Curriculum Vitae and Bibliography - Pål Rasmus Njølstad MD PhD

PERSONAL: INFORMATION

Male, Norwegian.Widower (Helga Salvesen), four boys.
Addresses: Dep. of Clinical Science, Univ. of Bergen, Norway; Metabolism Program, Broad Institute of Harvard and MIT, Cambridge, MA, USA. e-mail: pal.njolstad@uib.no, njolstad@broadinstitute.org; tel +47-48141578; ORCID 0000-0003-0304-6728. www.diabetes.uib/diabetes

CURRENT POSITIONS

2017-	**Head of Department**	Dep. of Clinical Science, Univ. of Bergen (350 empl)
2013-	**Visiting professor**	Broad Inst. of Harvard and MIT
2012-	**Center leader**	Center for Diabetes Research, Univ. of Bergen (70 empl)
2000-	Professor (full)	Univ. of Bergen
1999-	Consultant	Pediatric endocrinology, Haukeland Univ. Hospital

EDUCATION

1997	Specialist	Pediatrics (EU requirements)
1989	PhD	University of Bergen (molecular biology)
1986	MD	University of Bergen, Norway

PREVIOUS POSITIONS AND MOBILITY

2018-2019	Visiting professor	Genetic endocrinology	Mass. General Hospital, Boston
2018-2019	Visiting professor	Genetic epidemiology	Broad Inst. of Harvard and MIT
2012-2013	Visiting scientist	Molecular genetics	Broad Inst. of Harvard and MIT
2004-2005	Visiting professor	Cell biology, physiology	Harvard Medical School, Boston
1998-1999	Visiting scholar	Diabetes genetics	Univ. of Chicago, Chicago
1992-1997	Residency, fellow	Pediatrics, ped endocrinology	Haukeland Univ. Hospital

MAJOR INSTITUTIONAL RESPONSIBILITIES

2016-	Member	**Norwegian Society of Science and Letters (Medicine)**
2016-	Steering committee	Nordic Society for Precision Medicine
2016-17	Member	Health Data Task Force (Helsedatautvalget, Health Dep., Norway)
2012-17	Leader	Organizing building of 1200 sqm. in new university hospital
2012-14	President	EASD-Study Group for the Genetics of Diabetes
1999-07	Leader	Norwegian Study Group for Childhood Diabetes
1999-12	Leader	Diab. Research Group, Univ. of Bergen/Haukeland Univ. Hosp.

COMMISSIONS OF TRUST

2014-16	European Union (ERC panel)	Research grants (200 biannually)
2012-18	Novo Nordisk Fonden	Research grants (300 yearly)
2003-18	Helse Vest, Helse Sørøst	Research grants, leader (100 yearly)
2007-	Helse SørØst (Akershus)	Research grants (10 yearly)
2006-	Lund, Harvard, Exeter, Oslo, Israel Univ.	Professorships
2007 & 10	Univ. of Chicago, USA	Pilot and Feasibility Grant
2008 & 10	Diabetes UK	Research grants
1998-	Ad hoc reviewer *New Engl J Med*, *Lancet*, *Nat Med*, *Nat Commun*, *Lancet Diab & Met*, PNAS, *Hum Molec Genet*, *Diabetes*, *J Clin Endo Met*, *Diabetologia*, *Diabetes Care*, *Diabetes Med*, *Pediatrics*, *J Pediatrics*, *Arch Dis Child*, *Acta Ped*, *Am J Med Genet*, *Am J Hum Genet*, *PLoS Med*, *PLoS One*	

MEMBERSHIPS SCIENTIFIC SOCIETIES

Norwegian Medical Association, EASD, ADA, ASHG, ESPE, ISPAD, EASD-SGGD, T2GENES, GoT2D, EGG

FELLOWSHIPS AND AWARDS

2020 Senior Research Prize, Westen Norway Health Trust (Helse Vest), Norway
2002-18 **ERC Advanced Grant** (Novel Tools for Early Childhood Predisposition to Obesity and Diabetes)
2018 The Swedish Child Diabetes Foundation's **Johnny Ludvigsson-Prize** for excellent research in childhood and adolescent diabetes
2017 Norwegian Association of Diabetes Research Prize (first time in Norway)
2010 **Novo Nordic Research Award and Lecture**
2008 Research Group of the Year, Faculty of Medicine, Univ. of Bergen
2006 **Diabetes Program at Lund Univ.** Price to Younger Diabetes Scientist
2004 European Society for Pediatric Endocrinology Sabbatical Leave Program (Boston, USA)
2004 **Fulbright Foundation Award** for overseas studies (Boston, USA)
2001, 2006, 2007 Best Scientific Publication Award at Faculty of Medicine, Univ. of Bergen
1998 **Fulbright Foundation Award** for overseas studies (Chicago, USA)
1998 Unger Vetlesen Foundation Award for overseas studies (Chicago, USA)

SUPERVISION OF GRADUATE STUDENTS AND POSTDOCTORAL FELLOWS

Completed 18 PhDs (main supervisor for 80%), 9 MSc, 10 post docs
Ongoing 7 PhDs (main supervisor 50%), 4 post docs

MENTORING OF STUDENTS/FELLOWS

Present (highest ranked) positions for previous students/fellows

Full professor	Lise Bjørkhaug, Jørn Sagen, Helge Ræder, Stefan Johansson, Mette Vesterhus
Ass. professor	Ingvild Aukrust
Consultant physician	Erling Tjora, Henrik Irgens
Academic leader	Vice dean Helge Ræder; senior researchers Jens Hertel, Bente Johansson, and Karianne Fjeld; department directors Øyvind Helgeland and Kishan Chudasama

TEACHING ACTIVITIES

Since 2000, I regularly teach MD students and PhD students (lectures, group work, bed-side teaching) as well as pediatricians and internists for their continuing education (lectures), and have given more than 200 invited presentations at various meetings and courses in Norway.

MOST IMPORTANT ONGOING COLLABORATIONS

Prof. **Graeme I. Bell** PhD, Univ. of Chicago, Chicago, IL, USA, Profs. **C. Ronald Kahn** MD and **Rohit N. Kulkarn** MD PhD, Joslin Diabetes Center, Harvard Medical School, MA, USA, Prof. **Andrew T. Hattersley** MD PhD, and Ass. Prof. **Rachel Freathy** PhD, Exeter Univ., Exeter, UK, Prof. **Anna L. Gloyn** PhD and Prof. **Mark I. McCarthy** MD PhD, Oxford Univ.; Prof. **José Florez** MD PhD, Massachusetts General Hospital, Boston, MA; Prof. **Joel Hirschhorn**, Boston Children´s Hospital, Harvard Medical School; Prof. **V. Mohan**, Madras Diabetes Center, Chennai, India.

Ole A. Andreassen, **Knut Dahl-Jørgensen**, **Geir Joner**, **Torild Skrivarhaug**, Univ. Oslo; **Camilla Stoltenberg**, **Per Magnus**, **Lars Christian Stene**, NIPH, Oslo; **Bjørn Olav Åsvold, Kristian Hveem**, HUNT/NTNU, Trondheim; many physicians and researchers at Univ. of Bergen and Haukeland Univ. Hospital, Bergen.

ORGANIZATION OF INTERNATIONAL CONFERENCES IN THE FIELD AND SCIENTIFIC MEETINGS

2020 *Prediabetes and obesity - who to treat and how?* Univ. Bergen/Univ. Lund. Norway (prog. committee)
2019 *Diabetes Research Conference*. Norway (program committee)
2018 *Nordic Society for Human Genetics and Precision Medicine Meeting*, Malmø (program committee)
2015 *Bioinformatics in Diabetes and Obesity.* Norway (chairman)

2011 *The Genetics of Diabetes in the post-GWAS Area* (EASD-SGGD). Slovak Republic (scient. board)
2009 *The Genotypes and Phenotypes of Diabetes*: *Joint Meeting,* EASD-Study Group on Genetics of Diabetes & Scandinavian Society for the Study of Diabetes Annual Meeting, Norway (chairman)
2007 *Scandinavian Society for the Study of Diabetes Annual Meeting*, Denmark (steering committee)
2004 *EASD-Study Group on Genetics of Diabetes Annual Meeting*, Spain (scientific board)
2003 *International Beta-Cell Meeting,* Norway (chairman)

INVITED PRESENTATIONS TO INTERNATIONAL CONFERENCES AND OTHER MEETINGS

2021 Annual Meeting, Scandinavian Society for the Study of Diabetes, Trondheim
2019 International Society of Pediatric and Adolescent Diabetes, Boston, MA, USA
2019 Joslin Diabetes Center, Harvard Medical School, Boston, MA, USA
2019 European Society for Pediatric Endocrinology, Vienna, Austria
2018 Finnish Institute for Molecular Medicine, Helsinki, Finland
2018 The Swedish Child Diabetes Foundation's Johnny Ludvigsson Award Lecture, Turku, Finland
2017 Norwegian Association of Diabetes Research Award Lecture, Oslo, Norway
2017 Study Group on the Genetics of Diabetes, Leiden, Holland
2016 Banbury Meeting - precision medicine initiative in the Nordic region, Cold Spring Harbor, NY, USA
2016 Meeting, Early Growth and Genetics, Copenhagen, Denmark
2016 Annual Meeting, International Diabetes Federation, Valencia, Spain
2016 ISPAD Science School, Hønefoss, Norway
2015 Nordic Health Research and Innovation Networks Annual Meeting, Bergen, Norway
2015 Study Group on the Genetics of Diabetes, Krakow, Poland
2015 Annual Meeting Diabetes Research at Karolinska, Stockholm, Sweden
2014 Endocrine Society, Chicago, IL, USA
2013 International Diabetes Federation, Gothenburg, Sweden
2013 4th. Nordic Bioscience Conference, Copenhagen, Denmark
2013 Broad Institute of Harvard and MIT, Cambridge, MA, USA
2012 Endocrine Society, Reykjavik, Iceland
2011 Annual Meeting, Europediatrics, Vienna, Austria
2011 The Genetics of Diabetes in the post-GWAS Area (EASD-SGGD), Slovak Republic
2011 Annual Meeting, International Diabetes Federation, Dubai, Arabic Emirates
2010 70th Scientific Sessions, American Diabetes Association, New Orleans, USA
2010 Annual Meeting, European Society for Human Genetics (**opening lecture**), Sweden
2010 Annual Meeting, European Society for Pediatric Endocrinology, Czech Republic
2010 6th International Pediatric Endocrinology Symposium, Germany
2010 EMBO Workshop on Disease, Development, and Stem Cells of the Pancreas, Sweden
2008 Workshop on Exocrine Dysfunction in Diabetes, Germany
2008 Annual Meeting of European Congress of Endocrinology, Germany
2007 Monogenic Diabetes Diagnostics Meeting, France
2007 ISPAD Science School, Denmark
2007 Lund University, Malmø, Sweden
2007 Danish Diabetes Registry Annual Meeting, Copenhagen, Denmark
2006 Genomics of Hyperglycemia (EASD-SGGD), Denmark
2006 Annual Meeting Diabetes, Obesity and Hypertension, Germany
2006 Congenital Hyperinsulinism, Philadelphia, USA
2005 Grand Round, Joslin Diabetes Center, Boston, MA, USA
2005 University of Chicago, Chicago, IL, USA
2004 Grand Round, Rhode Island Hospital, RI, USA
2004 MODY in Malaga (EASD-SGGD), Spain
2002 Civil Hospital, Malaga, Spain
2002 Clinical Research Center, Malmø, Sweden
2001 Showa-mashi, Maebashi City, Gunma, Japan

2001 Annual Meeting, ISPAD, Siena, Italy

ON-GOING GRANTS of around 20 M € in funding 1997-date

Project Title	*Funding source*	*Amount (Euros)*	*Period*	*Role*
Childhood predisposition to obesity	European Research Council AdG incl. add-on from MED and UiB	5.0M	2012-18	PI
Towards personalized therapies in diabetes	Novo Nordisk Fonden	1.4M	2020-24	PI
Genomics of childhood diabetes and precision medicine	Health Authorities of Western Norway	0.5M	2019-21	PI
PERSON-MED-DIA: precision medicine in diabetes	Health Authorities of Western Norway	2.0M	2015-22	PI
Exome sequencing in diabetes	Research Council of Norway	1.2M	2016-19	PI
GWAS in Norw. Mother Child Cohort Study for better health	Bergen Research Foundation	1.0M	2016-18	PI
Center for Diabetes Research	Stiftelsen Kristian Gerhard Jebsen	2.5M	2012-18	PI
HARVEST genotyping 60 000 samples from Mother Child Cohort of Norway	Research Council of Norway	2.5M	2014-16	Co-PI

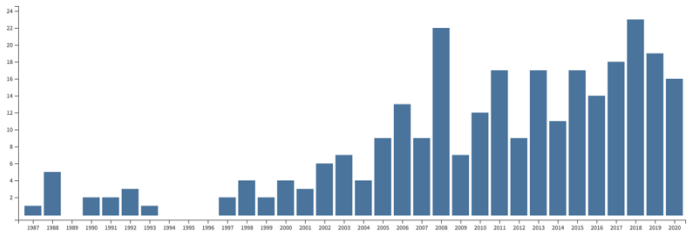
GRANTED PATENTS

Publication #WO/2007/063405; International Filing Date: 01.12.06; Title: Diagnosis and treatment of exocrine pancreatic dysfunction and diabetes.

BIBLIOGRAPHY 1987-2020 IN NUMBERS

Web of Science: *h*-index 50; 9551 citations. Of my 10 most cited papers, I am first or last author on 50%.

Number of publications per year 1997-2021



Number of citations per year 1997-2021

PubMed: 220 listed papers incl. *Nature* (3), *N Engl J Med* (3), *Nat Genet* (7), *Nat Commun* (3), *JAMA* (2), *Lancet D&E* (1), *Nat Rev Endocrinol* (2), *Am J Hum Genet* (3), *J Clin Invest* (1), *PNAS* (2), Diabetes (17), *J Biol Chem* (6), *PLoS Med* (3), *EMBO J* (3), and *Bioinformatics* (1). Of these, **80 as first, second, second last, last and/or corresponding author.** 16 PubMed papers in 2020.
Popular science and papers in Norwegian: 100

EXTRACURRICULUM ACTIVITY

Rank as captain in the Norwegian Navy
Competitive large yacht regatta sailing (short-handed, 2 sailors): At best, ranked no 21 in Norway (2019)
Deck Office Class 5 Pleasure Craft certificate (sailing and navigation world-around, 80 feet vessel) (2017)

Publications Pål Rasmus Njølstad 1987-2021

PubMed-listed papers

1: Hannigan LJ, Askeland RB, Ask H, Tesli M, Corfield E, Ayorech Z, Helgeland Ø, Magnus P, **Njølstad PR**, Øyen AS, Stoltenberg C, Andreassen OA, Davey Smith G, Reichborn-Kjennerud T, Havdahl A. Genetic Liability for Schizophrenia and Childhood Psychopathology in the General Population. *Schizophr Bull*. 2021 Feb 9:sbaa193. PMID: 33561255.

2: Lavrichenko K, Helgeland Ø, **Njølstad PR**, Jonassen I, Johansson S. SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. *Bioinformatics*. 2021 Jan 18:btab028. PMID: 33459766.

3: Sole-Navais P, Bacelis J, Helgeland Ø, Modzelewska D, Vaudel M, Flatley C, Andreassen O, **Njølstad PR**, Muglia LJ, Johansson S, Zhang G, Jacobsson B. Autozygosity mapping and time-to-spontaneous delivery in Norwegian parent-offspring trios. *Hum Mol Genet*. 2021 Feb 4;29(23):3845-3858. PMID: 33291140.

4: Laisk T, Soares ALG, Ferreira T, Painter JN, Censin JC, Laber S, Bacelis J, Chen CY, Lepamets M, Lin K, Liu S, Millwood IY, Ramu A, Southcombe J, Andersen MS, Yang L, Becker CM, Børglum AD, Gordon SD, Bybjerg-Grauholm J, Helgeland Ø, Hougaard DM, Jin X, Johansson S, Juodakis J, Kartsonaki C, Kukushkina V, Lind PA, Metspalu A, Montgomery GW, Morris AP, Mors O, Mortensen PB, **Njølstad PR**, Nordentoft M, Nyholt DR, Lippincott M, Seminara S, Salumets A, Snieder H, Zondervan K, Werge T, Chen Z, Conrad DF, Jacobsson B, Li L, Martin NG, Neale BM, Nielsen R, Walters RG, Granne I, Medland SE, Mägi R, Lawlor DA, Lindgren CM. The genetic architecture of sporadic and multiple consecutive miscarriage. *Nat Commun*. 2020 Nov 25;11(1):5980. doi: PMID: 33239672.

5: Bowman P, Mathews F, Barbetti F, Shepherd MH, Sanchez J, Piccini B, Beltrand J, Letourneau-Freiberg LR, Polak M, Greeley SAW, Rawlins E, Babiker T, Thomas NJ, De Franco E, Ellard S, Flanagan SE, Hattersley AT; Neonatal Diabetes International Collaborative Group (**Njølstad PR**). Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated ABCC8 Permanent Neonatal Diabetes. *Diabetes Care*. 2021 Jan;44(1):35-42. PMID: 33184150.

6: Cheesman R, Eilertsen EM, Ahmadzadeh YI, Gjerde LC, Hannigan LJ, Havdahl A, Young AI, Eley TC, **Njølstad PR**, Magnus P, Andreassen OA, Ystrom E, McAdams TA. 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*). BMC Med*. 2020 Oct 27;18(1):284. PMID: 33106172.

