

Pl R Njlstad

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

117
papers

7,559
citations

35
h-index

86
g-index

137
ext. papers

9,149
ext. citations

10.1
avg, IF

4.73
L-index

#	Paper	IF	Citations
117	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. <i>New England Journal of Medicine</i> , 2004 , 350, 1838-49	59.2	930
116	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. <i>New England Journal of Medicine</i> , 2006 , 355, 467-77	59.2	740
115	The genetic architecture of type 2 diabetes. <i>Nature</i> , 2016 , 536, 41-47	50.4	704
114	Rare and low-frequency coding variants alter human adult height. <i>Nature</i> , 2017 , 542, 186-190	50.4	412
113	Physical activity attenuates the influence of FTO variants on obesity risk: a meta-analysis of 218,166 adults and 19,268 children. <i>PLoS Medicine</i> , 2011 , 8, e1001116	11.6	379
112	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. <i>Nature Genetics</i> , 2014 , 46, 357-63	36.3	351
111	Neonatal diabetes mellitus due to complete glucokinase deficiency. <i>New England Journal of Medicine</i> , 2001 , 344, 1588-92	59.2	350
110	Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. <i>Diabetes</i> , 2004 , 53, 2713-8	0.9	314
109	Mutations in the CEL VNTR cause a syndrome of diabetes and pancreatic exocrine dysfunction. <i>Nature Genetics</i> , 2006 , 38, 54-62	36.3	246
108	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. <i>Nature Genetics</i> , 2018 , 50, 26-41	36.3	186
107	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. <i>Nature Genetics</i> , 2019 , 51, 804-814	36.3	181
106	Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. <i>JAMA - Journal of the American Medical Association</i> , 2014 , 311, 2305-14	27.4	164
105	Mutations in the insulin gene can cause MODY and autoantibody-negative type 1 diabetes. <i>Diabetes</i> , 2008 , 57, 1131-5	0.9	154
104	Permanent neonatal diabetes caused by glucokinase deficiency: inborn error of the glucose-insulin signaling pathway. <i>Diabetes</i> , 2003 , 52, 2854-60	0.9	150
103	A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. <i>Nature Genetics</i> , 2015 , 47, 518-522	36.3	111
102	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. <i>Nature Genetics</i> , 2013 , 45, 1380-5	36.3	103
101	Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. <i>Human Molecular Genetics</i> , 2018 , 27, 742-756	5.6	98

