Pl R Njlstad

List of Publications by Year in Descending Order

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	Fucosylated AGP glycopeptides as biomarkers of HNF1A-Maturity Onset Diabetes of the Young.. *Diabetes Research and Clinical Practice*, **2022**, 109226	7.4	0
116	Characterization of the genetic architecture of infant and early childhood body mass index.. *Nature Metabolism*, **2022**,	14.6	2
115	Modeling assortative mating and genetic similarities between partners, siblings, and in-laws.. *Nature Communications*, **2022**, 13, 1108	17.4	0
114	Genome-wide Association Meta-analysis of Childhood and Adolescent Internalizing Symptoms.. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2022**,	7.2	2
113	Structural and biophysical characterization of transcription factor HNF-1A as a tool to study MODY3 diabetes variants.. *Journal of Biological Chemistry*, **2022**, 101803	5.4	0
112	Within-sibship genome-wide association analyses decrease bias in estimates of direct genetic effects.. *Nature Genetics*, **2022**, 54, 581-592	36.3	6
111	Autozygosity mapping and time-to-spontaneous delivery in Norwegian parent-offspring trios. *Human Molecular Genetics*, **2021**, 29, 3845-3858	5.6	1
110	Maturity onset diabetes of the young type 2 (MODY2): Insight from an extended family. *Diabetes Research and Clinical Practice*, **2021**, 175, 108791	7.4	1
109	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	5.6	2
108	Genetic association study of childhood aggression across raters, instruments, and age. *Translational Psychiatry*, **2021**, 11, 413	8.6	7
107	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	5.3	9
106	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	5.4	3
105	SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. *Bioinformatics*, **2021**,	7.2	1
104	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	3.6	1
103	Impact of overweight on glucose homeostasis in MODY2 and MODY3. *Diabetic Medicine*, **2021**, 38, e14649	3.5	0
102	Continuity of Genetic Risk for Aggressive Behavior Across the Life-Course. *Behavior Genetics*, **2021**, 51, 592-606	3.2	2
101	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. *American Journal of Human Genetics*, **2020**, 107, 670-682	11	9

100	Intellectual Disability in K Channel Neonatal Diabetes. *Diabetes Care*, **2020**, 43, 526-533	14.6	7
99	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study. *PLoS Medicine*, **2020**, 17, e1003032	11.6	9
98	Functional Analyses of HNF1A-MODY Variants Refine the Interpretation of Identified Sequence Variants. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	9
97	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	3.8	2
96	Pathogenic Carboxyl Ester Lipase (CEL) Variants Interact with the Normal CEL Protein in Pancreatic Cells. *Cells*, **2020**, 9,	7.9	7
95	Introducing M-GCTA a Software Package to Estimate Maternal (or Paternal) Genetic Effects on Offspring Phenotypes. *Behavior Genetics*, **2020**, 50, 51-66	3.2	5
94	The homeobox factor Irx3 maintains adipogenic identity. *Metabolism: Clinical and Experimental*, **2020**, 103, 154014	12.7	6
93	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. *PLoS Genetics*, **2020**, 16, e1008718	6	25
92	The genetic architecture of sporadic and multiple consecutive miscarriage. *Nature Communications*, **2020**, 11, 5980	17.4	11
91	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study **2020**, 17, e1003032		
90	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study **2020**, 17, e1003032		
89	Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study **2020**, 17, e1003032		
88	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. *Nature Communications*, **2019**, 10, 3927	17.4	21
87	Maternal and Newborn Vitamin D-Binding Protein, Vitamin D Levels, Vitamin D Receptor Genotype, and Childhood Type 1 Diabetes. *Diabetes Care*, **2019**, 42, 553-559	14.6	30
86	Maternal microchimerism in cord blood and risk of childhood-onset type 1 diabetes. *Pediatric Diabetes*, **2019**, 20, 728-735	3.6	2
85	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. *Nature Genetics*, **2019**, 51, 804-814	36.3	181
84	Roadmap for a precision-medicine initiative in the Nordic region. *Nature Genetics*, **2019**, 51, 924-930	36.3	12
83	A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. *Human Molecular Genetics*, **2019**, 28, 3327-3338	5.6	30