7: Vogelezang S, Bradfield JP, Ahluwalia TS, Curtin JA, Lakka TA, Grarup N, Scholz M, van der Most PJ, Monnereau C, Stergiakouli E, Heiskala A, Horikoshi M, Fedko IO, Vilor-Tejedor N, Cousminer DL, Standl M, Wang CA, Viikari J, Geller F, Íñiguez C, Pitkänen N, Chesi A, Bacelis J, Yengo L, Torrent M, Ntalla I, Helgeland Ø, Selzam S, Vonk JM, Zafarmand MH, Heude B, Farooqi IS, Alyass A, Beaumont RN, Have CT, Rzehak P, Bilbao JR, Schnurr TM, Barroso I, Bønnelykke K, Beilin LJ, Carstensen L, Charles MA, Chawes B, Clément K, Closa-Monasterolo R, Custovic A, Eriksson JG, Escribano J, Groen-Blokhuis M, Grote V, Gruszfeld D, Hakonarson H, Hansen T, Hattersley AT, Hollensted M, Hottenga JJ, Hyppönen E, Johansson S, Joro R, Kähönen M, Karhunen V, Kiess W, Knight BA, Koletzko B, Kühnapfel A, Landgraf K, Langhendries JP, Lehtimäki T, Leinonen JT, Li A, Lindi V, Lowry E, Bustamante M, Medina-Gomez C, Melbye M, Michaelsen KF, Morgen CS, Mori TA, Nielsen TRH, Niinikoski H, Oldehinkel AJ, Pahkala K, Panoutsopoulou K, Pedersen O, Pennell CE, Power C, Reijneveld SA, Rivadeneira F, Simpson A, Sly PD, Stokholm J, Teo KK, Thiering E, Timpson NJ, Uitterlinden AG, van Beijsterveldt CEM, van Schaik BDC, Vaudel M, Verduci E, Vinding RK, Vogel M, Zeggini E, Sebert S, Lind MV, Brown CD, Santa-Marina L, Reischl E, Frithioff-Bøjsøe C, Meyre D, Wheeler E, Ong K, Nohr EA, Vrijkotte TGM, Koppelman GH, Plomin R, **Njølstad PR**, Dedoussis GD, Froguel P, Sørensen TIA, Jacobsson B, Freathy RM, Zemel BS, Raitakari O, Vrijheid M, Feenstra B, Lyytikäinen LP,

Publications Pål Rasmus Njølstad 1987-2021

Snieder H, Kirsten H, Holt PG, Heinrich J, Widén E, Sunyer J, Boomsma DI, Järvelin MR, Körner A, Davey Smith G, Holm JC, Atalay M, Murray C, Bisgaard H, McCarthy MI; Early Growth Genetics Consortium, Jaddoe VWV, Grant SFA, Felix JF. Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. *PLoS Genet*. 2020 Oct 12;16(10):e1008718. PMID: 33045005.

8: Althari S, Najmi LA, Bennett AJ, Aukrust I, Rundle JK, Colclough K, Molnes J, Kaci A, Nawaz S, van der Lugt T, Hassanali N, Mahajan A, Molven A, Ellard S, McCarthy MI, Bjørkhaug L, **Njølstad PR**, Gloyn AL. Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. *Am J Hum Genet*. 2020 Oct 1;107(4):670-682. PMID: 32910913.

9: Velasco K, St-Louis JL, Hovland HN, Thompson N, Ottesen Å, Choi MH, Pedersen L, **Njølstad PR**, Arnesen T, Fjeld K, Aukrust I, Myklebust LM, Molven A. Functional evaluation of 16 SCHAD missense variants: Only amino acid substitutions causing congenital hyperinsulinism of infancy lead to loss-of-function phenotypes in vitro. *J Inherit Metab Dis.* 2021 Jan;44(1):240-252. PMID: 32876354.

10: Smajlagić D, Lavrichenko K, Berland S, Helgeland Ø, Knudsen GP, Vaudel M, Haavik J, Knappskog PM, **Njølstad PR**, Houge G, Johansson S. Population prevalence and inheritance pattern of recurrent CNVs associated with neurodevelopmental disorders in 12,252 newborns and their parents*. Eur J Hum Genet*. 2021 Jan;29(1):205-215. PMID: 32778765.

11: Winnay JN, Solheim MH, Sakaguchi M, **Njølstad PR**, Kahn CR. Inhibition of the PI 3-kinase pathway disrupts the unfolded protein response and reduces sensitivity to ER stress-dependent apoptosis. *FASEB J.* 2020 Sep;34(9):12521-12532. PMID: 32744782.

12: Lund-Blix NA, Tapia G, Mårild K, Brantsaeter AL, **Njølstad PR**, Joner G, Skrivarhaug T, Størdal K, Stene LC. Maternal and child gluten intake and association with type 1 diabetes: The Norwegian Mother and Child Cohort Study. *PLoS Med.* 2020 Mar 2;17(3):e1003032. PMID: 32119659.

13: Malikova J, Kaci A, Dusatkova P, Aukrust I, Torsvik J, Vesela K, Kankova PD, **Njølstad PR**, Pruhova S, Bjørkhaug L. Functional analyses of HNF1A-MODY variants refine the interpretation of identified sequence variants. *J Clin Endocrinol Metab.* 2020 Feb 4. PMID: 32017842.

14: Fjeld K, Masson E, Lin JH, Michl P, Stokowy T, Gravdal A, El Jellas K, Steine SJ, Hoem D, Johansson BB, Dalva M, Ruffert C, Zou WB, Li ZS, **Njølstad PR**, Chen JM, Liao Z, Johansson S, Rosendahl J, Férec C, Molven A. Characterization of CEL-DUP2: Complete duplication of the carboxyl ester lipase gene is unlikely to influence risk of chronic pancreatitis. *Pancreatology.* 2020 Jan 20. PMID: 32007358.

15: Dalva M, Lavik IK, El Jellas K, Gravdal A, Lugea A, Pandol SJ, **Njølstad PR**, Waldron RT, Fjeld K, Johansson BB, Molven A. Pathogenic Carboxyl Ester Lipase (CEL) Variants Interact with the Normal CEL Protein in Pancreatic Cells. *Cells.* 2020 Jan 18;9(1). PMID: 31963687.

16: Svalastoga P, Sulen Å, Fehn JR, Aukland SM, Irgens H, Sirnes E, Fevang SKE, Valen E, Elgen IB, **Njølstad PR**. Intellectual Disability in K(ATP) Channel Neonatal Diabetes. *Diabetes Care.* 2020 Mar;43(3):526-533. PMID: 31932458.

17: Bjune JI, Dyer L, Røsland GV, Tronstad KJ, **Njølstad PR**, Sagen JV, Dankel SN, Mellgren G. The homeobox factor Irx3 maintains adipogenic identity. *Metabolism.* 2020 Feb;103:154014. PMID: 31751577.

Publications Pål Rasmus Njølstad 1987-2021

18: Helgeland Ø, Vaudel M, Juliusson PB, Lingaas Holmen O, Juodakis J, Bacelis J, Jacobsson B, Lindekleiv H, Hveem K, Lie RT, Knudsen GP, Stoltenberg C, Magnus P, Sagen JV, Molven A, Johansson S, **Njølstad PR**. Genome-wide association study reveals dynamic role of genetic variation in infant and early childhood growth. *Nat Commun.* 2019 Oct 1;10(1):4448. PMID: 31575865.

19: Bradfield JP, Vogelezang S, Felix JF, Chesi A, Helgeland Ø, Horikoshi M, Karhunen V, Lowry E, Cousminer DL, Ahluwalia TS, Thiering E, Boh ET, Zafarmand MH, Vilor-Tejedor N, Wang CA, Joro R, Chen Z, Gauderman WJ, Pitkänen N, Parra EJ, Fernandez-Rhodes L, Alyass A, Monnereau C, Curtin JA, Have CT, McCormack SE, Hollensted M, Frithioff-Bøjsøe C, Valladares-Salgado A, Peralta-Romero J, Teo YY, Standl M, Leinonen JT, Holm JC, Peters T, Vioque J, Vrijheid M, Simpson A, Custovic A, Vaudel M, Canouil M, Lindi V, Atalay M, Kähönen M, Raitakari OT, van Schaik BDC, Berkowitz RI, Cole SA, Voruganti VS, Wang Y, Highland HM, Comuzzie AG, Butte NF, Justice AE, Gahagan S, Blanco E, Lehtimäki T, Lakka TA, Hebebrand J, Bonnefond A, Grarup N, Froguel P, Lyytikäinen LP, Cruz M, Kobes S, Hanson RL, Zemel BS, Hinney A, Teo KK, Meyre D, North KE, Gilliland FD, Bisgaard H, Bustamante M, Bonnelykke K, Pennell CE, Rivadeneira F, Uitterlinden AG, Baier LJ, Vrijkotte TGM, Heinrich J, Sørensen TIA, Saw SM, Pedersen O, Hansen T, Eriksson J, Widén E, McCarthy MI, **Njølstad PR**, Power C, Hyppönen E, Sebert S, Brown CD, Järvelin MR, Timpson NJ, Johansson S, Hakonarson H, Jaddoe VWV; Early Growth Genetics Consortium, Grant SFA. A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. *Hum Mol Genet.* 2019 Oct 1;28(19):3327-3338. PMID: 31504550.

20: Qiao Z, Zheng J, Helgeland Ø, Vaudel M, Johansson S, **Njølstad PR**, Smith GD, Warrington NM, Evans DM. Introducing M-GCTA a Software Package to Estimate Maternal (or Paternal) Genetic Effects on Offspring Phenotypes. *Behav Genet*. 2020 Jan;50(1):51-66. PMID: 31493278.

21: Liu X, Helenius D, Skotte L, Beaumont RN, Wielscher M, Geller F, Juodakis J, Mahajan A, Bradfield JP, Lin FTJ, Vogelezang S, Bustamante M, Ahluwalia TS, Pitkänen N, Wang CA, Bacelis J, Borges MC, Zhang G, Bedell BA, Rossi RM, Skogstrand K, Peng S, Thompson WK, Appadurai V, Lawlor DA, Kalliala I, Power C, McCarthy MI, Boyd HA, Marazita ML, Hakonarson H, Hayes MG, Scholtens DM, Rivadeneira F, Jaddoe VWV, Vinding RK, Bisgaard H, Knight BA, Pahkala K, Raitakari O, Helgeland Ø, Johansson S, **Njølstad PR**, Fadista J, Schork AJ, Nudel R, Miller DE, Chen X, Weirauch MT, Mortensen PB, Børglum AD, Nordentoft M, Mors O, Hao K, Ryckman KK, Hougaard DM, Kottyan LC, Pennell CE, Lyytikainen LP, Bønnelykke K, Vrijheid M, Felix JF, Lowe WL Jr, Grant SFA, Hyppönen E, Jacobsson B, Jarvelin MR, Muglia LJ, Murray JC, Freathy RM, Werge TM, Melbye M, Buil A, Feenstra B. Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. *Nat Commun*. 2019 Sep 2;10(1):3927. PMID: 31477735.

22: Sánchez LFH, Burger B, Horro C, Fabregat A, Johansson S, **Njølstad PR**, Barsnes H, Hermjakob H, Vaudel M. PathwayMatcher: proteoform-centric network construction enables fine-granularity multiomics pathway mapping. *Gigascience*. 2019 Aug 1;8(8). PMID: 31363752.

23: Tapia G, Mortimer G, Ye J, Gillard BT, Chipper-Keating S, Mårild K, Viken MK, Lie BA, Joner G, Skrivarhaug T, **Njølstad PR**, Størdal K, Gillespie KM, Stene LC. Maternal microchimerism in cord blood and risk of childhood-onset type 1 diabetes. *Pediatr Diabetes*. 2019 Sep;20(6):728-735. PMID: 31173445.

24: **Njølstad PR**, Andreassen OA, Brunak S, Børglum AD, Dillner J, Esko T, Franks PW, Freimer N, Groop L, Heimer H, Hougaard DM, Hovig E, Hveem K, Jalanko A, Kaprio J, Knudsen GP, Melbye M, Metspalu A, Mortensen PB, Palmgren J, Palotie A, Reed W, Stefánsson H, Stitziel NO, Sullivan PF, Thorsteinsdóttir U, Vaudel M, Vuorio E, Werge T, Stoltenberg C, Stefánsson K. Roadmap for a precision-medicine initiative in the Nordic region. *Nat Genet*. 2019 Jun;51(6):924-930. PMID: 30988515.

25: Warrington NM, Beaumont RN, Horikoshi M, Day FR, Helgeland Ø, Laurin C, Bacelis J, Peng S, Hao K, Feenstra B, Wood AR, Mahajan A, Tyrrell J, Robertson NR, Rayner NW, Qiao Z, Moen GHØ, Vaudel M, Marsit CJ, Chen J, Nodzenski M, Schnurr TM, Zafarmand MH, Bradfield JP, Grarup N, Kooijman NM, Li-Gao R, Geller F, Ahluwalia TS, Paternoster L, Rueedi R, Huikari V, Hottenga JJ, Lyytikäinen LP, Cavadino A, Metrustry S, Cousminer DL, Wu Y, Thiering E, Wang CA, Have CT, Vilor-Tejedor N, Joshi PK, Painter JN, Ntalla I, Myhre R, Pitkänen N, van Leeuwen EM, Joro R, Lagou V, Richmond RC, Espinosa A, Barton SJ, Inskip HM, Holloway JW, Santa-Marina L, Estivill X, Ang Q, Marsh JA, Reichetzeder C, Marullo L, Hocher B, Lunetta CL, Murabito JM, Relton CL, Kogevinas M, Chatzi L, Allard A, Bouchard L, Hivert MF, Zhang G, Muglia LJ, Heikkinen J, Early Growth Genetics (EGG) Consortium, Morgen CS, van Kampen AHC, van Schaik BDC, Mentch FD, Langenberg C, Luan J, Scott RA, Zhao JH, Hemani G, Ring SM, Bennett AJ, Gaulton KJ, Fernandez-Tajes F, van Zuydam NR, Medina-Gomez C, de Haan HG, Rosendaal FR, Kutalik Z, Marques-Vidal P, Das S, Willemsen G, Mbarek H, Müller-Nurasyid M, Standl M, Appel EVR, Fonvig CE, Trier C, van Beijsterveldt CEM, Murcia M, Bustamante M, Bonas-Guarch S, Hougaard DM, Mercader JM, Linneberg A, Schraut KE, Lind PA, Medland DSE, Shields BM, Knight BA, Chai JF, Panoutsopoulou K, Bartels M, Sánchez F, Stokholm J, Torrents D, Vinding RK, Willems SM, Atalay M, Chawes BL, Kovacs P, Prokopenko I, Tuke MA, Yaghootkar H, Ruth KS, Jones SE, Loh PR, Murray A, Weedon MN, Tönjes A, Stumvoll M, Michaelsen KF, Eloranta EM, Lakka TA, van Duijn CM, Kiess W, Körner A, Niinikoski H, Pahkala K, Raitakari OY, Jacobsson B, Zeggini E, Dedoussis GV, Teo YY, Saw SM, Montgomery GW, Campbell H, Wilson GF, Vrijkotte TGM, Vrijheid M, de Geus EJCN, Hayes EG, Kadarmideen HN, Holm JC, Beilin LJ, Pennell CE, Heinrich J, Adair LS, Borja JB, Mohlke KL, Eriksson JG, Widén EE, Hattersley AT, Spector TD, Kähönen M, Viikari JS, Lehtimäki T, Boomsma DI, Sebert S, Vollenweider P Sørensen YIA, Bisgaard H, Bønnelykke K, Murray JC, Melbye M, Nohr EA, Mook-Kanamori DO, Rivadeneira F, Hofman A, Felix JF, Jaddoe VWV, Hansen T, Pisinger C, Vaag AA, Pedersen O, Uitterlinden AG, Järvelin MR, Power C, Hyppönen E, Scholtens DM, Lowe Jr WL, Smith GD, Timpson NJ, Morris AP, Wareham NJ, Hakonarson H , Grant SFA, Frayling TM, Lawlor DA, **Njølstad PR**, Johansson S, Ong K,McCarthy MI, Perry JRB, Evans DM, Freathy RM. Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. *Nat Genet*. 2019, 2019 May;51(5):804-814. PMID: 31043758.

26: Tapia G, Mårild K, Dahl SR, Lund-Blix NA, Viken MK, Lie BA, **Njølstad PR**, Joner G, Skrivarhaug T, Cohen AS, Størdal K, Stene LC. Maternal and Newborn Vitamin D-Binding Protein, Vitamin D Levels, Vitamin D Receptor Genotype, and Childhood Type 1 Diabetes. *Diabetes Care*. 2019 Apr;42(4):553-559. PMID: 30692241.

27: Bjune JI, Haugen C, Gudbrandsen O, Nordbø OP, Nielsen HJ, Våge V, **Njølstad PR**, Sagen JV, Dankel SN, Mellgren G. IRX5 regulates adipocyte amyloid precursor protein and mitochondrial respiration in obesity. *Int J Obes (Lond).* 2018 Dec 11. PMID: 30538277.

28: El Jellas K, Johansson BB, Fjeld K, Antonopoulos A, Immervoll H, Choi MH, Hoem D, Lowe ME, Lombardo D, **Njølstad PR**, Dell A, Mas E, Haslam SM, Molven A. 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. *J Biol Chem*. 2018 Dec 14;293(50):19476-19491. PMID: 30315106.

29: Hattersley AT, Greeley SAW, Polak M, Rubio-Cabezas O, **Njølstad PR**, Mlynarski W, Castano L, Carlsson A, Raile K, Chi DV, Ellard S, Craig ME. ISPAD Clinical Practice Consensus Guidelines 2018: The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatr Diabetes*. 2018 Oct;19 Suppl 27:47-63. PMID: 30225972.

30: Kaci A, Keindl M, Solheim MH, **Njølstad PR**, Bjørkhaug L, Aukrust I. The E3 SUMO ligase PIASγ is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1α. *Sci Rep*. 2018 Aug 24;8(1):12780. PMID: 30143652.

31: Magnus MC, Tapia G, Olsen SF, Granstrom C, Mårild K, Ueland PM, Midttun Ø, Svensson J, Johannesen J, Skrivarhaug T, Joner G, **Njølstad PR**, Størdal K, Stene LC. Parental Smoking and Risk of Childhood-onset Type 1 Diabetes. *Epidemiology*. 2018 Nov;29(6):848-856. PMID: 30074542.

32: Engjom T, Kavaliauskiene G, Tjora E, Erchinger F, Wathle G, Lærum BN, **Njølstad PR**, Frøkjær JB, Gilja OH, Dimcevski G, Haldorsen IS. Sonographic pancreas echogenicity in cystic fibrosis compared to exocrine pancreatic function and pancreas fat content at Dixon-MRI. *PLoS One*. 2018 Jul 26;13(7):e0201019. PMID: 30048483.

33: Størdal K, McArdle HJ, Hayes H, Tapia G, Viken MK, Lund-Blix NA, Haugen M, Joner G, Skrivarhaug T, Mårild K, **Njølstad PR**, Eggesbø M, Mandal S, Page CM, London SJ, Lie BA, Stene LC. Prenatal iron exposure and childhood type 1 diabetes. *Sci Rep*. 2018 Jun 13;8(1):9067. PMID: 29899542.