100	Common and rare forms of diabetes mellitus: towards a continuum of diabetes subtypes. <i>Nature Reviews Endocrinology</i> , 2016 , 12, 394-406	15.2	79
99	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. <i>Lancet Diabetes and Endocrinology</i> , 2018 , 6, 637-646	18.1	77
98	Exome sequencing and genetic testing for MODY. <i>PLoS ONE</i> , 2012 , 7, e38050	3.7	76
97	Pancreatic lipomatosis is a structural marker in nondiabetic children with mutations in carboxyl-ester lipase. <i>Diabetes</i> , 2007 , 56, 444-9	0.9	74
96	Assessing the Causal Relationship of Maternal Height on Birth Size and Gestational Age at Birth: A Mendelian Randomization Analysis. <i>PLoS Medicine</i> , 2015 , 12, e1001865	11.6	69
95	Targeted next-generation sequencing reveals MODY in up to 6.5% of antibody-negative diabetes cases listed in the Norwegian Childhood Diabetes Registry. <i>Diabetologia</i> , 2017 , 60, 625-635	10.3	67
94	Hepatocyte nuclear factor-1 alpha gene mutations and diabetes in Norway. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003 , 88, 920-31	5.6	63
93	From clinicogenetic studies of maturity-onset diabetes of the young to unraveling complex mechanisms of glucokinase regulation. <i>Diabetes</i> , 2006 , 55, 1713-22	0.9	61
92	Diabetes and pancreatic exocrine dysfunction due to mutations in the carboxyl ester lipase gene-maturity onset diabetes of the young (CEL-MODY): a protein misfolding disease. <i>Journal of Biological Chemistry</i> , 2011 , 286, 34593-605	5.4	58
91	Kir6.2 mutations causing neonatal diabetes provide new insights into Kir6.2-SUR1 interactions. <i>EMBO Journal</i> , 2005 , 24, 2318-30	13	56
90	Prospective Associations of Systemic and Urinary Choline Metabolites with Incident Type 2 Diabetes. <i>Clinical Chemistry</i> , 2016 , 62, 755-65	5.5	52
89	A zebrafish engrailed-like homeobox sequence expressed during embryogenesis. <i>FEBS Letters</i> , 1988 , 231, 355-60	3.8	50
88	Role of molecular genetics in transforming diagnosis of diabetes mellitus. <i>Expert Review of Molecular Diagnostics</i> , 2011 , 11, 313-20	3.8	48
87	Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. <i>Human Genetics</i> , 2010 , 127, 55-64	6.3	44
86	Structure and early embryonic expression of the zebrafish engrailed-2 gene. <i>Mechanisms of Development</i> , 1992 , 39, 51-62	1.7	44
85	Infant Feeding and Risk of Type 1 Diabetes in Two Large Scandinavian Birth Cohorts. <i>Diabetes Care</i> , 2017 , 40, 920-927	14.6	40
84	Studies in 3,523 Norwegians and meta-analysis in 11,571 subjects indicate that variants in the hepatocyte nuclear factor 4 alpha (HNF4A) P2 region are associated with type 2 diabetes in Scandinavians. <i>Diabetes</i> , 2007 , 56, 3112-7	0.9	37
83	Diagnostic screening of MODY2/GCK mutations in the Norwegian MODY Registry. <i>Pediatric Diabetes</i> , 2008 , 9, 442-9	3.6	36

82	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. <i>Diabetes</i> , 2017 , 66, 335-346	0.9	35
81	Genome-wide association study reveals dynamic role of genetic variation in infant and early childhood growth. <i>Nature Communications</i> , 2019 , 10, 4448	17.4	34
80	The number of tandem repeats in the carboxyl-ester lipase (CEL) gene as a risk factor in alcoholic and idiopathic chronic pancreatitis. <i>Pancreatology</i> , 2013 , 13, 29-32	3.8	31
79	Primary structure, developmentally regulated expression and potential duplication of the zebrafish homeobox gene ZF-21. <i>Nucleic Acids Research</i> , 1988 , 16, 9097-111	20.1	31
78	Maternal and Newborn Vitamin D-Binding Protein, Vitamin D Levels, Vitamin D Receptor Genotype, and Childhood Type 1 Diabetes. <i>Diabetes Care</i> , 2019 , 42, 553-559	14.6	30
77	A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. <i>Human Molecular Genetics</i> , 2019 , 28, 3327-3338	5.6	30
76	Allosteric activation of human glucokinase by free polyubiquitin chains and its ubiquitin-dependent cotranslational proteasomal degradation. <i>Journal of Biological Chemistry</i> , 2007 , 282, 22757-64	5.4	29
75	The role of pancreatic imaging in monogenic diabetes mellitus. <i>Nature Reviews Endocrinology</i> , 2011 , 8, 148-59	15.2	28
74	The role of the carboxyl ester lipase (CEL) gene in pancreatic disease. <i>Pancreatology</i> , 2018 , 18, 12-19	3.8	26
73	A hepatocyte nuclear factor-4 alpha gene (HNF4A) P2 promoter haplotype linked with late-onset diabetes: studies of HNF4A variants in the Norwegian MODY registry. <i>Diabetes</i> , 2006 , 55, 1899-903	0.9	26
72	High Incidence of Heterozygous ABCC8 and HNF1A Mutations in Czech Patients With Congenital Hyperinsulinism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, E1540-9	5.6	25
71	SUMOylation of pancreatic glucokinase regulates its cellular stability and activity. <i>Journal of Biological Chemistry</i> , 2013 , 288, 5951-62	5.4	25
70	MODY associated with two novel hepatocyte nuclear factor-1alpha loss-of-function mutations (P112L and Q466X). <i>Biochemical and Biophysical Research Communications</i> , 2000 , 279, 792-8	3.4	25
69	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. <i>PLoS Genetics</i> , 2020 , 16, e1008718	6	25
68	Endocytosis of secreted carboxyl ester lipase in a syndrome of diabetes and pancreatic exocrine dysfunction. <i>Journal of Biological Chemistry</i> , 2014 , 289, 29097-111	5.4	24
67	Paternal and maternal obesity but not gestational weight gain is associated with type 1 diabetes. <i>International Journal of Epidemiology</i> , 2018 , 47, 417-426	7.8	23
66	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. <i>Diabetes</i> , 2014 , 63, 259-69	0.9	23
65	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. <i>Nature Communications</i> , 2019 , 10, 3927	17.4	21