82	PathwayMatcher: proteoform-centric network construction enables fine-granularity multiomics pathway mapping. *GigaScience*, **2019**, 8,	7.6	2
81	Genome-wide association study reveals dynamic role of genetic variation in infant and early childhood growth. *Nature Communications*, **2019**, 10, 4448	17.4	34
80	Paternal and maternal obesity but not gestational weight gain is associated with type 1 diabetes. *International Journal of Epidemiology*, **2018**, 47, 417-426	7.8	23
79	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. *Human Molecular Genetics*, **2018**, 27, 742-756	5.6	98
78	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	3.8	20
77	The role of the carboxyl ester lipase (CEL) gene in pancreatic disease. *Pancreatology*, **2018**, 18, 12-19	3.8	26
76	Plasma immunological markers in pregnancy and cord blood: A possible link between macrophage chemo-attractants and risk of childhood type 1 diabetes. *American Journal of Reproductive Immunology*, **2018**, 79, e12802	3.8	6
75	Sonographic pancreas echogenicity in cystic fibrosis compared to exocrine pancreatic function and pancreas fat content at Dixon-MRI. *PLoS ONE*, **2018**, 13, e0201019	3.7	5
74	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	18.1	77
73	Prenatal iron exposure and childhood type 1 diabetes. *Scientific Reports*, **2018**, 8, 9067	4.9	11
72	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
71	Parental Smoking and Risk of Childhood-onset Type 1 Diabetes. *Epidemiology*, **2018**, 29, 848-856	3.1	19
70	The mucinous domain of pancreatic carboxyl-ester lipase (CEL) contains core 1/core 2 glycans that can be modified by ABO blood group determinants. *Journal of Biological Chemistry*, **2018**, 293, 19476-19491	5.4	7
69	The E3 SUMO ligase PIAS is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1 *Scientific Reports*, **2018**, 8, 12780	4.9	5
68	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
67	Infant Feeding and Risk of Type 1 Diabetes in Two Large Scandinavian Birth Cohorts. *Diabetes Care*, **2017**, 40, 920-927	14.6	40
66	PeptideMapper: efficient and versatile amino acid sequence and tag mapping. *Bioinformatics*, **2017**, 33, 2042-2044	7.2	6
65	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. *Diabetes*, **2017**, 66, 335-346	0.9	35

64	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	10.3	67
63	Iris Malformation and Anterior Segment Dysgenesis in Mice and Humans With a Mutation in PI 3-Kinase **2017**, 58, 3100-3106		5
62	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	4.4	5
61	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	3.8	17
60	Prospective Associations of Systemic and Urinary Choline Metabolites with Incident Type 2 Diabetes. *Clinical Chemistry*, **2016**, 62, 755-65	5.5	52
59	The Hypoglycemic Phenotype Is Islet Cell-Autonomous in Short-Chain Hydroxyacyl-CoA Dehydrogenase-Deficient Mice. *Diabetes*, **2016**, 65, 1672-8	0.9	9
58	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	3.7	11
57	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
56	Common and rare forms of diabetes mellitus: towards a continuum of diabetes subtypes. *Nature Reviews Endocrinology*, **2016**, 12, 394-406	15.2	79
55	A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. *Nature Genetics*, **2015**, 47, 518-522	36.3	111
54	High Incidence of Heterozygous ABCC8 and HNF1A Mutations in Czech Patients With Congenital Hyperinsulinism. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1540-9	5.6	25
53	The Chromosome 9p21 CVD- and T2D-Associated Regions in a Norwegian Population (The HUNT2 Survey). *International Journal of Endocrinology*, **2015**, 2015, 164652	2.7	3
52	Assessing the Causal Relationship of Maternal Height on Birth Size and Gestational Age at Birth: A Mendelian Randomization Analysis. *PLoS Medicine*, **2015**, 12, e1001865	11.6	69
51	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	4.4	11
50	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. *Nature Genetics*, **2014**, 46, 357-63	36.3	351
49	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, 259-69	0.9	23
48	Endocytosis of secreted carboxyl ester lipase in a syndrome of diabetes and pancreatic exocrine dysfunction. *Journal of Biological Chemistry*, **2014**, 289, 29097-111	5.4	24
47	Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. *JAMA - Journal of the American Medical Association*, **2014**, 311, 2305-14	27.4	164

46	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, 801-5	3.9	2
45	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. *Nature Genetics*, **2013**, 45, 1380-5	36.3	103
44	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. *Pediatric Diabetes*, **2013**, 14, 466-72	3.6	14
43	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	3.8	31
42	SUMOylation of pancreatic glucokinase regulates its cellular stability and activity. *Journal of Biological Chemistry*, **2013**, 288, 5951-62	5.4	25
41	A novel GATA6 mutation in a child with congenital heart malformation and neonatal diabetes. *Clinical Case Reports (discontinued)*, **2013**, 1, 86-90	0.7	8
40	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. *Pediatric Diabetes*, **2012**, 13, 155-62	3.6	15
39	GCK-MODY diabetes associated with protein misfolding, cellular self-association and degradation. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2012**, 1822, 1705-15	6.9	12
38	Exome sequencing and genetic testing for MODY. *PLoS ONE*, **2012**, 7, e38050	3.7	76
37	The role of pancreatic imaging in monogenic diabetes mellitus. *Nature Reviews Endocrinology*, **2011**, 8, 148-59	15.2	28
36	Role of molecular genetics in transforming diagnosis of diabetes mellitus. *Expert Review of Molecular Diagnostics*, **2011**, 11, 313-20	3.8	48
35	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. *Journal of Biological Chemistry*, **2011**, 286, 34593-605	5.4	58
34	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	11.6	379
33	Pancreatic function in carboxyl-ester lipase knockout mice. *Pancreatology*, **2010**, 10, 467-76	3.8	18
32	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. *European Journal of Pediatrics*, **2010**, 169, 207-13	4.1	19
31	Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. *Human Genetics*, **2010**, 127, 55-64	6.3	44
30	Diagnostic screening of MODY2/GCK mutations in the Norwegian MODY Registry. *Pediatric Diabetes*, **2008**, 9, 442-9	3.6	36
29	Mutations in the insulin gene can cause MODY and autoantibody-negative type 1 diabetes. *Diabetes* , **2008**, 57, 1131-5	0.9	154