34: Bowman P, Sulen Å, Barbetti F, Beltrand J, Svalastoga P, Codner E, Tessmann EH, Juliusson PB, Skrivarhaug T, Pearson ER, Flanagan SE, Babiker T, Thomas NJ, Shepherd MH, Ellard S, Klimes I, Szopa M, Polak M, Iafusco D, Hattersley AT, **Njølstad PR**; Neonatal Diabetes International Collaborative Group. Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. *Lancet Diabetes Endocrinol*. 2018 Aug;6(8):637-646. PMID: 29880308.

35: Tapia G, Størdal K, Mårild K, Kahrs CR, Skrivarhaug T, **Njølstad PR**, Joner G, Stene LC. Antibiotics, acetaminophen and infections during prenatal and early life in relation to type 1 diabetes. *Int J Epidemiol*. 2018 Oct 1;47(5):1538-1548. PMID: 29868838.

36: Solheim MH, Winnay JN, Batista TM, Molven A, **Njølstad PR**, Kahn CR. Mice carrying a dominant-negative human PI3K mutation are protected from obesity and hepatic steatosis but not diabetes. *Diabetes*. 2018 Jul;67(7):1297-1309. PMID: 29724723.

37: Magnus MC, Olsen SF, Granstrom C, Lund-Blix NA, Svensson J, Johannesen J, Fraser A, Skrivarhaug T, Joner G, **Njølstad PR**, Størdal K, Stene LC. Paternal and maternal obesity but not gestational weight gain is associated with type 1 diabetes. *Int J Epidemiol.* 2018 Apr 1;47(2):417-426. PMID: 29415279.

38: Beaumont RN, Warrington NM, Cavadino A, Tyrrell J, Nodzenski M, Horikoshi M, Geller F, Myhre R, Richmond RC, Paternoster L, Bradfield JP, Kreiner-Møller E, Huikari V, Metrustry S, Lunetta KL, Painter JN, Hottenga JJ, Allard C, Barton SJ, Espinosa A, Marsh JA, Potter C, Zhang G, Ang W, Berry DJ, Bouchard L, Das S; Early Growth Genetics (EGG) Consortium, Hakonarson H, Heikkinen J, Helgeland Ø, Hocher B, Hofman A, Inskip HM, Jones SE, Kogevinas M, Lind PA, Marullo L, Medland SE, Murray A, Murray JC, **Njølstad PR**, Nohr EA, Reichetzeder C, Ring SM, Ruth KS, Santa-Marina L, Scholtens DM, Sebert S, Sengpiel V, Tuke MA, Vaudel M, Weedon MN, Willemsen G, Wood AR, Yaghootkar H, Muglia LJ, Bartels M, Relton CL, Pennell CE, Chatzi L, Estivill X, Holloway JW, Boomsma DI, Montgomery GW, Murabito JM, Spector TD, Power C, Järvelin MR, Bisgaard H, Grant SFA, Sørensen TIA, Jaddoe VW, Jacobsson B, Melbye M, McCarthy MI, Hattersley AT, Hayes MG, Frayling TM, Hivert MF, Felix JF, Hyppönen E, Lowe WL Jr, Evans DM, Lawlor DA, Feenstra B, Freathy RM. 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. *Hum Mol Genet*. 2018 Feb 15;27(4):742-756. PMID: 29309628.

39: Turcot V, Lu Y, Highland HM, Schurmann C, Justice AE, Fine RS, Bradfield JP, Esko T, Giri A, Graff M, Guo X, Hendricks AE, Karaderi T, Lempradl A, Locke AE, Mahajan A, Marouli E, Sivapalaratnam S, Young KL, Alfred T, Feitosa MF, Masca NGD, Manning AK, Medina-Gomez C, Mudgal P, Ng MCY, Reiner AP, Vedantam S, Willems SM, Winkler TW, Abecasis G, Aben KK, Alam DS, Alharthi SE, Allison M, Amouyel P, Asselbergs FW, Auer PL, Balkau B, Bang LE, Barroso I, Bastarache L, Benn M, Bergmann S, Bielak LF, Blüher M, Boehnke M, Boeing H, Boerwinkle E, Böger CA, Bork-Jensen J, Bots ML, Bottinger EP, Bowden DW, Brandslund I, Breen G, Brilliant MH, Broer L, Brumat

M, Burt AA, Butterworth AS, Campbell PT, Cappellani S, Carey DJ, Catamo E, Caulfield MJ, Chambers JC, Chasman DI, Chen YI, Chowdhury R, Christensen C, Chu AY, Cocca M, Collins FS, Cook JP, Corley J, Corominas Galbany J, Cox AJ, Crosslin DS, Cuellar-Partida G, D'Eustacchio A, Danesh J, Davies G, Bakker PIW, Groot MCH, Mutsert R, Deary IJ, Dedoussis G, Demerath EW, Heijer M, Hollander AI, Ruijter HM, Dennis JG, Denny JC, Angelantonio E, Drenos F, Du M, Dubé MP, Dunning AM, Easton DF, Edwards TL, Ellinghaus D, Ellinor PT, Elliott P, Evangelou E, Farmaki AE, Farooqi IS, Faul JD, Fauser S, Feng S, Ferrannini E, Ferrieres J, Florez JC, Ford I, Fornage M, Franco OH, Franke A, Franks PW, Friedrich N, Frikke-Schmidt R, Galesloot TE, Gan W, Gandin I, Gasparini P, Gibson J, Giedraitis V, Gjesing AP, Gordon-Larsen P, Gorski M, Grabe HJ, Grant SFA, Grarup N, Griffiths HL, Grove ML, Gudnason V, Gustafsson S, Haessler J, Hakonarson H, Hammerschlag AR, Hansen T, Harris KM, Harris TB, Hattersley AT, Have CT, Hayward C, He L, Heard-Costa NL, Heath AC, Heid IM, Helgeland Ø, Hernesniemi J, Hewitt AW, Holmen OL, Hovingh GK, Howson JMM, Hu Y, Huang PL, Huffman JE, Ikram MA, Ingelsson E, Jackson AU, Jansson JH, Jarvik GP, Jensen GB, Jia Y, Johansson S, Jørgensen ME, Jørgensen T, Jukema JW, Kahali B, Kahn RS, Kähönen M, Kamstrup PR, Kanoni S, Kaprio J, Karaleftheri M, Kardia SLR, Karpe F, Kathiresan S, Kee F, Kiemeney LA, Kim E, Kitajima H, Komulainen P, Kooner JS, Kooperberg C, Korhonen T, Kovacs P, Kuivaniemi H, Kutalik Z, Kuulasmaa K, Kuusisto J, Laakso M, Lakka TA, Lamparter D, Lange EM, Lange LA, Langenberg C, Larson EB, Lee NR, Lehtimäki T, Lewis CE, Li H, Li J, Li-Gao R, Lin H, Lin KH, Lin LA, Lin X, Lind L, Lindström J, Linneberg A, Liu CT, Liu DJ, Liu Y, Lo KS, Lophatananon A, Lotery AJ, Loukola A, Luan J, Lubitz SA, Lyytikäinen LP, Männistö S, Marenne G, Mazul AL, McCarthy MI, McKean-Cowdin R, Medland SE, Meidtner K, Milani L, Mistry V, Mitchell P, Mohlke KL, Moilanen L, Moitry M, Montgomery GW, Mook-Kanamori DO, Moore C, Mori TA, Morris AD, Morris AP, Müller-Nurasyid M, Munroe PB, Nalls MA, Narisu N, Nelson CP, Neville M, Nielsen SF, Nikus K, **Njølstad PR**, Nordestgaard BG, Nyholt DR, O'Connel JR, O'Donoghue ML, Olde Loohuis LM, Ophoff RA, Owen KR, Packard CJ, Padmanabhan S, Palmer CNA, Palmer ND, Pasterkamp G, Patel AP, Pattie A, Pedersen O, Peissig PL, Peloso GM, Pennell CE, Perola M, Perry JA, Perry JRB, Pers TH, Person TN, Peters A, Petersen ERB, Peyser PA, Pirie A, Polasek O, Polderman TJ, Puolijoki H, Raitakari OT, Rasheed A, Rauramaa R, Reilly DF, Renström F, Rheinberger M, Ridker PM, Rioux JD, Rivas MA, Roberts DJ, Robertson NR, Robino A, Rolandsson O, Rudan I, Ruth KS, Saleheen D, Salomaa V, Samani NJ, Sapkota Y, Sattar N, Schoen RE, Schreiner PJ, Schulze MB, Scott RA, Segura-Lepe MP, Shah SH, Sheu WH, Sim X, Slater AJ, Small KS, Smith AV, Southam L, Spector TD, Speliotes EK, Starr JM, Stefansson K, Steinthorsdottir V, Stirrups KE, Strauch K, Stringham HM, Stumvoll M, Sun L, Surendran P, Swift AJ, Tada H, Tansey KE, Tardif JC, Taylor KD, Teumer A, Thompson DJ, Thorleifsson G, Thorsteinsdottir U, Thuesen BH, Tönjes A, Tromp G, Trompet S, Tsafantakis E, Tuomilehto J, Tybjaerg-Hansen A, Tyrer JP, Uher R, Uitterlinden AG, Uusitupa M, Laan SW, Duijn CM, Leeuwen N, van Setten J, Vanhala M, Varbo A, Varga TV, Varma R, Velez Edwards DR, Vermeulen SH, Veronesi G, Vestergaard H, Vitart V, Vogt TF, Völker U, Vuckovic D, Wagenknecht LE, Walker M, Wallentin L, Wang F, Wang CA, Wang S, Wang Y, Ware EB, Wareham NJ, Warren HR, Waterworth DM, Wessel J, White HD, Willer CJ, Wilson JG, Witte DR, Wood AR, Wu Y, Yaghootkar H, Yao J, Yao P, Yerges-Armstrong LM, Young R, Zeggini E, Zhan X, Zhang W, Zhao JH, Zhao W, Zhou W, Zondervan KT, Rotter JI, Pospisilik JA, Rivadeneira F, Borecki IB, Deloukas P, Frayling TM, Lettre G, North KE, Lindgren CM, Hirschhorn JN, Loos RJF; CHD Exome+Consortium; EPIC-CVD Consortium; ExomeBP Consortium; Global Lipids Genetic Consortium; GoT2D Genes Consortium; EPIC InterAct Consortium; INTERVAL Study; ReproGen Consortium; T2D-Genes Consortium; MAGIC Investigators; Understanding Society Scientific Group. Publisher Correction: Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nat Genet*. 2018 May;50(5):765-766. PMID: 29273807.

40: Vistnes M, Tapia G, Mårild K, Midttun Ø, Ueland PM, Viken MK, Magnus P, Berg JP, Gillespie KM, Skrivarhaug T, **Njølstad PR**, Joner G, Størdal K, Stene LC. Plasma immunological markers in pregnancy and cord blood: A possible link between macrophage chemo-attractants and risk of childhood type 1 diabetes. *Am J Reprod Immunol*. 2018 Mar;79(3). PMID: 29266506.

41: Flannick J, Fuchsberger C, Mahajan A, Teslovich TM, Agarwala V, Gaulton KJ, Caulkins L, Koesterer R, Ma C, Moutsianas L, McCarthy DJ, Rivas MA, Perry JRB, Sim X, Blackwell TW, Robertson NR, Rayner NW, Cingolani P,

Locke AE, Tajes JF, Highland HM, Dupuis J, Chines PS, Lindgren CM, Hartl C, Jackson AU, Chen H, Huyghe JR, van de Bunt M, Pearson RD, Kumar A, Müller-Nurasyid M, Grarup N, Stringham HM, Gamazon ER, Lee J, Chen Y, Scott RA, Below JE, Chen P, Huang J, Go MJ, Stitzel ML, Pasko D, Parker SCJ, Varga TV, Green T, Beer NL, Day-Williams AG, Ferreira T, Fingerlin T, Horikoshi M, Hu C, Huh I, Ikram MK, Kim BJ, Kim Y, Kim YJ, Kwon MS, Lee J, Lee S, Lin KH, Maxwell TJ, Nagai Y, Wang X, Welch RP, Yoon J, Zhang W, Barzilai N, Voight BF, Han BG, Jenkinson CP, Kuulasmaa T, Kuusisto J, Manning A, Ng MCY, Palmer ND, Balkau B, Stančáková A, Abboud HE, Boeing H, Giedraitis V, Prabhakaran D, Gottesman O, Scott J, Carey J, Kwan P, Grant G, Smith JD, Neale BM, Purcell S, Butterworth AS, Howson JMM, Lee HM, Lu Y, Kwak SH, Zhao W, Danesh J, Lam VKL, Park KS, Saleheen D, So WY, Tam CHT, Afzal U, Aguilar D, Arya R, Aung T, Chan E, Navarro C, Cheng CY, Palli D, Correa A, Curran JE, Rybin D, Farook VS, Fowler SP, Freedman BI, Griswold M, Hale DE, Hicks PJ, Khor CC, Kumar S, Lehne B, Thuillier D, Lim WY, Liu J, Loh M, Musani SK, Puppala S, Scott WR, Yengo L, Tan ST, Taylor HA, Thameem F, Wilson G, Wong TY, **Njølstad PR**, Levy JC, Mangino M, Bonnycastle LL, Schwarzmayr T, Fadista J, Surdulescu GL, Herder C, Groves CJ, Wieland T, Bork-Jensen J, Brandslund I, Christensen C, Koistinen HA, Doney ASF, Kinnunen L, Esko T, Farmer AJ, Hakaste L, Hodgkiss D, Kravic J, Lyssenko V, Hollensted M, Jørgensen ME, Jørgensen T, Ladenvall C, Justesen JM, Käräjämäki A, Kriebel J, Rathmann W, Lannfelt L, Lauritzen T, Narisu N, Linneberg A, Melander O, Milani L, Neville M, Orho-Melander M, Qi L, Qi Q, Roden M, Rolandsson O, Swift A, Rosengren AH, Stirrups K, Wood AR, Mihailov E, Blancher C, Carneiro MO, Maguire J, Poplin R, Shakir K, Fennell T, DePristo M, de Angelis MH, Deloukas P, Gjesing AP, Jun G, Nilsson P, Murphy J, Onofrio R, Thorand B, Hansen T, Meisinger C, Hu FB, Isomaa B, Karpe F, Liang L, Peters A, Huth C, O'Rahilly SP, Palmer CNA, Pedersen O, Rauramaa R, Tuomilehto J, Salomaa V, Watanabe RM, Syvänen AC, Bergman RN, Bharadwaj D, Bottinger EP, Cho YS, Chandak GR, Chan JCN, Chia KS, Daly MJ, Ebrahim SB, Langenberg C, Elliott P, Jablonski KA, Lehman DM, Jia W, Ma RCW, Pollin TI, Sandhu M, Tandon N, Froguel P, Barroso I, Teo YY, Zeggini E, Loos RJF, Small KS, Ried JS, DeFronzo RA, Grallert H, Glaser B, Metspalu A, Wareham NJ, Walker M, Banks E, Gieger C, Ingelsson E, Im HK, Illig T, Franks PW, Buck G, Trakalo J, Buck D, Prokopenko I, Mägi R, Lind L, Farjoun Y, Owen KR, Gloyn AL, Strauch K, Tuomi T, Kooner JS, Lee JY, Park T, Donnelly P, Morris AD, Hattersley AT, Bowden DW, Collins FS, Atzmon G, Chambers JC, Spector TD, Laakso M, Strom TM, Bell GI, Blangero J, Duggirala R, Tai ES, McVean G, Hanis CL, Wilson JG, Seielstad M, Frayling TM, Meigs JB, Cox NJ, Sladek R, Lander ES, Gabriel S, Mohlke KL, Meitinger T, Groop L, Abecasis G, Scott LJ, Morris AP, Kang HM, Altshuler D, Burtt NP, Florez JC, Boehnke M, McCarthy MI. Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Sci Data*. 2018 Jan 23;5:180002. PMID: 29257133.

42: Johansson BB, Fjeld K, El Jellas K, Gravdal A, Dalva M, Tjora E, Ræder H, Kulkarni RN, Johansson S, **Njølstad PR**, Molven A. The role of the carboxyl ester lipase (CEL) gene in pancreatic disease. *Pancreatology*. 2018 Jan;18(1):12-19. PMID: 29233499.

43: Thorsen SU, Mårild K, Olsen SF, Holst KK, Tapia G, Granström C, Halldorsson TI, Cohen AS, Haugen M, Lundqvist M, Skrivarhaug T, **Njølstad PR**, Joner G, Magnus P, Størdal K, Svensson J, Stene LC. Lack of Association Between Maternal or Neonatal Vitamin D Status and Risk of Childhood Type 1 Diabetes: A Scandinavian Case-Cohort Study. *Am J Epidemiol*. 2018 Jun 1;187(6):1174-1181. PMID: 29186303.

44: Skinner TC, Lange KS, Hoey H, Mortensen HB, Aanstoot HJ, Castaño L, Skovlund S, Swift PG, Cameron FJ, Dorchy HR, Palmert MR, Kaprio E, Robert JJ, Danne T, Neu A, Shalitin S, Chiarelli F, Chiari G, Urakami T, **Njølstad PR**, Jarosz-Chobot PK, Roche EF, Castro-Correia CG, Kocova M, Åman J, Schönle E, Barrett TG, Fisher L, de Beaufort CE; Hvidoere Study Group. Targets and teamwork: Understanding differences in pediatric diabetes centers treatment outcomes. *Pediatr Diabetes*. 2018 May;19(3):559-565. PMID: 29159931.

45: Sagen JV, Bjørkhaug L, Haukanes BI, Grevle L, Molnes J, Nedrebø BG, Søvik O, **Njølstad PR**, Johansson S, Molven A. The HNF1A mutant Ala180Val: Clinical challenges in determining causality of a rare HNF1A variant in familial diabetes. *Diabetes Res Clin Pract*. 2017 Nov;133:142-149. PMID: 28934671.

46: Johansson BB, Fjeld K, Solheim MH, Shirakawa J, Zhang E, Keindl M, Hu J, Lindqvist A, Døskeland A, Mellgren G, Flatmark T, **Njølstad PR**, Kulkarni RN, Wierup N, Aukrust I, Bjørkhaug L. Nuclear import of glucokinase in pancreatic beta-cells is mediated by a nuclear localization signal and modulated by SUMOylation. *Mol Cell Endocrinol.* 2017 Oct 15;454:146-157. PMID: 28648619.

47: Solheim MH, Clermont AC, Winnay JN, Hallstensen E, Molven A, **Njølstad PR**, Rødahl E, Kahn CR. Iris malformation and anterior segment dysgenesis in mice and humans with a mutation in PI 3-kinase. *Invest Ophthalmol Vis Sci*. 2017 Jun 1;58(7):3100-3106. PMID: 28632845.