64	Lack of Association Between Maternal or Neonatal Vitamin D Status and Risk of Childhood Type 1 Diabetes: A Scandinavian Case-Cohort Study. <i>American Journal of Epidemiology</i> , 2018 , 187, 1174-1181	3.8	20
63	A zebrafish homologue of the murine Hox-2.1 gene. <i>FEBS Letters</i> , 1988 , 230, 25-30	3.8	20
62	In situ hybridization patterns of zebrafish homeobox genes homologous to Hox-2.1 and En-2 of mouse. <i>Biochemical and Biophysical Research Communications</i> , 1988 , 157, 426-32	3.4	20
61	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. <i>European Journal of Pediatrics</i> , 2010 , 169, 207-13	4.1	19
60	Parental Smoking and Risk of Childhood-onset Type 1 Diabetes. <i>Epidemiology</i> , 2018 , 29, 848-856	3.1	19
59	Pancreatic function in carboxyl-ester lipase knockout mice. <i>Pancreatology</i> , 2010 , 10, 467-76	3.8	18
58	Functional dissection of the HNF-1alpha transcription factor: a study on nuclear localization and transcriptional activation. <i>DNA and Cell Biology</i> , 2005 , 24, 661-9	3.6	18
57	Copy number variants and VNTR length polymorphisms of the carboxyl-ester lipase (CEL) gene as risk factors in pancreatic cancer. <i>Pancreatology</i> , 2017 , 17, 83-88	3.8	17
56	Structure and neural expression of a zebrafish homeobox sequence. <i>Gene</i> , 1988 , 73, 33-46	3.8	17
55	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. <i>Pediatric Diabetes</i> , 2012 , 13, 155-62	3.6	15
54	An Egyptian family with H syndrome due to a novel mutation in SLC29A3 illustrating overlapping features with pigmented hypertrichotic dermatosis with insulin-dependent diabetes and Faisalabad histiocytosis. <i>Pediatric Diabetes</i> , 2013 , 14, 466-72	3.6	14
53	Enlarged nephrons and severe nondiabetic nephropathy in hepatocyte nuclear factor-1beta (HNF-1beta) mutation carriers. <i>Kidney International</i> , 2003 , 64, 793-800	9.9	14
52	Roadmap for a precision-medicine initiative in the Nordic region. <i>Nature Genetics</i> , 2019 , 51, 924-930	36.3	12
51	GCK-MODY diabetes associated with protein misfolding, cellular self-association and degradation. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012 , 1822, 1705-15	6.9	12
50	Prenatal iron exposure and childhood type 1 diabetes. <i>Scientific Reports</i> , 2018 , 8, 9067	4.9	11
49	GCK-MODY diabetes as a protein misfolding disease: the mutation R275C promotes protein misfolding, self-association and cellular degradation. <i>Molecular and Cellular Endocrinology</i> , 2014 , 382, 55-65	4.4	11
48	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. <i>PLoS ONE</i> , 2016 , 11, e0165567	3.7	11
47	The genetic architecture of sporadic and multiple consecutive miscarriage. <i>Nature Communications</i> , 2020 , 11, 5980	17.4	11

