwegian Mother, Father and Child Cohort Study. Fertility and Sterility, 2022, 118, 180-190.	1.0	11
97	Sequence analysis of the zebrafish hox-B5 / B6 region. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1993, 1173, 102-106.	2.4	10
98	A novel <scp>GATA</scp>6 mutation in a child with congenital heart malformation and neonatal diabetes. Clinical Case Reports (discontinued), 2013, 1, 86-90.	0.5	10
99	Developmental milestones in early childhood and genetic liability to neurodevelopmental disorders. Psychological Medicine, 2023, 53, 1750-1758.	4.5	10
100	Sonographic pancreas echogenicity in cystic fibrosis compared to exocrine pancreatic function and pancreas fat content at Dixon-MRI. PLoS ONE, 2018, 13, e0201019.	2.5	9
101	Social and genetic associations with educational performance in a Scandinavian welfare state. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	7
102	Prediction of Type 1 Diabetes at Birth: Cord Blood Metabolites vs Genetic Risk Score in the Norwegian Mother, Father, and Child Cohort. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e4062-e4071.	3.6	6
103	The Chromosome 9p21 CVD- and T2D-Associated Regions in a Norwegian Population (The HUNT2 Survey). International Journal of Endocrinology, 2015, 2015, 1-9.	1.5	5
104	Nuclear import of glucokinase in pancreatic beta-cells is mediated by a nuclear localization signal and modulated by SUMOylation. Molecular and Cellular Endocrinology, 2017, 454, 146-157.	3.2	5
105	Characterization of CEL-DUP2: Complete duplication of the carboxyl ester lipase gene is unlikely to influence risk of chronic pancreatitis. Pancreatology, 2020, 20, 377-384.	1.1	5
106	PathwayMatcher: proteoform-centric network construction enables fine-granularity multiomics pathway mapping. GigaScience, 2019, 8, .	6.4	4
107	Maternal microchimerism in cord blood and risk of childhoodâ€onset type 1 diabetes. Pediatric Diabetes, 2019, 20, 728-735.	2.9	4
108	SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. Bioinformatics, 2021, 37, 1876-1883.	4.1	4

#	Article	IF	Citations
109	Maturity onset diabetes of the young type 2 (MODY2): Insight from an extended family. Diabetes Research and Clinical Practice, 2021, 175, 108791.	2.8	4
110	Spatial Environment Affects HNF4A Mutation-Specific Proteome Signatures and Cellular Morphology in hiPSC-Derived Î²-Like Cells. Diabetes, 2022, 71, 862-869.	0.6	4
111	Fucosylated AGP glycopeptides as biomarkers of HNF1A-Maturity onset diabetes of the young. Diabetes Research and Clinical Practice, 2022, 185, 109226.	2.8	4
112	Structural and biophysical characterization of transcription factor HNF-1A as a tool to study MODY3 diabetes variants. Journal of Biological Chemistry, 2022, 298, 101803.	3.4	4
113	Impact of overweight on glucose homeostasis in MODY2 and MODY3. Diabetic Medicine, 2021, 38, e14649.	2.3	3
114	Homeobox sequences of Atlantic salmon (Salmo salar) and zebrafish (Brachydanio rerio). Aquaculture, 1990, 85, 51-60.	3.5	2
115	An Egyptian case of congenital hyperinsulinism of infancy due to a novel mutation in KCNJ11 encoding Kir6.2 and response to octreotide. Acta Diabetologica, 2013, 50, 801-805.	2.5	2
116	Autozygosity mapping and time-to-spontaneous delivery in Norwegian parent-offspring trios. Human Molecular Genetics, 2021, 29, 3845-3858.	2.9	1
117	Title is missing!. , 2020, 17, e1003032.		0
118	Title is missing!. , 2020, 17, e1003032.		0
119	Title is missing!. , 2020, 17, e1003032.		0