48: Rebnord EW, Strand E, Midttun Ø, Svingen GFT, Christensen MHE, Ueland PM, Mellgren G, **Njølstad PR**, Tell GS, Nygård OK, Pedersen ER. The kynurenine:tryptophan ratio as a predictor of incident type 2 diabetes mellitus in individuals with coronary artery disease. *Diabetologia*. 2017 Sep;60(9):1712-1721. PMID: 28612106.

49: Lund-Blix NA, Dydensborg Sander S, Størdal K, Nybo Andersen AM, Rønningen KS, Joner G, Skrivarhaug T, **Njølstad PR**, Husby S, Stene LC. Infant Feeding and Risk of Type 1 Diabetes in Two Large Scandinavian Birth Cohorts. *Diabetes Care*. 2017 Jul;40(7):920-927. PMID: 28487451.

50: Kopczynski D, Barsnes H, **Njølstad PR**, Sickmann A, Vaudel M, Ahrends R. PeptideMapper: efficient and versatile amino acid sequence and tag mapping. *Bioinformatics.* 2017 Jul 1;33(13):2042-2044. PMID: 28334306.

51: Strand E, Pedersen ER, Svingen GF, Olsen T, Bjørndal B, Karlsson T, Dierkes J, **Njølstad PR**, Mellgren G, Tell GS, Berge RK, Svardal A, Nygård O. Serum Acylcarnitines and Risk of Cardiovascular Death and Acute Myocardial Infarction in Patients With Stable Angina Pectoris. *J Am Heart Assoc*. 2017 Feb 3;6(2). pii: e003620. PMID: 28159823.

52: Marouli E, Graff M, Medina-Gomez C, Lo KS, Wood AR, Kjaer TR, Fine RS, Lu Y, Schurmann C, Highland HM, Rüeger S, Thorleifsson G, Justice AE, Lamparter D, Stirrups KE, Turcot V, Young KL, Winkler TW, Esko T, Karaderi T, Locke AE, Masca NG, Ng MC, Mudgal P, Rivas MA, Vedantam S, Mahajan A, Guo X, Abecasis G, Aben KK, Adair LS, Alam DS, Albrecht E, Allin KH, Allison M, Amouyel P, Appel EV, Arveiler D, Asselbergs FW, Auer PL, Balkau B, Banas B, Bang LE, Benn M, Bergmann S, Bielak LF, Blüher M, Boeing H, Boerwinkle E, Böger CA, Bonnycastle LL, Bork-Jensen J, Bots ML, Bottinger EP, Bowden DW, Brandslund I, Breen G, Brilliant MH, Broer L, Burt AA, Butterworth AS, Carey DJ, Caulfield MJ, Chambers JC, Chasman DI, Chen YI, Chowdhury R, Christensen C, Chu AY, Cocca M, Collins FS, Cook JP, Corley J, Galbany JC, Cox AJ, Cuellar-Partida G, Danesh J, Davies G, de Bakker PI, de Borst GJ, de Denus S, de Groot MC, de Mutsert R, Deary IJ, Dedoussis G, Demerath EW, den Hollander AI, Dennis JG, Di Angelantonio E, Drenos F, Du M, Dunning AM, Easton DF, Ebeling T, Edwards TL, Ellinor PT, Elliott P, Evangelou E, Farmaki AE, Faul JD, Feitosa MF, Feng S, Ferrannini E, Ferrario MM, Ferrieres J, Florez JC, Ford I, Fornage M, Franks PW, Frikke-Schmidt R, Galesloot TE, Gan W, Gandin I, Gasparini P, Giedraitis V, Giri A, Girotto G, Gordon SD, Gordon-Larsen P, Gorski M, Grarup N, Grove ML, Gudnason V, Gustafsson S, Hansen T, Harris KM, Harris TB, Hattersley AT, Hayward C, He L, Heid IM, Heikkilä K, Helgeland Ø, Hernesniemi J, Hewitt AW, Hocking LJ, Hollensted M, Holmen OL, Hovingh GK, Howson JM, Hoyng CB, Huang PL, Hveem K, Ikram MA, Ingelsson E, Jackson AU, Jansson JH, Jarvik GP, Jensen GB, Jhun MA, Jia Y, Jiang X, Johansson S, Jørgensen ME, Jørgensen T, Jousilahti P, Jukema JW, Kahali B, Kahn RS, Kähönen M, Kamstrup PR, Kanoni S, Kaprio J, Karaleftheri M, Kardia SL, Karpe F, Kee F, Keeman R, Kiemeney LA, Kitajima H, Kluivers KB, Kocher T, Komulainen P, Kontto J, Kooner JS, Kooperberg C, Kovacs P, Kriebel J, Kuivaniemi H, Küry S, Kuusisto J, La Bianca M, Laakso M, Lakka TA, Lange EM, Lange LA, Langefeld CD, Langenberg C, Larson EB, Lee IT, Lehtimäki T, Lewis CE, Li H, Li J, Li-Gao R, Lin H, Lin LA, Lin X, Lind L, Lindström J, Linneberg A, Liu Y, Liu Y, Lophatananon A, Luan J, Lubitz SA, Lyytikäinen LP, Mackey DA, Madden PA, Manning AK, Männistö S, Marenne G, Marten J, Martin NG, Mazul AL, Meidtner K, Metspalu A, Mitchell P, Mohlke KL, Mook-Kanamori DO, Morgan A, Morris AD, Morris AP, Müller-Nurasyid M, Munroe PB, Nalls MA, Nauck M, Nelson CP, Neville M, Nielsen SF, Nikus K, **Njølstad PR**, Nordestgaard BG, Ntalla I, O'Connel

JR, Oksa H, Loohuis LM, Ophoff RA, Owen KR, Packard CJ, Padmanabhan S, Palmer CN, Pasterkamp G, Patel AP, Pattie A, Pedersen O, Peissig PL, Peloso GM, Pennell CE, Perola M, Perry JA, Perry JR, Person TN, Pirie A, Polasek O, Posthuma D, Raitakari OT, Rasheed A, Rauramaa R, Reilly DF, Reiner AP, Renström F, Ridker PM, Rioux JD, Robertson N, Robino A, Rolandsson O, Rudan I, Ruth KS, Saleheen D, Salomaa V, Samani NJ, Sandow K, Sapkota Y, Sattar N, Schmidt MK, Schreiner PJ, Schulze MB, Scott RA, Segura-Lepe MP, Shah S, Sim X, Sivapalaratnam S, Small KS, Smith AV, Smith JA, Southam L, Spector TD, Speliotes EK, Starr JM, Steinthorsdottir V, Stringham HM, Stumvoll M, Surendran P, 't Hart LM, Tansey KE, Tardif JC, Taylor KD, Teumer A, Thompson DJ, Thorsteinsdottir U, Thuesen BH, Tönjes A, Tromp G, Trompet S, Tsafantakis E, Tuomilehto J, Tybjaerg-Hansen A, Tyrer JP, Uher R, Uitterlinden AG, Ulivi S, van der Laan SW, Van Der Leij AR, van Duijn CM, van Schoor NM, van Setten J, Varbo A, Varga TV, Varma R, Edwards DR, Vermeulen SH, Vestergaard H, Vitart V, Vogt TF, Vozzi D, Walker M, Wang F, Wang CA, Wang S, Wang Y, Wareham NJ, Warren HR, Wessel J, Willems SM, Wilson JG, Witte DR, Woods MO, Wu Y, Yaghootkar H, Yao J, Yao P, Yerges-Armstrong LM, Young R, Zeggini E, Zhan X, Zhang W, Zhao JH, Zhao W, Zhao W, Zheng H, Zhou W; EPIC-InterAct Consortium; CHD Exome+ Consortium; ExomeBP Consortium; T2D-Genes Consortium; GoT2D Genes Consortium; Global Lipids Genetics Consortium; ReproGen Consortium; MAGIC Investigators, Rotter JI, Boehnke M, Kathiresan S, McCarthy MI, Willer CJ, Stefansson K, Borecki IB, Liu DJ, North KE, Heard-Costa NL, Pers TH, Lindgren CM, Oxvig C, Kutalik Z, Rivadeneira F, Loos RJ, Frayling TM, Hirschhorn JN, Deloukas P, Lettre G. Rare and low-frequency coding variants alter human adult height. *Nature*. 2017 Feb 9;542(7640):186-190. PMID: 28146470.

53: Johansson BB, Irgens HU, Molnes J, Sztromwasser P, Aukrust I, Juliusson PB, Søvik O, Levy S, Skrivarhaug T, Joner G, Molven A, Johansson S, **Njølstad PR**. 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 Apr;60(4):625-635. PMID: 27913849.

54: Najmi LA, Aukrust I, Flannick J, Molnes J, Burtt N, Molven A, Groop L, Altshuler D, Johansson S, Bjørkhaug L, **Njølstad PR**. Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. *Diabetes*. 2017 Feb;66(2):335-346. PMID: 27899486.

55: Madzak A, Engjom T, Wathle GK, Olesen SS, Tjora E, **Njølstad PR**, Lærum BN, Drewes AM, Dimcevski G, Frøkjær JB, Haldorsen IS. Secretin-stimulated MRI assessment of exocrine pancreatic function in patients with cystic fibrosis and healthy controls. *Abdom Radiol (NY).* 2017 Mar;42(3):890-899. PMID: 27832324.

56: Fjeld K, Beer S, Johnstone M, Zimmer C, Mössner J, Ruffert C, Krehan M, Zapf C, **Njølstad PR**, Johansson S, Bugert P, Miyajima F, Liloglou T, Brown LJ, Winn SA, Davies K, Latawiec D, Gunson BK, Criddle DN, Pirmohamed M, Grützmann R, Michl P, Greenhalf W, Molven A, Sutton R, Rosendahl J. 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 Nov 1;11(11):e0165567. PMID: 27802312.

57: Dalva M, El Jellas K, Steine SJ, Johansson BB, Ringdal M, Torsvik J, Immervoll H, Hoem D, Laemmerhirt F, Simon P, Lerch MM, Johansson S, **Njølstad PR**, Weiss FU, Fjeld K, Molven A. Copy number variants and VNTR length polymorphisms of the carboxyl-ester lipase (CEL) gene as risk factors in pancreatic cancer. *Pancreatology*. 2017 Jan - Feb;17(1):83-88. PMID: 27773618.

58: Lek M, Karczewski KJ, Minikel EV, Samocha KE, Banks E, Fennell T, O'Donnell-Luria AH, Ware JS, Hill AJ, Cummings BB, Tukiainen T, Birnbaum DP, Kosmicki JA, Duncan LE, Estrada K, Zhao F, Zou J, Pierce-Hoffman E, Berghout J, Cooper DN, Deflaux N, DePristo M, Do R, Flannick J, Fromer M, Gauthier L, Goldstein J, Gupta N, Howrigan D, Kiezun A, Kurki MI, Moonshine AL, Natarajan P, Orozco L, Peloso GM, Poplin R, Rivas MA, Ruano-Rubio V, Rose SA, Ruderfer DM, Shakir K, Stenson PD, Stevens C, Thomas BP, Tiao G, Tusie-Luna MT, Weisburd B, Won HH, Yu D, Altshuler DM, Ardissino D, Boehnke M, Danesh J, Donnelly S, Elosua R, Florez JC, Gabriel SB,

Getz G, Glatt SJ, Hultman CM, Kathiresan S, Laakso M, McCarroll S, McCarthy MI, McGovern D, McPherson R, Neale BM, Palotie A, Purcell SM, Saleheen D, Scharf JM, Sklar P, Sullivan PF, Tuomilehto J, Tsuang MT, Watkins HC, Wilson JG, Daly MJ, MacArthur DG; Exome Aggregation Consortium [incl. **Njølstad PR**]. Analysis of protein-coding genetic variation in 60,706 humans. *Nature*. 2016 Aug 18;536(7616):285-91. PMID: 27535533.

59: Fuchsberger C, Flannick J, Teslovich TM, Mahajan A, Agarwala V, Gaulton KJ, Ma C, Fontanillas P, Moutsianas L, McCarthy DJ, Rivas MA, Perry JRB, Sim X, Blackwell TW, Robertson NR, Rayner NW, Cingolani P, Locke AE, Tajes JF, Highland HM, Dupuis J, Chines PS, Lindgren CM, Hartl C, Jackson AU, Chen H, Huyghe JR, van de Bunt M, Pearson RD, Kumar A, Müller-Nurasyid M, Grarup N, Stringham HM, Gamazon ER, Lee J, Chen Y, Scott RA, Below JE, Chen P, Huang J, Go MJ, Stitzel ML, Pasko D, Parker SCJ, Varga TV, Green T, Beer NL, Day-Williams AG, Ferreira T, Fingerlin T, Horikoshi M, Hu C, Huh I, Ikram MK, Kim BJ, Kim Y, Kim YJ, Kwon MS, Lee J, Lee S, Lin KH, Maxwell TJ, Nagai Y, Wang X, Welch RP, Yoon J, Zhang W, Barzilai N, Voight BF, Han BG, Jenkinson CP, Kuulasmaa T, Kuusisto J, Manning A, Ng MCY, Palmer ND, Balkau B, Stančáková A, Abboud HE, Boeing H, Giedraitis V, Prabhakaran D, Gottesman O, Scott J, Carey J, Kwan P, Grant G, Smith JD, Neale BM, Purcell S, Butterworth AS, Howson JMM, Lee HM, Lu Y, Kwak SH, Zhao W, Danesh J, Lam VKL, Park KS, Saleheen D, So WY, Tam CHT, Afzal U, Aguilar D, Arya R, Aung T, Chan E, Navarro C, Cheng CY, Palli D, Correa A, Curran JE, Rybin D, Farook VS, Fowler SP, Freedman BI, Griswold M, Hale DE, Hicks PJ, Khor CC, Kumar S, Lehne B, Thuillier D, Lim WY, Liu J, van der Schouw YT, Loh M, Musani SK, Puppala S, Scott WR, Yengo L, Tan ST, Taylor HA Jr, Thameem F, Wilson G Sr, Wong TY, **Njølstad PR**, Levy JC, Mangino M, Bonnycastle LL, Schwarzmayr T, Fadista J, Surdulescu GL, Herder C, Groves CJ, Wieland T, Bork-Jensen J, Brandslund I, Christensen C, Koistinen HA, Doney ASF, Kinnunen L, Esko T, Farmer AJ, Hakaste L, Hodgkiss D, Kravic J, Lyssenko V, Hollensted M, Jørgensen ME, Jørgensen T, Ladenvall C, Justesen JM, Käräjämäki A, Kriebel J, Rathmann W, Lannfelt L, Lauritzen T, Narisu N, Linneberg A, Melander O, Milani L, Neville M, Orho-Melander M, Qi L, Qi Q, Roden M, Rolandsson O, Swift A, Rosengren AH, Stirrups K, Wood AR, Mihailov E, Blancher C, Carneiro MO, Maguire J, Poplin R, Shakir K, Fennell T, DePristo M, de Angelis MH, Deloukas P, Gjesing AP, Jun G, Nilsson P, Murphy J, Onofrio R, Thorand B, Hansen T, Meisinger C, Hu FB, Isomaa B, Karpe F, Liang L, Peters A, Huth C, O'Rahilly SP, Palmer CNA, Pedersen O, Rauramaa R, Tuomilehto J, Salomaa V, Watanabe RM, Syvänen AC, Bergman RN, Bharadwaj D, Bottinger EP, Cho YS, Chandak GR, Chan JCN, Chia KS, Daly MJ, Ebrahim SB, Langenberg C, Elliott P, Jablonski KA, Lehman DM, Jia W, Ma RCW, Pollin TI, Sandhu M, Tandon N, Froguel P, Barroso I, Teo YY, Zeggini E, Loos RJF, Small KS, Ried JS, DeFronzo RA, Grallert H, Glaser B, Metspalu A, Wareham NJ, Walker M, Banks E, Gieger C, Ingelsson E, Im HK, Illig T, Franks PW, Buck G, Trakalo J, Buck D, Prokopenko I, Mägi R, Lind L, Farjoun Y, Owen KR, Gloyn AL, Strauch K, Tuomi T, Kooner JS, Lee JY, Park T, Donnelly P, Morris AD, Hattersley AT, Bowden DW, Collins FS, Atzmon G, Chambers JC, Spector TD, Laakso M, Strom TM, Bell GI, Blangero J, Duggirala R, Tai ES, McVean G, Hanis CL, Wilson JG, Seielstad M, Frayling TM, Meigs JB, Cox NJ, Sladek R, Lander ES, Gabriel S, Burtt NP, Mohlke KL, Meitinger T, Groop L, Abecasis G, Florez JC, Scott LJ, Morris AP, Kang HM, Boehnke M, Altshuler D, McCarthy MI. The genetic architecture of type 2 diabetes. *Nature*. 2016 Aug 4;536(7614):41-47. PMID: 27398621.

60: Flannick J, Johansson S, **Njølstad PR**. Common and rare forms of diabetes mellitus: towards a continuum of diabetes subtypes. *Nat Rev Endocrinol*. 2016 Jul;12(7):394-406. PMID: 27080136.

61: Gagnum V, Stene LC, Jenssen TG, Berteussen LM, Sandvik L, Joner G, **Njølstad PR**, Skrivarhaug T. Causes of death in childhood-onset Type 1 diabetes: long-term follow-up. *Diabet Med*. 2017 Jan;34(1):56-63. PMID: 26996105.

62: Svingen GF, Schartum-Hansen H, Pedersen ER, Ueland PM, Tell GS, Mellgren G, **Njølstad PR**, Seifert R, Strand E, Karlsson T, Nygård O. Prospective Associations of Systemic and Urinary Choline Metabolites with Incident Type 2 Diabetes. *Clin Chem.* 2016 May;62(5):755-65. PMID: 26980210.

63: Winnay JN, Solheim MH, Dirice E, Sakaguchi M, Noh HL, Kang HJ, Takahashi H, Chudasama KK, Kim JK, Molven A, Kahn CR, **Njølstad PR**. PI3-kinase mutation linked to insulin and growth factor resistance in vivo. *J Clin Invest*. 2016 Apr 1;126(4):1401-12. PMID: 26974159.

64: Molven A, Hollister-Lock J, Hu J, Martinez R, **Njølstad PR**, Liew CW, Weir G, Kulkarni RN. The hypoglycemic phenotype Is islet cell-autonomous in short-chain hydroxyacyl-CoA dehydrogenase-deficient mice. *Diabetes*. 2016 Jun;65(6):1672-8. PMID: 26953163.