46	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. <i>American Journal of Human Genetics</i> , 2020 , 107, 670-682	11	9
45	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study. <i>PLoS Medicine</i> , 2020 , 17, e1003032	11.6	9
44	Functional Analyses of HNF1A-MODY Variants Refine the Interpretation of Identified Sequence Variants. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	9
43	The Hypoglycemic Phenotype Is Islet Cell-Autonomous in Short-Chain Hydroxyacyl-CoA Dehydrogenase-Deficient Mice. <i>Diabetes</i> , 2016 , 65, 1672-8	0.9	9
42	Population prevalence and inheritance pattern of recurrent CNVs associated with neurodevelopmental disorders in 12,252 newborns and their parents. <i>European Journal of Human Genetics</i> , 2021 , 29, 205-215	5.3	9
41	A novel GATA6 mutation in a child with congenital heart malformation and neonatal diabetes. <i>Clinical Case Reports (discontinued)</i> , 2013 , 1, 86-90	0.7	8
40	Sequence analysis of the zebrafish hox-B5/B6 region. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1993 , 1173, 102-6		8
39	Intellectual Disability in K Channel Neonatal Diabetes. <i>Diabetes Care</i> , 2020 , 43, 526-533	14.6	7
38	Pathogenic Carboxyl Ester Lipase (CEL) Variants Interact with the Normal CEL Protein in Pancreatic Cells. <i>Cells</i> , 2020 , 9,	7.9	7
37	Genetic association study of childhood aggression across raters, instruments, and age. <i>Translational Psychiatry</i> , 2021 , 11, 413	8.6	7
36	The mucinous domain of pancreatic carboxyl-ester lipase (CEL) contains core 1/core 2 glycans that can be modified by ABO blood group determinants. <i>Journal of Biological Chemistry</i> , 2018 , 293, 19476-19491	5.4	7
35	PeptideMapper: efficient and versatile amino acid sequence and tag mapping. <i>Bioinformatics</i> , 2017 , 33, 2042-2044	7.2	6
34	Plasma immunological markers in pregnancy and cord blood: A possible link between macrophage chemo-attractants and risk of childhood type 1 diabetes. <i>American Journal of Reproductive Immunology</i> , 2018 , 79, e12802	3.8	6
33	The homeobox factor Irx3 maintains adipogenic identity. <i>Metabolism: Clinical and Experimental</i> , 2020 , 103, 154014	12.7	6
32	Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects.. <i>Nature Genetics</i> , 2022 , 54, 581-592	36.3	6
31	Iris Malformation and Anterior Segment Dysgenesis in Mice and Humans With a Mutation in PI 3-Kinase 2017 , 58, 3100-3106		5
30	Sonographic pancreas echogenicity in cystic fibrosis compared to exocrine pancreatic function and pancreas fat content at Dixon-MRI. <i>PLoS ONE</i> , 2018 , 13, e0201019	3.7	5
29	Nuclear import of glucokinase in pancreatic beta-cells is mediated by a nuclear localization signal and modulated by SUMOylation. <i>Molecular and Cellular Endocrinology</i> , 2017 , 454, 146-157	4.4	5