28	Pancreatic lipomatosis is a structural marker in nondiabetic children with mutations in carboxyl-ester lipase. *Diabetes*, **2007**, 56, 444-9	0.9	74
27	Allosteric activation of human glucokinase by free polyubiquitin chains and its ubiquitin-dependent cotranslational proteasomal degradation. *Journal of Biological Chemistry*, **2007**, 282, 22757-64	5.4	29
26	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. *Diabetes*, **2007**, 56, 3112-7	0.9	37
25	From clinicogenetic studies of maturity-onset diabetes of the young to unraveling complex mechanisms of glucokinase regulation. *Diabetes*, **2006**, 55, 1713-22	0.9	61
24	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. *Diabetes*, **2006**, 55, 1899-903	0.9	26
23	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. *New England Journal of Medicine*, **2006**, 355, 467-77	59.2	740
22	Mutations in the CEL VNTR cause a syndrome of diabetes and pancreatic exocrine dysfunction. *Nature Genetics*, **2006**, 38, 54-62	36.3	246
21	Kir6.2 mutations causing neonatal diabetes provide new insights into Kir6.2-SUR1 interactions. *EMBO Journal*, **2005**, 24, 2318-30	13	56
20	Functional dissection of the HNF-1alpha transcription factor: a study on nuclear localization and transcriptional activation. *DNA and Cell Biology*, **2005**, 24, 661-9	3.6	18
19	Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. *Diabetes*, **2004**, 53, 2713-8	0.9	314
18	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, 1838-49	59.2	930
17	Enlarged nephrons and severe nondiabetic nephropathy in hepatocyte nuclear factor-1beta (HNF-1beta) mutation carriers. *Kidney International*, **2003**, 64, 793-800	9.9	14
16	Hepatocyte nuclear factor-1 alpha gene mutations and diabetes in Norway. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 920-31	5.6	63
15	Permanent neonatal diabetes caused by glucokinase deficiency: inborn error of the glucose-insulin signaling pathway. *Diabetes*, **2003**, 52, 2854-60	0.9	150
14	Neonatal diabetes mellitus due to complete glucokinase deficiency. *New England Journal of Medicine*, **2001**, 344, 1588-92	59.2	350
13	MODY associated with two novel hepatocyte nuclear factor-1alpha loss-of-function mutations (P112L and Q466X). *Biochemical and Biophysical Research Communications*, **2000**, 279, 792-8	3.4	25
12	Sequence analysis of the zebrafish hox-B5/B6 region. *Biochimica Et Biophysica Acta Gene Regulatory Mechanisms*, **1993**, 1173, 102-6		8
11	Structure and early embryonic expression of the zebrafish engrailed-2 gene. *Mechanisms of Development*, **1992**, 39, 51-62	1.7	44

10	Homeobox sequences of Atlantic salmon (Salmo salar) and zebrafish (Brachydanio rerio). *Aquaculture*, **1990**, 85, 51-60	4.4	2
9	A zebrafish homologue of the murine Hox-2.1 gene. *FEBS Letters*, **1988**, 230, 25-30	3.8	20
8	A zebrafish engrailed-like homeobox sequence expressed during embryogenesis. *FEBS Letters*, **1988**, 231, 355-60	3.8	50
7	In situ hybridization patterns of zebrafish homeobox genes homologous to Hox-2.1 and En-2 of mouse. *Biochemical and Biophysical Research Communications*, **1988**, 157, 426-32	3.4	20
6	Structure and neural expression of a zebrafish homeobox sequence. *Gene*, **1988**, 73, 33-46	3.8	17
5	Primary structure, developmentally regulated expression and potential duplication of the zebrafish homeobox gene ZF-21. *Nucleic Acids Research*, **1988**, 16, 9097-111	20.1	31
4	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
3	Genome-wide association meta-analysis of childhood and adolescent internalising symptoms		3
2	Characterization of the genetic architecture of BMI in infancy and early childhood reveals age-specific effects and implicates pathways involved in Mendelian obesity		1
1	Developmental milestones in early childhood and genetic liability to neurodevelopmental disorders. *Psychological Medicine*,1-9	6.9	0