65: Balamurugan K, Bjørkhaug L, Mahajan S, Kanthimathi S, **Njølstad PR**, Srinivasan N, Mohan V, Radha V. Structure-function studies of HNF1A (MODY3) gene mutations in South Indian patients with monogenic diabetes. *Clin Genet*. 2016 Dec;90(6):486-495. PMID: 26853433.

66: Magnus MC, Olsen SF, Granström C, Joner G, Skrivarhaug T, Svensson J, Johannesen J, **Njølstad PR**, Magnus P, Størdal K, Stene LC. Infant growth and risk of childhood-onset type 1 diabetes in children from 2 Scandinavian birth cohorts. *JAMA Pediatr*. 2015 Dec;169(12):e153759. PMID: 26642117.

67: Avila M, Dyment DA, Sagen JV, St-Onge J, Moog U, Chung BHY, Mo S, Mansour S, Albanese A, Garcia S, Martin DO, Lopez AA, Claudi T, König R, White SM, Sawyer SL, Bernstein JA, Slattery L, Jobling RK, Yoon G, Curry CJ, Merrer ML, Luyer BL, Héron D, Mathieu-Dramard M, Bitoun P, Odent S, Amiel J, Kuentz P, Thevenon J, Laville M, Reznik Y, Fagour C, Nunes ML, Delesalle D, Manouvrier S, Lascols O, Huet F, Binquet C, Faivre L, Rivière JB, Vigouroux C, **Njølstad PR**, Innes AM, Thauvin-Robinet C. Clinical reappraisal of SHORT syndrome with PIK3R1 mutations: toward recommendation for molecular testing and management. *Clin Genet*. 2016 Apr;89(4):501-506. PMID: 26497935.

68: Kaur S, Mirza AH, Brorsson CA, Fløyel T, Størling J, Mortensen HB, Pociot F; Hvidoere International Study Group [invl. **Njølstad PR**]. The genetic and regulatory architecture of ERBB3-type 1 diabetes susceptibility locus. *Mol Cell Endocrinol*. 2016 Jan 5;419:83-91. PMID: 26450151.

69: Rozenkova K, Malikova J, Nessa A, Dusatkova L, Bjørkhaug L, Obermannova B, Dusatkova P, Kytnarova J, Aukrust I, Najmi LA, Rypackova B, Sumnik Z, Lebl J, **Njølstad PR**, Hussain K, Pruhova S. High incidence of heterozygous ABCC8 and HNF1A mutations in Czech patients with congenital hyperinsulinism. *J Clin Endocrinol Metab.* 2015 Dec;100(12):E1540-9. PMID: 26431509.

70: Molven A, **Njølstad PR**, Weiss FU. Lipase gene fusion: a new route to chronic pancreatitis*. Oncotarget*. 2015 Oct 13;6(31):30443-4. Review. PMID: 26387137.

71: Zhang G, Bacelis J, Lengyel C, Teramo K, Hallman M, Helgeland Ø, Johansson S, Myhre R, Sengpiel V, **Njølstad PR**, Jacobsson B, Muglia L. Assessing the causal relationship of maternal height on birth size and gestational age at birth: A Mendelian randomization analysis. *PLoS Med*. 2015 Aug 18;12(8):e1001865. PMID: 26284790.

72: Helgeland Ø, Hertel JK, Molven A, Ræder H, Platou CG, Midthjell K, Hveem K, Nygård O, **Njølstad PR**, Johansson S. The chromosome 9p21 CVD- and T2D-associated regions in a Norwegian population (The HUNT2 Survey). *Int J Endocrinol*. 2015;2015:164652. PMID: 26089876.

73: Gagnum V, Stene LC, Sandvik L, Fagerland MW, **Njølstad PR**, Joner G, Skrivarhaug T. All-cause mortality in a nationwide cohort of childhood-onset diabetes in Norway 1973-2013. *Diabetologia*. 2015 Aug;58(8):1779-86. PMID: 25972232.

74: Neu A, Lange K, Barrett T, Cameron F, Dorchy H, Hoey H, Jarosz-Chobot P, Mortensen HB, Robert JJ, Robertson K, de Beaufort C; Hvidoere Study Group [incl. **Njølstad PR**]. Classifying insulin regimens--difficulties and proposal for comprehensive new definitions. *Pediatr Diabetes*. 2015 Sep;16(6):402-6. PMID: 25865149.

75: Fjeld K, Weiss FU, Lasher D, Rosendahl J, Chen JM, Johansson BB, Kirsten H, Ruffert C, Masson E, Steine SJ, Bugert P, Cnop M, Grützmann R, Mayerle J, Mössner J, Ringdal M, Schulz HU, Sendler M, Simon P, Sztromwasser P, Torsvik J, Scholz M, Tjora E, Férec C, Witt H, Lerch MM, **Njølstad PR**, Johansson S, Molven A. A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. *Nat Genet*. 2015 May;47(5):518-522. PMID: 25774637.

76: Irgens HU, Fjeld K, Johansson BB, Ringdal M, Immervoll H, Leh S, Søvik O, Johansson S, Molven A, **Njølstad PR**. Glycogenin-2 is dispensable for liver glycogen synthesis and glucagon-stimulated glucose release. *J Clin Endocrinol Metab*. 2015 May;100(5):E767-75. PMID: 25751106.

77: Eifes S, Chudasama KK, Molnes J, Wagner K, Hoang T, Schierloh U, Rocour-Brumioul D, Johansson S, **Njølstad PR**, de Beaufort C. A novel GATA6 mutation in a child with congenital heart malformation and neonatal diabetes. *Clin Case Rep*. 2013 Dec;1(2):86-90. PMID: 25356219.

78: Rubio-Cabezas O, Hattersley AT, **Njølstad PR**, Mlynarski W, Ellard S, White N, Chi DV, Craig ME; International Society for Pediatric and Adolescent Diabetes. ISPAD Clinical Practice Consensus Guidelines 2014. The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatr Diabetes*. 2014 Sep;15 Suppl 20:47-64. PMID: 25182307.

79: Torsvik J, Johansson BB, Dalva M, Marie M, Fjeld K, Johansson S, Bjørkøy G, Saraste J, **Njølstad PR**, Molven A. Endocytosis of secreted carboxyl ester lipase in a syndrome of diabetes and pancreatic exocrine dysfunction. *J Biol Chem*. 2014 Oct 17;289(42):29097-111. PMID: 25160620.

80: Majithia AR, Flannick J, Shahinian P, Guo M, Bray MA, Fontanillas P, Gabriel SB; Estrada K, Mercader J, MacArthur D, GoT2D Consortium; NHGRI JHS/FHS Allelic Spectrum Project; SIGMA T2D Consortium [incl. **Njølstad PR**]; T2D-GENES Consortium, Rosen ED, Altshuler D. Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. *Proc Natl Acad Sci U S A*. 2014 Sep 9;111(36):13127-32. PMID: 25157153.

81: SIGMA Type 2 Diabetes Consortium, Estrada K, Aukrust I, Bjørkhaug L, Burtt NP, Mercader JM, García-Ortiz H, Huerta-Chagoya A, Moreno-Macías H, Walford G, Flannick J, Williams AL, Gómez-Vázquez MJ, Fernandez-Lopez JC, Martínez-Hernández A, Jiménez-Morales S, Centeno-Cruz F, Mendoza-Caamal E, Revilla-Monsalve C, Islas-Andrade S, Córdova EJ, Soberón X, González-Villalpando ME, Henderson E, Wilkens LR, Le Marchand L, Arellano-Campos O, Ordóñez-Sánchez ML, Rodríguez-Torres M, Rodríguez-Guillén R, Riba L, Najmi LA, Jacobs SB, Fennell T, Gabriel S, Fontanillas P, Hanis CL, Lehman DM, Jenkinson CP, Abboud HE, Bell GI, Cortes ML, Boehnke M, González-Villalpando C, Orozco L, Haiman CA, Tusié-Luna T, Aguilar-Salinas CA, Altshuler D, **Njølstad PR**, Florez JC, MacArthur DG. Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. *JAMA*. 2014 Jun 11;311(22):2305-14. PMID: 24915262.

82: Flannick J, Thorleifsson G, Beer NL, Jacobs SB, Grarup N, Burtt NP, Mahajan A, Fuchsberger C, Atzmon G, Benediktsson R, Blangero J, Bowden DW, Brandslund I, Brosnan J, Burslem F, Chambers J, Cho YS, Christensen C, Douglas DA, Duggirala R, Dymek Z, Farjoun Y, Fennell T, Fontanillas P, Forsén T, Gabriel S, Glaser B, Gudbjartsson DF, Hanis C, Hansen T, Hreidarsson AB, Hveem K, Ingelsson E, Isomaa B, Johansson S, Jørgensen T, Jørgensen ME, Kathiresan S, Kong A, Kooner J, Kravic J, Laakso M, Lee JY, Lind L, Lindgren CM, Linneberg A, Masson G, Meitinger T, Mohlke KL, Molven A, Morris AP, Potluri S, Rauramaa R, Ribel-Madsen R, Richard AM, Rolph T, Salomaa V,

Segrè AV, Skärstrand H, Steinthorsdottir V, Stringham HM, Sulem P, Tai ES, Teo YY, Teslovich T, Thorsteinsdottir U, Trimmer JK, Tuomi T, Tuomilehto J, Vaziri-Sani F, Voight BF, Wilson JG, Boehnke M, McCarthy MI, **Njølstad PR**, Pedersen O; Go-T2D Consortium; T2D-GENES Consortium, Groop L, Cox DR, Stefansson K, Altshuler D. Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. *Nat Genet*. 2014 Apr;46(4):357-63. PMID: 24584071.

83: Flannick J, Beer NL, Bick AG, Agarwala V, Molnes J, Gupta N, Burtt NP, Florez JC, Meigs JB, Taylor H, Lyssenko V, Irgens H, Fox E, Burslem F, Johansson S, Brosnan MJ, Trimmer JK, Newton-Cheh C, Tuomi T, Molven A, Wilson JG, O'Donnell CJ, Kathiresan S, Hirschhorn JN, **Njølstad PR**, Rolph T, Seidman JG, Gabriel S, Cox DR, Seidman CE, Groop L, Altshuler D. Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. *Nat Genet.* 2013 Nov;45(11):1380-5. PMID: 24097065.

84: Ræder H, McAllister FE, Tjora E, Bhatt S, Haldorsen I, Hu J, Willems SM, Vesterhus M, El Ouaamari A, Liu M, Ræder MB, Immervoll H, Hoem D, Dimcevski G, **Njølstad PR**, Molven A, Gygi SP, Kulkarni RN. 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 Jan;63(1):259-69. PMID: 24062244.

85: Negahdar M, Aukrust I, Molnes J, Solheim MH, Johansson BB, Sagen JV, Dahl-Jørgensen K, Kulkarni RN, Søvik O, Flatmark T, **Njølstad PR**, Bjørkhaug L. GCK-MODY diabetes as a protein misfolding disease: the mutation R275C promotes protein misfolding, self-association and cellular degradation. *Mol Cell Endocrinol*. 2014 Jan 25;382(1):55-65. PMID: 24001579.

86: Chudasama KK, Winnay J, Johansson S, Claudi T, König R, Haldorsen I, Johansson B, Woo JR, Aarskog D, Sagen JV, Kahn CR, Molven A, **Njølstad PR**. SHORT syndrome with partial lipodystrophy due to impaired phosphatidylinositol 3 kinase signaling. *Am J Hum Genet*. 2013 Jul 11;93(1):150-7. PMID: 23810379.

87: Due P, de Beaufort C, Damsgaard MT, Mortensen HB, Rasmussen M, Ahluwalia N, Skinner T, Swift P; HSG and HBSC consortia [incl. **Njølstad PR**]. Do eating behaviors in the general population account for country variance in glycemic control among adolescents with diabetes: the Hvidoere Study Group and the Health Behaviour in School-Aged Children study. *Pediatr Diabetes*. 2013 Dec;14(8):554-61. PMID: 23773782.

88: Tjora E, Wathle G, Engjom T, Erchinger F, Molven A, Aksnes L, Haldorsen IS, Dimcevski G, **Njølstad PR**, Ræder H. Severe pancreatic dysfunction but compensated nutritional status in monogenic pancreatic disease caused by carboxyl-ester lipase mutations*. Pancreas*. 2013 Oct;42(7):1078-84. PMID: 23770712.

89: Wathle GK, Tjora E, Ersland L, Dimcevski G, Salvesen ØO, Molven A, **Njølstad PR**, Haldorsen IS. Assessment of exocrine pancreatic function by secretin-stimulated magnetic resonance cholangiopancreaticography and diffusion-weighted imaging in healthy controls. *J Magn Reson Imaging*. 2014 Feb;39(2):448-54. PMID: 23649590.

90: Cameron FJ, de Beaufort C, Aanstoot HJ, Hoey H, Lange K, Castano L, Mortensen HB; Hvidoere International Study Group [incl. **Njølstad PR**]. Lessons from the Hvidoere International Study Group on childhood diabetes: be dogmatic about outcome and flexible in approach. *Pediatr Diabetes*. 2013 Nov;14(7):473-80. PMID: 23627895.

91: Irgens HU, Molnes J, Johansson BB, Ringdal M, Skrivarhaug T, Undlien DE, Søvik O, Joner G, Molven A, **Njølstad PR**. Prevalence of monogenic diabetes in the population-based Norwegian Childhood Diabetes Registry. *Diabetologia*. 2013 Jul;56(7):1512-9. PMID: 23624530.

92: Tjora E, Wathle G, Erchinger F, Engjom T, Molven A, Aksnes L, Haldorsen IS, Dimcevski G, Raeder H, **Njølstad PR**. Exocrine pancreatic function in hepatocyte nuclear factor 1β-maturity-onset diabetes of the young (HNF1B-

MODY) is only moderately reduced: compensatory hypersecretion from a hypoplastic pancreas. *Diabet Med*. 2013 Aug;30(8):946-55. PMID: 23600988.

93: Ræder H, Vesterhus M, El Ouaamari A, Paulo JA, McAllister FE, Liew CW, Hu J, Kawamori D, Molven A, Gygi SP, **Njølstad PR**, Kahn CR, Kulkarni RN. Absence of diabetes and pancreatic exocrine dysfunction in a transgenic model of carboxyl-ester lipase-MODY (maturity-onset diabetes of the young). *PLoS One*. 2013;8(4):e60229. PMID: 23565203.

94: Ragvin A, Fjeld K, Weiss FU, Torsvik J, Aghdassi A, Mayerle J, Simon P, **Njølstad PR**, Lerch MM, Johansson S, Molven A. The number of tandem repeats in the carboxyl-ester lipase (CEL) gene as a risk factor in alcoholic and idiopathic chronic pancreatitis. *Pancreatology*. 2013 Jan-Feb;13(1):29-32. PMID: 23395566.

95: Teo AK, Windmueller R, Johansson BB, Dirice E, **Njølstad PR**, Tjora E, Raeder H, Kulkarni RN. Derivation of human induced pluripotent stem cells from patients with maturity onset diabetes of the young. *J Biol Chem*. 2013 Feb 22;288(8):5353-6. PMID: 23306198.

96: Aukrust I, Bjørkhaug L, Negahdar M, Molnes J, Johansson BB, Müller Y, Haas W, Gygi SP, Søvik O, Flatmark T, Kulkarni RN, **Njølstad PR**. SUMOylation of pancreatic glucokinase regulates its cellular stability and activity. *J Biol Chem*. 2013 Feb 22;288(8):5951-62. PMID: 23297408.

97: Thanabalasingham G, Huffman JE, Kattla JJ, Novokmet M, Rudan I, Gloyn AL, Hayward C, Adamczyk B, Reynolds RM, Muzinic A, Hassanali N, Pucic M, Bennett AJ, Essafi A, Polasek O, Mughal SA, Redzic I, Primorac D, Zgaga L, Kolcic I, Hansen T,Gasperikova D, Tjora E, Strachan MW, Nielsen T, Stanik J, Klimes I, Pedersen OB, **Njølstad PR**, Wild SH, Gyllensten U, Gornik O, Wilson JF, Hastie ND, Campbell H, McCarthy MI, Rudd PM, Owen KR, Lauc G, Wright AF. Mutations in HNF1A result in marked alterations of plasma glycan profile. *Diabetes*. 2013 Apr;62(4):1329-37. PMID: 23274891.

98: Elbarbary NS, Tjora E, Molnes J, Lie BA, Habib MA, Salem MA, **Njølstad PR**. 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. *Pediatr Diabetes*. 2013 Sep;14(6):466-72. PMID: 22989030.

99: Ræder H, Molven A, **Njølstad PR**. [Tailored medicine or narcissomics?]. *Tidsskr Nor Laegeforen*. 2012 Sep 4;132(16):1844-5. PMID: 22986961.

100: de Beaufort CE, Lange K, Swift PG, Aman J, Cameron F, Castano L, Dorchy H, Fisher LK, Hoey H, Kaprio E, Kocova M, Neu A, **Njølstad PR**, Phillip M, Schoenle E, Robert JJ, Urukami T, Vanelli M, Danne T, Barrett T, Chiarelli F, Aanstoot HJ, Mortensen HB; Hvidoere Study Group. Metabolic outcomes in young children with type 1 diabetes differ between treatment centers: the Hvidoere Study in Young Children 2009. *Pediatr Diabetes*. 2013 Sep;14(6):422-8. PMID: 22957743.

102: Jahnavi S, Poovazhagi V, Mohan V, Bodhini D, Raghupathy P, Amutha A, Suresh Kumar P, Adhikari P, Shriraam M, Kaur T, Das AK, Molnes J, **Njølstad PR**, Unnikrishnan R, Radha V. Clinical and molecular characterization of neonatal diabetes and monogenic syndromic diabetes in Asian Indian children. *Clin Genet*. 2013 May;83(5):439-45. PMID: 22831748.

102: Negahdar M, Aukrust I, Johansson BB, Molnes J, Molven A, Matschinsky FM, Søvik O, Kulkarni RN, Flatmark T, **Njølstad PR**, Bjørkhaug L. GCK-MODY diabetes associated with protein misfolding, cellular self-association and degradation. *Biochim Biophys Acta*. 2012 Nov;1822(11):1705-15. PMID: 22820548.

103: Johansson S, Irgens H, Chudasama KK, Molnes J, Aerts J, Roque FS, Jonassen I, Levy S, Lima K, Knappskog PM, Bell GI, Molven A, **Njølstad PR**. Exome sequencing and genetic testing for MODY. *PLoS One*. 2012;7(5):e38050. PMID: 22662265.