28	Introducing M-GCTA a Software Package to Estimate Maternal (or Paternal) Genetic Effects on Offspring Phenotypes. <i>Behavior Genetics</i> , 2020 , 50, 51-66	3.2	5
27	The E3 SUMO ligase PIAS1 is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1. <i>Scientific Reports</i> , 2018 , 8, 12780	4.9	5
26	The Chromosome 9p21 CVD- and T2D-Associated Regions in a Norwegian Population (The HUNT2 Survey). <i>International Journal of Endocrinology</i> , 2015 , 2015, 164652	2.7	3
25	Genome-wide association meta-analysis of childhood and adolescent internalising symptoms		3
24	The position of single-base deletions in the VNTR sequence of the carboxyl ester lipase (CEL) gene determines proteotoxicity. <i>Journal of Biological Chemistry</i> , 2021 , 296, 100661	5.4	3
23	Maternal microchimerism in cord blood and risk of childhood-onset type 1 diabetes. <i>Pediatric Diabetes</i> , 2019 , 20, 728-735	3.6	2
22	Characterization of CEL-DUP2: Complete duplication of the carboxyl ester lipase gene is unlikely to influence risk of chronic pancreatitis. <i>Pancreatology</i> , 2020 , 20, 377-384	3.8	2
21	PathwayMatcher: proteoform-centric network construction enables fine-granularity multiomics pathway mapping. <i>GigaScience</i> , 2019 , 8,	7.6	2
20	An Egyptian case of congenital hyperinsulinism of infancy due to a novel mutation in KCNJ11 encoding Kir6.2 and response to octreotide. <i>Acta Diabetologica</i> , 2013 , 50, 801-5	3.9	2
19	Homeobox sequences of Atlantic salmon (<i>Salmo salar</i>) and zebrafish (<i>Brachydanio rerio</i>). <i>Aquaculture</i> , 1990 , 85, 51-60	4.4	2
18	How important are parents in the development of child anxiety and depression? A genomic analysis of parent-offspring trios in the Norwegian Mother Father and Child Cohort Study (MoBa)		2
17	Prediction of Type 1 Diabetes at Birth: Cord Blood Metabolites vs Genetic Risk Score in the Norwegian Mother, Father, and Child Cohort. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e4062-e4071	5.6	2
16	Continuity of Genetic Risk for Aggressive Behavior Across the Life-Course. <i>Behavior Genetics</i> , 2021 , 51, 592-606	3.2	2
15	Characterization of the genetic architecture of infant and early childhood body mass index.. <i>Nature Metabolism</i> , 2022 ,	14.6	2
14	Genome-wide Association Meta-analysis of Childhood and Adolescent Internalizing Symptoms.. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2022 ,	7.2	2
13	Autozygosity mapping and time-to-spontaneous delivery in Norwegian parent-offspring trios. <i>Human Molecular Genetics</i> , 2021 , 29, 3845-3858	5.6	1
12	Characterization of the genetic architecture of BMI in infancy and early childhood reveals age-specific effects and implicates pathways involved in Mendelian obesity		1
11	Maturity onset diabetes of the young type 2 (MODY2): Insight from an extended family. <i>Diabetes Research and Clinical Practice</i> , 2021 , 175, 108791	7.4	1

10	SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. <i>Bioinformatics</i> , 2021 ,	7.2	1
9	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. <i>Diabetes Therapy</i> , 2021 , 12, 2571-2583	3.6	1
8	Fucosylated AGP glycopeptides as biomarkers of HNF1A-Maturity Onset Diabetes of the Young.. <i>Diabetes Research and Clinical Practice</i> , 2022 , 109226	7.4	0
7	Impact of overweight on glucose homeostasis in MODY2 and MODY3. <i>Diabetic Medicine</i> , 2021 , 38, e14649-5	5.5	0
6	Developmental milestones in early childhood and genetic liability to neurodevelopmental disorders. <i>Psychological Medicine</i> , 1-9	6.9	0
5	Modeling assortative mating and genetic similarities between partners, siblings, and in-laws.. <i>Nature Communications</i> , 2022 , 13, 1108	17.4	0
4	Structural and biophysical characterization of transcription factor HNF-1A as a tool to study MODY3 diabetes variants.. <i>Journal of Biological Chemistry</i> , 2022 , 101803	5.4	0
3	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study 2020 , 17, e1003032		
2	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study 2020 , 17, e1003032		
1	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study 2020 , 17, e1003032		