104: Eike MC, Skinningsrud B, Ronninger M, Stormyr A, Kvien TK, Joner G, **Njølstad PR**, Førre O, Flatø B, Alfredsson L, Padyukov L, Undlien DE, Lie BA. CIITA gene variants are associated with rheumatoid arthritis in Scandinavian populations. *Genes Immun*. 2012 Jul;13(5):431-6. PMID: 22513452.

105: **Njølstad PR**, Molven A. To test, or not to test: time for a MODY calculator? *Diabetologia*. 2012 May;55(5):1231-4. PMID: 22382521.

106: Haldorsen IS, Ræder H, Vesterhus M, Molven A, **Njølstad PR**. The role of pancreatic imaging in monogenic diabetes mellitus. *Nat Rev Endocrinol*. 2011 Nov 29;8(3):148-59. Review. PMID: 22124438.

107: Kilpeläinen TO, Qi L, Brage S, Sharp SJ, Sonestedt E, Demerath E, Ahmad T, Mora S, Kaakinen M, Sandholt CH, Holzapfel C, Autenrieth CS, Hyppönen E, Cauchi S, He M, Kutalik Z, Kumari M, Stančáková A, Meidtner K, Balkau B, Tan JT, Mangino M, Timpson NJ, Song Y, Zillikens MC, Jablonski KA, Garcia ME, Johansson S, Bragg-Gresham JL, Wu Y, van Vliet-Ostaptchouk JV, Onland-Moret NC, Zimmermann E, Rivera NV, Tanaka T, Stringham HM, Silbernagel G, Kanoni S, Feitosa MF, Snitker S, Ruiz JR, Metter J, Larrad MT, Atalay M, Hakanen M, Amin N, Cavalcanti-Proença C, Grøntved A, Hallmans G, Jansson JO, Kuusisto J, Kähönen M, Lutsey PL, Nolan JJ, Palla L, Pedersen O, Pérusse L, Renström F, Scott RA, Shungin D, Sovio U, Tammelin TH, Rönnemaa T, Lakka TA, Uusitupa M, Rios MS, Ferrucci L, Bouchard C, Meirhaeghe A, Fu M, Walker M, Borecki IB, Dedoussis GV, Fritsche A, Ohlsson C, Boehnke M, Bandinelli S, van Duijn CM, Ebrahim S, Lawlor DA, Gudnason V, Harris TB, Sørensen TI, Mohlke KL, Hofman A, Uitterlinden AG, Tuomilehto J, Lehtimäki T, Raitakari O, Isomaa B, **Njølstad PR**, Florez JC, Liu S, Ness A, Spector TD, Tai ES, Froguel P, Boeing H, Laakso M, Marmot M, Bergmann S, Power C, Khaw KT, Chasman D, Ridker P, Hansen T, Monda KL, Illig T, Järvelin MR, Wareham NJ, Hu FB, Groop LC, Orho-Melander M, Ekelund U, Franks PW, Loos RJ. Physical activity attenuates the influence of FTO variants on obesity risk: a meta-analysis of 218,166 adults and 19,268 children. *PLoS Med*. 2011 Nov;8(11):e1001116. PMID: 22069379.

108: Thanabalasingham G, Shah N, Vaxillaire M, Hansen T, Tuomi T, Gašperíková D, Szopa M, Tjora E, James TJ, Kokko P, Loiseleur F, Andersson E, Gaget S, Isomaa B, Nowak N, Raeder H, Stanik J, **Njølstad PR**, Malecki MT, Klimes I, Groop L, Pedersen O, Froguel P, McCarthy MI, Gloyn AL, Owen KR. A large multi-centre European study validates high-sensitivity C-reactive protein (hsCRP) as a clinical biomarker for the diagnosis of diabetes subtypes. *Diabetologia*. 2011 Nov;54(11):2801-10. PMID: 21814873.

109: Shahawy S, Chan NK, Ellard S, Young E, Shahawy H, Mace J, Peverini R, Chinnock R, **Njølstad PR**, Hattersley AT, Hathout E. A pathway to insulin independence in newborns and infants with diabetes. *J Perinatol*. 2011 Aug;31(8):567-70. PMID: 21796147.

110: Johansson BB, Torsvik J, Bjørkhaug L, Vesterhus M, Ragvin A, Tjora E, Fjeld K, Hoem D, Johansson S, Ræder H, Lindquist S, Hernell O, Cnop M, Saraste J, Flatmark T, Molven A, **Njølstad PR**. 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. *J Biol Chem*. 2011 Oct 7;286(40):34593-605. PMID: 21784842.

111: Gonc EN, Ozturk BB, Haldorsen IS, Molnes J, Immervoll H, Raeder H, Molven A, Søvik O, **Njølstad PR**. HNF1B mutation in a Turkish child with renal and exocrine pancreas insufficiency, diabetes and liver disease. *Pediatr Diabetes*. 2012 Mar;13(2):e1-5. PMID: 21767339.

Publications Pål Rasmus Njølstad 1987-2021

112: Trewick AL, Moustafa JS, de Smith AJ, Froguel P, Greve G, **Njølstad PR**, Coin LJ, Blakemore AI. Accurate single-nucleotide polymorphism allele assignment in trisomic or duplicated regions by using a single base-extension assay with MALDI-TOF mass spectrometry. *Clin Chem*. 2011 Aug;57(8):1188-95. PMID: 21677093.

113: Molnes J, Teigen K, Aukrust I, Bjørkhaug L, Søvik O, Flatmark T, **Njølstad PR**. Binding of ATP at the active site of human pancreatic glucokinase--nucleotide-induced conformational changes with possible implications for its kinetic cooperativity. *FEBS J*. 2011 Jul;278(13):2372-86. PMID: 21569204.

114: Søvik O, Aagenaes O, Eide SÅ, Mackay D, Temple IK, Molven A, **Njølstad PR**. Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. *Pediatr Diabetes*. 2012 Mar;13(2):155-62. PMID: 21518169.

115: Molven A, **Njølstad PR**. Role of molecular genetics in transforming diagnosis of diabetes mellitus. *Expert Rev Mol Diagn*. 2011 Apr;11(3):313-20. PMID: 21463240.

116: Hertel JK, Johansson S, Sonestedt E, Jonsson A, Lie RT, Platou CG, Nilsson PM, Rukh G, Midthjell K, Hveem K, Melander O, Groop L, Lyssenko V, Molven A, Orho-Melander M, **Njølstad PR**. FTO, type 2 diabetes, and weight gain throughout adult life: a meta-analysis of 41,504 subjects from the Scandinavian HUNT, MDC, and MPP studies. *Diabetes*. 2011 May;60(5):1637-44. PMID: 21398525.

117: Hertel JK, Johansson S, Ræder H, Platou CG, Midthjell K, Hveem K, Molven A, **Njølstad PR**. Evaluation of four novel genetic variants affecting hemoglobin A1c levels in a population-based type 2 diabetes cohort (the HUNT2 study). *BMC Med Genet*. 2011 Feb 4;12:20. PMID: 21294870.

118: Pörksen S, Laborie LB, Nielsen L, Louise Max Andersen M, Sandal T, de Wet H, Schwarcz E, Aman J, Swift P, Kocova M, Schönle EJ, de Beaufort C, Hougaard P, Ashcroft F, Molven A, Knip M, Mortensen HB, Hansen L, **Njølstad PR**; Hvidøre Study Group on Childhood Diabetes. Disease progression and search for monogenic diabetes among children with new onset type 1 diabetes negative for ICA, GAD- and IA-2 Antibodies. *BMC Endocr Disord*. 2010 Sep 23;10:16. PMID: 20863361.

119: Vesterhus M, Raeder H, Kurpad AJ, Kawamori D, Molven A, Kulkarni RN, Kahn CR, **Njølstad PR**. Pancreatic function in carboxyl-ester lipase knockout mice. *Pancreatology*. 2010;10(4):467-76. PMID: 20720448.

120: Sherif EM, Abdelmaksoud AA, Elbarbary NS, **Njølstad PR**. An Egyptian case of congenital hyperinsulinism of infancy due to a novel mutation in KCNJ11 encoding Kir6.2 and response to octreotide. *Acta Diabetol*. 2013 Oct;50(5):801-5. PMID: 20686794.

121: **Njølstad PR**, Hertel JK, Søvik O, Raeder H, Johansson S, Molven A. [Progress in diabetes genetics]. *Tidsskr Nor Laegeforen*. 2010 Jun 3;130(11):1145-9. Review. PMID: 20531501.

122: Ragvin A, Moro E, Fredman D, Navratilova P, Drivenes Ø, Engström PG, Alonso ME, de la Calle Mustienes E, Gómez Skarmeta JL, Tavares MJ, Casares F, Manzanares M, van Heyningen V, Molven A, **Njølstad PR**, Argenton F, Lenhard B, Becker TS. Long-range gene regulation links genomic type 2 diabetes and obesity risk regions to HHEX, SOX4, and IRX3. *Proc Natl Acad Sci U S A*. 2010 Jan 12;107(2):775-80. PMID: 20080751.

123 Molven A, Søvik O, von der Lippe C, Steine SJ, **Njølstad PR**, Houge G, Prescott TE. [Molecular genetic diagnostics in syndromes associated with the RAS/MAPK signalling pathway]. *Tidsskr Nor Laegeforen*. 2009 Nov 19;129(22):2358-61. PMID: 19935936.

124: Hoey H; Hvidoere Study Group on Childhood Diabetes [incl. **Njølstad PR**]. Psychosocial factors are associated with metabolic control in adolescents: research from the Hvidoere Study Group on Childhood Diabetes. *Pediatr Diabetes*. 2009 Dec;10 Suppl 13:9-14. PMID: 19930221.

125: Swift PG, Skinner TC, de Beaufort CE, Cameron FJ, Aman J, Aanstoot HJ, Castaño L, Chiarelli F, Daneman D, Danne T, Dorchy H, Hoey H, Kaprio EA, Kaufman F, Kocova M, Mortensen HB, **Njølstad PR**, Phillip M, Robertson KJ, Schoenle EJ, Urakami T, Vanelli M, Ackermann RW, Skovlund SE; Hvidoere Study Group on Childhood Diabetes. Target setting in intensive insulin management is associated with metabolic control: the Hvidoere childhood diabetes study group centre differences study 2005. *Pediatr Diabetes*. 2010 Jun;11(4):271-8. PMID: 19895567.

126: Lango Allen H, Johansson S, Ellard S, Shields B, Hertel JK, Raeder H, Colclough K, Molven A, Frayling TM, **Njølstad PR**, Hattersley AT, Weedon MN. Polygenic risk variants for type 2 diabetes susceptibility modify age at diagnosis in monogenic HNF1A diabetes. *Diabetes*. 2010 Jan;59(1):266-71. PMID: 19794065.

127: Torsvik J, Johansson S, Johansen A, Ek J, Minton J, Raeder H, Ellard S, Hattersley A, Pedersen O, Hansen T, Molven A, **Njølstad PR**. Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. *Hum Genet*. 2010 Jan;127(1):55-64. PMID: 19760265.

128: Hattersley A, Bruining J, Shield J, **Njølstad PR**, Donaghue KC. The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatr Diabetes*. 2009 Sep;10 Suppl 12:33-42. PMID: 19754616.

129: Skinningsrud B, Lie BA, Husebye ES, Kvien TK, Førre Ø, Flatø B, Stormyr A, Joner G, **Njølstad PR**, Egeland T, Undlien DE. A CLEC16A variant confers risk for juvenile idiopathic arthritis and anti-cyclic citrullinated peptide antibody negative rheumatoid arthritis. *Ann Rheum Dis*. 2010 Aug;69(8):1471-4. PMID: 19734133.

130: **Njølstad PR**, Molven A, Groop L. Diabetes genetics. A seventh sense for the successful sequel of 'come together'. "The genotypes and phenotypes of diabetes". Bergen, Norway. April 22-26, 2009. 2nd meeting of 'EASD Study Group on Genetics of Diabetes' and 44th annual meeting of the 'Scandinavian Society for the Study of Diabetes'. *JOP*. 2009 Jul 6;10(4):466-71. PMID: 19581761.

131: Laborie LB, Mackay DJ, Temple IK, Molven A, Søvik O, **Njølstad PR**. DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. *Eur J Pediatr*. 2010 Feb;169(2):207-13. PMID: 19521719.

132: Sandal T, Laborie LB, Brusgaard K, Eide SA, Christesen HB, Søvik O, **Njølstad PR**, Molven A. The spectrum of ABCC8 mutations in Norwegian patients with congenital hyperinsulinism of infancy. *Clin Genet*. 2009 May;75(5):440-8. PMID: 19475716.

133: Lacbawan F, Solomon BD, Roessler E, El-Jaick K, Domené S, Vélez JI, Zhou N, Hadley D, Balog JZ, Long R, Fryer A, Smith W, Omar S, McLean SD, Clarkson K, Lichty A, Clegg NJ, Delgado MR, Levey E, Stashinko E, Potocki L, Vanallen MI, Clayton-Smith J, Donnai D, Bianchi DW, Juliusson PB, **Njølstad PR**, Brunner HG, Carey JC, Hehr U, Müsebeck J, Wieacker PF, Postra A, Hennekam RC, van den Boogaard MJ, van Haeringen A, Paulussen A, Herbergs J, Schrander-Stumpel CT, Janecke AR, Chitayat D, Hahn J, McDonald-McGinn DM, Zackai EH, Dobyns WB, Muenke M. Clinical spectrum of SIX3-associated mutations in holoprosencephaly: correlation between genotype, phenotype and function. *J Med Genet*. 2009 Jun;46(6):389-98. PMID: 19346217.

Publications Pål Rasmus Njølstad 1987-2021

134: Bjørnvold M, Munthe-Kaas MC, Egeland T, Joner G, Dahl-Jørgensen K, **Njølstad PR**, Akselsen HE, Gervin K, Carlsen KC, Carlsen KH, Undlien DE. A TLR2 polymorphism is associated with type 1 diabetes and allergic asthma. *Genes Immun*. 2009 Mar;10(2):181-7. PMID: 19148143.

135: Aman J, Skinner TC, de Beaufort CE, Swift PG, Aanstoot HJ, Cameron F; Hvidoere Study Group on Childhood Diabetes [incl. **Njølstad PR**]. Associations between physical activity, sedentary behavior, and glycemic control in a large cohort of adolescents with type 1 diabetes: the Hvidoere Study Group on Childhood Diabetes. *Pediatr Diabetes*. 2009 Jun;10(4):234-9. PMID: 19140898.

136: Søvik O, Schubbert S, Houge G, Steine SJ, Norgård G, Engelsen B, **Njølstad PR**, Shannon K, Molven A. De novo HRAS and KRAS mutations in two siblings with short stature and neuro-cardio-facio-cutaneous features. *BMJ Case Rep*. 2009;2009. pii: bcr07.2008.0550. PMID: 21686750.

137: Magitta NF, Bøe Wolff AS, Johansson S, Skinningsrud B, Lie BA, Myhr KM, Undlien DE, Joner G, **Njølstad PR**, Kvien TK, Førre Ø, Knappskog PM, Husebye ES. A coding polymorphism in NALP1 confers risk for autoimmune Addison's disease and type 1 diabetes. *Genes Immun*. 2009 Mar;10(2):120-4. PMID: 18946481.

138: Øverby NC, Margeirsdottir HD, Brunborg C, Dahl-Jørgensen K, Andersen LF; Norwegian Study Group for Childhood Diabetes [incl. **Njølstad PR**]. Sweets, snacking habits, and skipping meals in children and adolescents on intensive insulin treatment. *Pediatr Diabetes.* 2008 Aug;9(4 Pt 2):393-400. PMID: 18774998.

139: Haldorsen IS, Vesterhus M, Raeder H, Jensen DK, Søvik O, Molven A, **Njølstad PR**. Lack of pancreatic body and tail in HNF1B mutation carriers. *Diabet Med*. 2008 Jul;25(7):782-7. PMID: 18644064.

140: Vesterhus M, Haldorsen IS, Raeder H, Molven A, **Njølstad PR**. Reduced pancreatic volume in hepatocyte nuclear factor 1A-maturity-onset diabetes of the young. *J Clin Endocrinol Metab*. 2008 Sep;93(9):3505-9. PMID: 18593771.

141: Vesterhus M, Raeder H, Aurlien H, Gjesdal CG, Bredrup C, Holm PI, Molven A, Bindoff L, Berstad A, **Njølstad PR**. Neurological features and enzyme therapy in patients with endocrine and exocrine pancreas dysfunction due to CEL mutations. *Diabetes* Care. 2008 Sep;31(9):1738-40. PMID: 18544793.

142: Eide SA, Raeder H, Johansson S, Midthjell K, Søvik O, **Njølstad PR**, Molven A. Prevalence of HNF1A (MODY3) mutations in a Norwegian population (the HUNT2 Study). *Diabet Med*. 2008 Jul;25(7):775-81. PMID: 18513305.

143: Søvik O, **Njølstad PR**, Jellum E, Molven A. Wolcott-Rallison syndrome with 3-hydroxydicarboxylic aciduria and lethal outcome. *J Inherit Metab Dis*. 2008 Dec;31 Suppl 2:S293-7. PMID: 18500571.

144: Christesen HB, Tribble ND, Molven A, Siddiqui J, Sandal T, Brusgaard K, Ellard S, **Njølstad PR**, Alm J, Brock Jacobsen B, Hussain K, Gloyn AL. Activating glucokinase (GCK) mutations as a cause of medically responsive congenital hyperinsulinism: prevalence in children and characterisation of a novel GCK mutation. *Eur J Endocrinol*. 2008 Jul;159(1):27-34. PMID: 18450771.

145: Hertel JK, Johansson S, Raeder H, Midthjell K, Lyssenko V, Groop L, Molven A, **Njølstad PR**. Genetic analysis of recently identified type 2 diabetes loci in 1,638 unselected patients with type 2 diabetes and 1,858 control participants from a Norwegian population-based cohort (the HUNT study). *Diabetologia*. 2008 Jun;51(6):971-7. PMID: 18437351.

146: Sagen JV, Bjørkhaug L, Molnes J, Raeder H, Grevle L, Søvik O, Molven A, **Njølstad PR**. Diagnostic screening of MODY2/GCK mutations in the Norwegian MODY
Registry. *Pediatr Diabetes*. 2008 Oct;9(5):442-9. PMID: 18399931.

147: Molnes J, Bjørkhaug L, Søvik O, **Njølstad PR**, Flatmark T. Catalytic activation of human glucokinase by substrate binding: residue contacts involved in the binding of D-glucose to the super-open form and conformational transitions. *FEBS J*. 2008 May;275(10):2467-81. PMID: 18397317.

148: Harries LW, Locke JM, Shields B, Hanley NA, Hanley KP, Steele A, **Njølstad PR**, Ellard S, Hattersley AT. The diabetic phenotype in HNF4A mutation carriers is moderated by the expression of HNF4A isoforms from the P1 promoter during fetal development. *Diabetes*. 2008 Jun;57(6):1745-52. PMID: 18356407.

149: Ellard S, Bellanné-Chantelot C, Hattersley AT; European Molecular Genetics Quality Network (EMQN) MODY group [incl. **Njølstad PR**]. Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. *Diabetologia*. 2008 Apr;51(4):546-53. PMID: 18297260.

150: Cameron FJ, Skinner TC, de Beaufort CE, Hoey H, Swift PG, Aanstoot H, Aman J, Martul P, Chiarelli F, Daneman D, Danne T, Dorchy H, Kaprio EA, Kaufman F, Kocova M, Mortensen HB, **Njølstad PR**, Phillip M, Robertson KJ, Schoenle EJ, Urakami T, Vanelli M, Ackermann RW, Skovlund SE; Hvidoere Study Group on Childhood Diabetes. Are family factors universally related to metabolic outcomes in adolescents with Type 1 diabetes? *Diabet Med*. 2008 Apr;25(4):463-8. PMID: 18294223.

151: Bjørnvold M, Undlien DE, Joner G, Dahl-Jørgensen K, **Njølstad PR**, Akselsen HE, Gervin K, Rønningen KS, Stene LC. Joint effects of HLA, INS, PTPN22 and CTLA4 genes on the risk of type 1 diabetes. *Diabetologia*. 2008 Apr;51(4):589-96. PMID: 18292987.

152: Molven A, Ringdal M, Nordbø AM, Raeder H, Støy J, Lipkind GM, Steiner DF, Philipson LH, Bergmann I, Aarskog D, Undlien DE, Joner G, Søvik O; Norwegian Childhood Diabetes Study Group, Bell GI, **Njølstad PR**. Mutations in the insulin gene can cause MODY and autoantibody-negative type 1 diabetes. *Diabetes*. 2008 Apr;57(4):1131-5. PMID: 18192540.

153: Vesterhus M, Raeder H, Johansson S, Molven A, **Njølstad PR**. Pancreatic exocrine dysfunction in maturity-onset diabetes of the young type 3. *Diabetes Care*. 2008 Feb;31(2):306-10. PMID: 17989309.

154: Søvik O, Tansek MZ, Sagen JV, **Njølstad PR**. Management of neonatal and infancy-onset diabetes mellitus. *Endocr Dev*. 2007;11:94-105. Review. PMID: 17986830.

155: Johansson S, Raeder H, Eide SA, Midthjell K, Hveem K, Søvik O, Molven A, **Njølstad PR**. 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 Dec;56(12):3112-7. PMID: 17827402.

156: Søvik O, Schubbert S, Houge G, Steine SJ, Norgård G, Engelsen B, **Njølstad PR**, Shannon K, Molven A. De novo HRAS and KRAS mutations in two siblings with short stature and neuro-cardio-facio-cutaneous features. *J Med Genet*. 2007 Jul;44(7):e84. PMID: 17601930.

157: Bjørkhaug L, Molnes J, Søvik O, **Njølstad PR**, Flatmark T. Allosteric activation of human glucokinase by free polyubiquitin chains and its ubiquitin-dependent cotranslational proteasomal degradation. *J Biol Chem*. 2007 Aug 3;282(31):22757-64. PMID: 17561510.

158: de Beaufort CE, Swift PG, Skinner CT, Aanstoot HJ, Aman J, Cameron F, Martul P, Chiarelli F, Daneman D, Danne T, Dorchy H, Hoey H, Kaprio EA, Kaufman F, Kocova M, Mortensen HB, **Njølstad PR**, Phillip M, Robertson KJ, Schoenle EJ, Urakami T, Vanelli M; Hvidoere Study Group on Childhood Diabetes 2005. Continuing stability of center differences in pediatric diabetes care: do advances in diabetes treatment improve outcome? The Hvidoere Study Group on Childhood Diabetes. *Diabetes Care*. 2007 Sep;30(9):2245-50. PMID: 17540955.

159: Aamodt G, Stene LC, **Njølstad PR**, Søvik O, Joner G; Norwegian Childhood Diabetes Study Group. Spatiotemporal trends and age-period-cohort modeling of the incidence of type 1 diabetes among children aged <15 years in Norway 1973-1982 and 1989-2003. *Diabetes Care*. 2007 Apr;30(4):884-9. PMID: 17392550.

160: Raeder H, Haldorsen IS, Ersland L, Grüner R, Taxt T, Søvik O, Molven A, **Njølstad PR**. Pancreatic lipomatosis is a structural marker in nondiabetic children with mutations in carboxyl-ester lipase. *Diabetes*. 2007 Feb;56(2):444-9. PMID: 17259390.

161: Hattersley A, Bruining J, Shield J, **Njølstad PR**, Donaghue K; International Society for Pediatric and Adolescent Diabetes. ISPAD Clinical Practice Consensus Guidelines 2006-2007. The diagnosis and management of monogenic diabetes in children. *Pediatr Diabetes*. 2006 Dec;7(6):352-60. PMID: 17212604.

162: Bjørnvold M, Amundsen SS, Stene LC, Joner G, Dahl-Jørgensen K, **Njølstad PR**, Ek J, Ascher H, Gudjònsdòttir AH, Lie BA, Skinningsrud B, Akselsen HE, Rønningen KS, Sollid LM, Undlien DE. FOXP3 polymorphisms in type 1 diabetes and coeliac disease. *J Autoimmun*. 2006 Sep;27(2):140-4. PMID: 16996248.

163: Pearson ER, Flechtner I, **Njølstad PR**, Malecki MT, Flanagan SE, Larkin B, Ashcroft FM, Klimes I, Codner E, Iotova V, Slingerland AS, Shield J, Robert JJ, Holst JJ, Clark PM, Ellard S, Søvik O, Polak M, Hattersley AT; Neonatal Diabetes International Collaborative Group. Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations*. N Engl J Med.* 2006 Aug 3;355(5):467-77. PMID: 16885550.

164: Mackay DJ, Boonen SE, Clayton-Smith J, Goodship J, Hahnemann JM, Kant SG, **Njølstad PR**, Robin NH, Robinson DO, Siebert R, Shield JP, White HE, Temple IK. A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. *Hum Genet*. 2006 Sep;120(2):262-9. PMID: 16816970.

165: Hathout E, Mace J, Bell GI, **Njølstad PR**. Treatment of hyperglycemia in a 7-year-old child diagnosed with neonatal diabetes. *Diabetes Care*. 2006 Jun;29(6):1458. PMID: 16732049.

166: Raeder H, Bjørkhaug L, Johansson S, Mangseth K, Sagen JV, Hunting A, Følling I, Johansen O, Bjørgaas M, Paus PN, Søvik O, Molven A, **Njølstad PR**. 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 Jun;55(6):1899-903. PMID: 16731861.

167: Proks P, Girard C, Baevre H, **Njølstad PR**, Ashcroft FM. Functional effects of mutations at F35 in the NH2-terminus of Kir6.2 (KCNJ11), causing neonatal diabetes, and response to sulfonylurea therapy. *Diabetes*. 2006 Jun;55(6):1731-7. PMID: 16731836.

168: Sagen JV, Odili S, Bjørkhaug L, Zelent D, Buettger C, Kwagh J, Stanley C, Dahl-Jørgensen K, de Beaufort C, Bell GI, Han Y, Grimsby J, Taub R, Molven A, Søvik O, **Njølstad PR**, Matschinsky FM. From clinicogenetic studies of maturity-onset diabetes of the young to unraveling complex mechanisms of glucokinase regulation. *Diabetes*. 2006 Jun;55(6):1713-22. PMID: 16731834.

169: Raeder H, Johansson S, Holm PI, Haldorsen IS, Mas E, Sbarra V, Nermoen I, Eide SA, Grevle L, Bjørkhaug L, Sagen JV, Aksnes L, Søvik O, Lombardo D, Molven A, **Njølstad PR**. Mutations in the CEL VNTR cause a syndrome of diabetes and pancreatic exocrine dysfunction. *Nat Genet*. 2006 Jan;38(1):54-62. PMID: 16369531.

170: Bjørkhaug L, Johansson S, Raeder H, Thorsby PM, Undlien DE, Søvik O, Molven A, Sagen JV, **Njølstad PR**. [Molecular diagnostics in diabetes mellitus]. *Tidsskr Nor Laegeforen*. 2005 Nov 3;125(21):2968-72. Review. PubMed PMID: 16276383.

171: Bjørkhaug L, Bratland A, **Njølstad PR**, Molven A. Functional dissection of the HNF-1alpha transcription factor: a study on nuclear localization and transcriptional activation. *DNA Cell Biol*. 2005 Nov;24(11):661-9. PMID: 16274290.

172: Sagen JV, Baumann ME, Salvesen HB, Molven A, Søvik O, **Njølstad PR**. Diagnostic screening of NEUROD1 (MODY6) in subjects with MODY or gestational diabetes mellitus. *Diabet Med*. 2005 Aug;22(8):1012-5. PMID: 16026366.

173: Shehadeh N, Bakri D, **Njølstad PR**, Gershoni-Baruch R. Clinical characteristics of mutation carriers in a la2rge family with glucokinase diabetes (MODY2). *Diabet Med*. 2005 Aug;22(8):994-8. PMID: 16026363.

174: Tammaro P, Girard C, Molnes J, **Njølstad PR**, Ashcroft FM. Kir6.2 mutations causing neonatal diabetes provide new insights into Kir6.2-SUR1 interactions. *EMBO J*. 2005 Jul 6;24(13):2318-30. PMID: 15962003.

175: Sagen JV, Pearson ER, Johansen A, Spyer G, Søvik O, Pedersen O, **Njølstad PR**, Hattersley AT, Hansen T. Preserved insulin response to tolbutamide in hepatocyte nuclear factor-1alpha mutation carriers. *Diabet Med*. 2005 Apr;22(4):406-9. PMID: 15787664.

176: Sagen JV, Raeder H, Hathout E, Shehadeh N, Gudmundsson K, Baevre H, Abuelo D, Phornphutkul C, Molnes J, Bell GI, Gloyn AL, Hattersley AT, Molven A, Søvik O, **Njølstad PR**. Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. *Diabetes*. 2004 Oct;53(10):2713-8. PMID: 15448106.

177: Gloyn AL, Pearson ER, Antcliff JF, Proks P, Bruining GJ, Slingerland AS, Howard N, Srinivasan S, Silva JM, Molnes J, Edghill EL, Frayling TM, Temple IK, Mackay D, Shield JP, Sumnik Z, van Rhijn A, Wales JK, Clark P, Gorman S, Aisenberg J, Ellard S, **Njølstad PR**, Ashcroft FM, Hattersley AT. Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. *N Engl J Med.* 2004 Apr 29;350(18):1838-49. PMID: 15115830.

178: Molven A, Matre GE, Duran M, Wanders RJ, Rishaug U, **Njølstad PR**, Jellum E, Søvik O. Familial hyperinsulinemic hypoglycemia caused by a defect in the SCHAD enzyme of mitochondrial fatty acid oxidation. *Diabetes.* 2004 Jan;53(1):221-7.
PMID: 14693719.

179: So WY, Ng MC, Horikawa Y, **Njølstad PR**, Li JK, Ma RC, Bell GI, Chan JC. Genetic variants of hepatocyte nuclear factor-1beta in Chinese young-onset diabetic patients with nephropathy. *J Diabetes Complications*. 2003 Nov-Dec;17(6):369-73. PMID: 14583183.

180: **Njølstad PR**, Sagen JV, Bjørkhaug L, Odili S, Shehadeh N, Bakry D, Sarici SU, Alpay F, Molnes J, Molven A, Søvik O, Matschinsky FM. Permanent neonatal diabetes caused by glucokinase deficiency: inborn error of the glucose-insulin signaling pathway. *Diabetes.* 2003 Nov;52(11):2854-60. PMID: 14578306.

181: Sagen JV, Bostad L, **Njølstad PR**, Søvik O. Enlarged nephrons and severe nondiabetic nephropathy in hepatocyte nuclear factor-1beta (HNF-1beta) mutation carriers. *Kidney Int*. 2003 Sep;64(3):793-800. PMID: 12911528.

182: Søvik O, Sagen J, **Njølstad PR**, Nyland H, Myhr KM. Contributions to the MODY5 phenotype. *J Inherit Metab Dis*. 2002 Nov;25(7):597-8. PMID: 12638944.

183: Bjørkhaug L, Sagen JV, Thorsby P, Søvik O, Molven A, **Njølstad PR**. Hepatocyte nuclear factor-1 alpha gene mutations and diabetes in Norway. *J Clin Endocrinol Metab.* 2003 Feb;88(2):920-31. PMID: 12574234.

184: Molven A, Rishaug U, Matre GE, **Njølstad PR**, Søvik O. Hunting for a hypoglycemia gene: severe neonatal hypoglycemia in a consanguineous family. *Am J Med Genet.* 2002 Nov 15;113(1):40-6. PMID: 12400064.

185: Sagen JV, **Njølstad PR**, Søvik O. Reduced prevalence of late-diabetic complications in MODY3 with early diagnosis. *Diabet Med*. 2002 Aug;19(8):697-8. PMID: 12147154.

186: Stride A, Vaxillaire M, Tuomi T, Barbetti F, **Njølstad PR**, Hansen T, Costa A, Conget I, Pedersen O, Søvik O, Lorini R, Groop L, Froguel P, Hattersley AT. The genetic abnormality in the beta cell determines the response to an oral glucose load. *Diabetologia.* 2002 Mar;45(3):427-35. PMID: 11914749.

187: **Njølstad PR**. [Just children?]. *Tidsskr Nor Laegeforen*. 2001 Sep 20;121(22):2575. Norwegian. PMID: 11668748.

188: **Njølstad PR**, Søvik O, Cuesta-Muñoz A, Bjørkhaug L, Massa O, Barbetti F, Undlien DE, Shiota C, Magnuson MA, Molven A, Matschinsky FM, Bell GI. Neonatal diabetes mellitus due to complete glucokinase deficiency. *N Engl J Med.* 2001 May 24;344(21):1588-92. PMID: 11372010.

189: Søvik O, **Njølstad PR**, Reigstad H, Brackman D, Teslo I, Brunvand L. [Diagnosis and treatment of congenital hyperinsulinism--to Paris at any price?]. *Tidsskr Nor Laegeforen.* 2001 Feb 20;121(5):612-4. PubMed PMID: 11301621.

190: Bjerknes R, Skadberg B, **Njølstad PR**. [Asthma, steroid inhalation and growth]. *Tidsskr Nor Laegeforen*. 2001 Jan 30;121(3):281. PMID: 11242865.

191: Bjørkhaug L, Ye H, Horikawa Y, Søvik O, Molven A, **Njølstad PR**. MODY associated with two novel hepatocyte nuclear factor-1alpha loss-of-function mutations (P112L and Q466X). *Biochem Biophys Res Commun*. 2000 Dec 29;279(3):792-8. PMID: 11162430.

192: Baptista MJ, Fairbrother UL, Howard CM, Farrer MJ, Davies GE, Trikka D, Maratou K, Redington A, Greve G, **Njølstad PR**, Kessling AM. Heterotrisomy, a significant contributing factor to ventricular septal defect associated with Down syndrome? *Hum Genet*. 2000 Nov;107(5):476-82. PMID: 11140945.

193: Bjørkhaug L, Søvik O, Bell GI, **Njølstad PR**, Molven A. A simple test for the high-frequency P291fsinsC mutation in the HNF1 alpha/MODY3 gene. *Diabetologia*. 2000 Jun;43(6):818-9. PMID: 10907131.

194: Lindner TH, **Njølstad PR**, Horikawa Y, Bostad L, Bell GI, Sovik O. A novel syndrome of diabetes mellitus, renal dysfunction and genital malformation associated with a partial deletion of the pseudo-POU domain of hepatocyte nuclear factor-1beta. *Hum Mol Genet*. 1999 Oct;8(11):2001-8. PMID: 10484768.

195: Johnson D, Horsley SW, Moloney DM, Oldridge M, Twigg SR, Walsh S, Barrow M, **Njølstad PR**, Kunz J, Ashworth GJ, Wall SA, Kearney L, Wilkie AO. A comprehensive screen for TWIST mutations in patients with craniosynostosis identifies a new microdeletion syndrome of chromosome band 7p21.1. *Am J Hum Genet.* 1998 Nov;63(5):1282-93. PubMed PMID: 9792856; PubMed Central PMCID: PMC1377539.

196: **Njølstad PR**, Cockburn BN, Bell GI, Søvik O. A missense mutation, Val62Ala, in the glucokinase gene in a Norwegian family with maturity-onset diabetes of the young. *Acta Paediatr*. 1998 Aug;87(8):853-6. PMID: 9736233.

197: **Njølstad PR**, Reigstad H, Westby J, Espeland A. Familial non-immune hydrops fetalis and congenital pulmonary lymphangiectasia. *Eur J Pediatr*. 1998 Jun;157(6):498-501. PMID: 9667408.

198: Søvik O, **Njølstad PR**, Følling I, Sagen J, Cockburn BN, Bell GI. Hyperexcitability to sulphonylurea in MODY3. *Diabetologia.* 1998 May;41(5):607-8. PMID: 9628283.

199: **Njølstad PR**, Søvik O, Bell GI, Cockburn BN, Følling I, Sagen J. [Non-insulin dependent diabetes in children and adolescents]. *Tidsskr Nor Laegeforen*. 1998 Mar 10;118(7):1054-7. PMID: 9531829.

200: Hassoun H, Vassiliadis JN, Murray J, **Njølstad PR**, Rogus JJ, Ballas SK, Schaffer F, Jarolim P, Brabec V, Palek J. Characterization of the underlying molecular defect in hereditary spherocytosis associated with spectrin deficiency. *Blood.* 1997 Jul 1;90(1):398-406. PMID: 9207476.

201: **Njølstad PR**, Skjeldal OH, Agsteribbe E, Huckriede A, Wannag E, Søvik O, Waaler PE. Medium chain acyl-CoA dehydrogenase deficiency and fatal valproate toxicity. *Pediatr Neurol*. 1997 Feb;16(2):160-2. PMID: 9090694.

202: **Njølstad PR**, Molven A. [The homeo box--a genetic key to fetal development]. *Tidsskr Nor Laegeforen*. 1993 May 10;113(12):1475-9. Review. PMID: 8101395.

203: Molven A, Hordvik I, **Njølstad PR**. Sequence analysis of the zebrafish hox-B5/B6 region. *Biochim Biophys Acta*. 1993 Apr 29;1173(1):102-6. PMID: 8097929.

204: Salvesen H, **Njølstad PR**, Akslen LA, Albrektsen G, Søreide O, Varhaug JE. Papillary thyroid carcinoma: a multivariate analysis of prognostic factors including an evaluation of the p-TNM staging system. *Eur J Surg*. 1992 Nov-Dec;158(11-12):583-9. PMID: 1363062.

205: Fjose A, **Njølstad PR**, Nornes S, Molven A, Krauss S. Structure and early embryonic expression of the zebrafish engrailed-2 gene. *Mech Dev.* 1992 Nov;39(1-2):51-62. PMID: 1362650.

206: Molven A, Hordvik I, **Njølstad PR**, van Ghelue M, Fjose A. The zebrafish homeobox gene hox[zf-114]: primary structure, expression pattern and evolutionary aspects. *Int J Dev Biol*. 1992 Jun;36(2):229-37. PMID: 1356006.

207: **Njølstad PR**, Vik IS, Nordbø SA. [Primary HIV-infection. A differential diagnosis of mononucleosis/influenza-like symptoms]. *Tidsskr Nor Laegeforen*. 1992 Apr 30;112(11):1446-8. PMID: 1631817.

208: Salvesen H, **Njølstad PR**, Akslen LA, Albrektsen G, Viste A, Søreide O, Varhaug JE. Thyroid carcinoma: results from surgical treatment in 211 consecutive patients. *Eur J Surg.* 1991 Sep;157(9):521-6. PMID: 1683575.

Publications Pål Rasmus Njølstad 1987-2021

209: Molven A, **Njølstad PR**, Fjose A. Genomic structure and restricted neural expression of the zebrafish wnt-1 (int-1) gene. *EMBO J.* 1991 Apr;10(4):799-807. PMID: 2009859.

210: **Njølstad PR**. [The molecular function of oncogenes]. *Tidsskr Nor Laegeforen*. 1990 Nov 20;110(28):3632-6. Review. PMID: 2260064.

211: **Njølstad PR**. [Transcription regulation--how are genes turned on and off?]. *Tidsskr Nor Laegeforen.* 1990 Oct 30;110(26):3366-71. Review. PMID: 2256060.

212: **Njølstad PR**, Molven A, Apold J, Fjose A. The zebrafish homeobox gene hox-2.2: transcription unit, potential regulatory regions and in situ localization of transcripts. *EMBO J.* 1990 Feb;9(2):515-24. PMID: 1968004.

213: **Njølstad PR**, Fjose A. In situ hybridization patterns of zebrafish homeobox genes homologous to Hox-2.1 and En-2 of mouse. *Biochem Biophys Res Commun*. 1988 Dec 15;157(2):426-32. PMID: 2904810.

214: **Njølstad PR**, Molven A, Eiken HG, Fjose A. Structure and neural expression of a zebrafish homeobox sequence. *Gene*. 1988 Dec 15;73(1):33-46. PMID: 2468579.

215: **Njølstad PR**, Molven A, Hordvik I, Apold J, Fjose A. Primary structure, developmentally regulated expression and potential duplication of the zebrafish homeobox gene ZF-21. *Nucleic Acids Res*. 1988 Oct 11;16(19):9097-111. PMID: 2902580.

216: Fjose A, Eiken HG, **Njølstad PR**, Molven A, Hordvik I. A zebrafish engrailed-like homeobox sequence expressed during embryogenesis. *FEBS Lett.* 1988 Apr 25;231(2):355-60. PMID: 2896133.

217: **Njølstad PR**, Molven A, Fjose A. A zebrafish homologue of the murine Hox-2.1 gene. *FEBS Lett*. 1988 Mar 28;230(1-2):25-30. PMID: 2895022.

218: Eiken HG, **Njølstad PR**, Molven A, Fjose A. A zebrafish homeobox-containing gene with embryonic transcription*. Biochem Biophys Res Commun*. 1987 Dec 31;149(3):1165-71. PMID: 2892487.

Review Papers, Popular Science & Various Papers in Norwegian

1: **Njølstad PR**. Structural and functional analysis of vertebrate homeobox genes using the zebrafish (*Brachydanio rerio*) as a model. Avhandling for den medisinske doktorgrad, Åsane Trykkeri A/S, Bergen 1989.
2: **Njølstad PR**. Transkripsjonsregulering: Hvordan skrus genene våre på og av? *Tidsskr* N*or* L*ægeforen.* 1990; 110, nr. 26: 3366-3371.
3: **Njølstad PR**. Onkogeners molekylære virkningsmekanismer. *Tidsskr Nor Lægeforen.* 1990; 110: nr. 28, 3632-3636.
4: **Njølstad PR**. Onkogener. Gener som kan forårsake kreftutvikling! *Norsk farmaceutisk tidsskr.* 1991; 12: 11-14.
5: Molven A, **Njølstad PR**. Når beina sitter på hodet. *Naturen* 1991; 5: 203-211.
6: **Njølstad PR**, Vik ISS, Nordbø SA. Primær HIV-infeksjon. En differensialdiagnose til influensa-/mononukleoseliknende sykdom! *Tidsskr Nor Lægeforen.* 1992;112: 1446-1448.
7: **Njølstad PR**, Molven A. Homeoboksen - en genetisk nøkkel til fosterutvikling. *Tidsskr Nor* L*ægeforen.* 1993; 113, nr. 12: 1475-1479.
8: Søvik O, Knutzon J, Bjerknes R, **Njølstad PR**. Nyoppdaget diabetes mellitus type 1. *Ped Endokrin.* 1997; 11: 5-15.

Publications Pål Rasmus Njølstad 1987-2021

9: **Njølstad PR**, Søvik O. Ikke insulin-krevende diabetes hos barn og unge voksne (MODY). *Ped Endokrin.* 1997; 11: 27-34.
10: **Njølstad PR**, Søvik O, Cockburn B, Bell GI, Følling I, Sagen JV. Ikke-insulinkrevende diabetes mellitus hos barn og unge voksne. *Tidsskr Norske Lægeforen.* 1998; 118: 1054-1057.
11: Bjerknes R, Rosendahl K, Gjelland K, **Njølstad PR**, Aarskaog D. Ultralydundersøkelse av genitalia interna hos jenter med pubertas precox. *Ped Endokrin.* 1998; 12: 20-28.
12: **Njølstad PR**, Bjerknes R, Aarskaog D. Apert syndrom. *Ped Endokrin.* 1998; 12: 29-3.
13: Bjerknes R, Kittang OB, **Njølstad PR**, Bland JD, Knutzon J. (1998) Utredning ved mistanke om Cushing syndrom. *Ped Endokrin.* 1998; 12: 67-78.
14: Salvesen H, **Njølstad PR**. Blandt mordere og Nobel-prisvinnere i Chicago. *Tidsskr Nor Lægeforen.* 1999; 119: 1164-1166.
15: **Njølstad PR**. Til Chicago for å studere diabetesgener. *Diabetikeren* 1999; 3: 27-28.
16: **Njølstad PR**. USAs kamp mot diabetes: En strategisk plan for det 21. århundre. *Diabetikeren* 1999; 3: 29-31.
17: **Njølstad PR**, Søvik O. Nå kartlegges MODY. *Diabetes for helsepersonell* 1999; 3: 24-25.
18: **Njølstad PR**. Erstatning og oppreisning for fjernelse av friske eggstokker. *Tidsskr Nor Lægeforen.* 2000; 120: 511.
19: **Njølstad PR**. Forsinket diagnostisering av medfødt hofteleddsdysplasi. *Tidsskr Nor Lægeforen.* 2000; 120: 950.
20: **Njølstad PR**, Rosendahl K, Ersland L. Magnetisk resonans - forkortinger og ordforklaringer. *Tidsskr Nor Lægeforen* 2000; 120: 941.
21: Aarskog D, **Njølstad PR**, Bjerknes R. Klinisk dysmorfologi: En oversikt. *Ped Endokrin.* 2000; 14: 29-37.
22: **Njølstad PR**. Norsk Pasientskadeerstatning 1999. *Tidsskr Nor Lægeforen.* 2000; 120:2188.
23: **Njølstad PR**. Endometriecancer etter østrogenbehandling. *Tidsskr Nor Lægeforen.* 2000; 120: 2329.
24: **Njølstad PR**. Ibuprofen ved persisterende ductus arteriosus. *Tidsskr Nor Lægeforen..* 2000; 120: 2463.
25: **Njølstad PR.** Klage til Norsk Pasienterstatning basert på opplysninger hentet fra Internett. *Tidsskr Nor Lægeforen.* 2000; 120: 2815.
26: **Njølstad PR**. Forsinket diagnose av hereditær episodisk ataksi. *Tidsskr Nor Lægeforen.* 2000; 120: 3324.
27: Søvik O, **Njølstad PR**, Reigstad H, Brackman D, Teslo I, Brunvand L. Undersøkelse og behandling av barn med kongenitt hyperinsulinisme - til Paris for enhver pris? *Tidsskr Nor Lægeforen.* 2001; 121, 612-614.
28: **Njølstad PR**, Søvik O. Løser gåten om arvelig diabetes. *Innsyn* 2001; 2, 6-10.
29: **Njølstad PR**, Søvik O. Ny type diabetes: Medfødt diabetes kan nå knyttes til MODY. *Diabetes for helsepersonell* 2001; 3: 25-27.
30: **Njølstad PR**, Søvik O. Når betacellens glukosesensor svikter. *Diabetes* 2001; 5: 58-59.
31: **Njølstad PR**, Engebretsen LF, Molven A, Søvik O. MODY-prosjektet ved Haukeland Sykehus: fra forskning til diagnostikk *Diabetes for helsepersonell* 2001; 3: 23-24.
32: **Njølstad PR**. Bare barn? *Tidsskr Nor Lægeforen.* 2001; 121: 2575.
33: **Njølstad PR**. Bra om barnemat. *Tidsskr Nor Lægeforen.* 2002; 122: 648.
34: **Njølstad PR**. Aktuell ny nordisk lærebok i pediatri. *Tidsskr Nor Lægeforen.* 2002; 122: 647.
35: **Njølstad PR**. Internasjonal pris til Dagfinn Aarskog. *Tidsskr Nor Lægeforen.* 2002; 122: 443.
36: **Njølstad PR**. Klinisk molekylærmedisin (2): Diagnostikk av mindre mutasjoner. *Ped Endokrin.* 2002; 16: 23-26.
37: Bjerknes R, Skadberg BT, **Njølstad PR**. Inhalasjonssteroider og vekst. *Tidsskr Nor Lægeforen* 2001; 121, 281.
38: Aarskog D, **Njølstad PR**. Klinisk molekylærmedisin: en introduksjon. *Ped Endokrin.* 2001; 15: 59-60.
39: **Njølstad PR**, Aarskog D. Klinisk molekylærmedisin (1): Diagnostikk av større delesjoner og rearrangementer. *Ped Endokrin.* 2001; 15: 61-65.
40: Ræder H, Ræder M, **Njølstad PR**. Klinisk molekylærmedisin (3): DNA-sekvensering. *Ped Endokrin.* 2002; 16: 51-56.
41: **Njølstad PR**, Sagen JV. Klinisk molekylærmedisin (4): Koblingsanalyser. *Ped Endokrin.* 2003; 16: 34-38.
42: **Njølstad PR**, Molven A. Forskere diskuterte arv og diabetes. *Diabetes for helsepersonell* 2003; 4: 12-14.
43: **Njølstad PR**, Søvik O. Neonatal diabetes – permanent eller transient. *Paidos*, in press. 2003

44: **Njølstad PR**, Bjørkhaug L. Klinisk molekylærmedisin (5): Eksempler på funksjonelle analyser. *Ped Endokrin.* 2003; 16: 64-69.
45: **Njølstad PR**. Forskning ved Barneklinikken og Seksjon for pediatri, Universitetet i Bergen. *Paidos*, 3: 52-53.
46: **Njølstad PR**, Søvik O. Neonatal diabetes – permanent eller transient. *Paidos*, 2003; 3:53-55.
47: **Njølstad PR**. Etablering av mastergradstudium i Molekylærmedisin ved Medisinsk fakultet, Universitetet i Bergen. In: Nordmo I: Universitetspedagogisk utviklingsarbeid i Bergen. UPED-skrift nr 1, 2004. Program for læringsforskning, Universitetet i Bergen.
48: Bjørkhaug L, Johansson S, Røder H, Thorsby PM, Undlien DE, Søvik O, Molven A, Sagen JV, **Njølstad PR**: Molekylærgenetisk diagnostikk ved diabetes mellitus. *Tidsskr Nor Lægeforen* 2005; 125: 2968-2972.
49: Ræder H, **Njølstad PR**. Ny type diabetes. *Diabetesforum* 2006; 1: 21.
50: Laborie LB, Søvik O, **Njølstad PR**. Diagnose og behandling av monogene former for nyfødt- og spedbarnsdiabetes. *Ped Endokrin.* 2007; 21: 23-28.
51: Sandal T, Søvik O, **Njølstad PR**, Molven A. Utredning og behandling av medfødt hyperinsulinisme (1): Molekylære og genetiske aspekter. *Ped Endokrin.* 2008; 22: 37-43.
52: **Njølstad PR**, Johansson S. EASD: genetikk og diabetes type 2. *Diabetesforum* 2008; 4: 4-5.
53: Søvik O, Hansen EV, Juliusson P, **Njølstad PR**. Utredning og behandling av medfødt hyperinsulinisme (2): Diagnostiske og terapeutiske aspekter. *Ped Endokrin.* 2008; 22:56-63.
54: Hagenäs L, **Njølstad PR**. Neuro-cardio-facio-cutana syndrom och dysreglering av RAS-MAPK signaleringen. *Ped Endokrin.* 2009;23:15-29.
55: Søvik O, **Njølstad PR**, Molven A. To søstre med cerebrale, faciale, kutane og kardiale symptomer og mutasjoner i to forskjellige RAS-gener. *Ped Endokrin.* 2009;23:30-37.
56: Molven A, Søvik O, von der Lippe C, Steine SJ, **Njølstad PR**, Houge G, Prescott TE. Ny molekylærgenetisk diagnostikk: Medfødte syndromer som skyldes genfeil i RAS/MAPK-signalveien. *Tidsskr Nor Lægeforen* 2009;129:2358-61.
57: **Njølstad PR**, Molven A, Søvik O. Internasjonalt forskermøte i Bergen: Genetikk er viktig for diagnose og behandling av diabetes! *Diabetesforum* 2009;4: 30-31.
58: **Njølstad PR**, Hertel J, Søvik O, Ræder H, Johansson S, Molven A. Fremskritt innen diabetesgenetikk. *Tidsskr Nor Lægeforen* 2010;130:1145-9.
59: **Njølstad PR**, Molven A. Genetikk i fokus. *Diabetesforum* 2010;2:6-9.
60: Molven A, **Njølstad PR**. Monogen diabetes i Norge. *Diabetesforum* 2010;2:10-15.
61: Haldorsen IS, **Njølstad PR**. Billeddiagnostikk av pankreas ved monogen diabetes. *Diabetesforum* 2011;4: 20-2.
62: Raeder H, Molven A, **Njølstad PR**. Skreddersydd medisin eller narsissomikk? *Tidsskr Den norske legeforen* 2012;132:1844-5.
63: **Njølstad PR.** De tunge barna (kronikk). *Bergens Tidende*, juli 2013.
64: Hertel JK, Johansson S, Midthjell K, Nygård O, **Njølstad PR**, Molven A. Type 2 diabetes genes: present status and data from Norwegian studies. *Norsk Epidemiology* 2013;23:9-22.
65: **Njølstad PR**. Hvorfor får man monogen diabetes? I: Vaaler S, Møinichen T (Ed): *Diabeteshåndboken*, pp. 45-52, Gyldendal Akademisk, 2014.
66: **Njølstad PR**. Behandling av monogen diabetes I: Vaaler S, Møinichen T (Ed): *Diabeteshåndboken*, pp. 99-102, Gyldendal Akademisk, 2014.
67: Søvik O, Sagen J, Haldorsen IS, Tjora E, **Njølstad PR**. HNF1B-MODY: A disorder of development with diabetes and congenital malformations in pancreas and urogenital tract due to mutations in *HNF1B*. In: Inborn Errors of Development (Epstein, Erickson and Wynshaw-Boris, eds), pp. 921-926. Oxford University Press, New York, 2016. ISBN 978-0-195-30691-0.
68: Njølstad TS, Ellard S, **Njølstad PR**. Insights from monogenic diabetes. In: The Genetics of Type 2 Diabetes and Related Traits - Biology, Physiology and Translation (Florez, ed.), pp. 223-240. Springer, 2016. ISBN 978-3-319-01573-8.
69: **Njølstad PR.** Ny som instituttleder: Wirkola, Boklöv og 1+1=3. Leder, *K2 Nytt*, uke 50, 2017.

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70: **Njølstad PR.** Godt nyttår. Leder, *K2 Nytt*, uke 1, 2018.
71: **Njølstad PR.** Årets forskningspresentasjoner. Leder, *K2 Nytt*, uke 3, 2018.
72: **Njølstad PR.** Økonomisk handlingsrom i 2018. Leder, *K2 Nytt*, uke 5, 2018.
73: **Njølstad PR.** Små driftsmidler. Leder, *K2 Nytt*, uke 7, 2018.
74: **Njølstad PR.** Bergen forskningsstiftelse. Leder, *K2 Nytt*, uke 8, 2018.
75: **Njølstad PR.** Xtramidler. Leder, *K2 Nytt*, uke 9, 2018.
76: **Njølstad PR.** Tid for å hedre og ære våre beste innen forskning, undervisning og formidling 2017. Leder, *K2 Nytt*, uke 11, 2018.
77: **Njølstad PR.** Faglige møter: Jakt på kunnskap og vennskap. Leder, *K2 Nytt*, uke 15, 2018.
78: **Njølstad PR.** Bli bedre kjent med K2! Leder, *K2 Nytt*, uke 17, 2018.
79: **Njølstad PR.** Faculty lunch 9. mai: Besøk av Victor Norman med viktige saker vedrørende undervisning og forskning. Leder, *K2 Nytt*, uke 19, 2018.
80: **Njølstad PR.** God sommer! Leder, *K2 Nytt*, uke 24, 2018.
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84: **Njølstad PR.** Undervisningsdagen 2019. Leder, *K2 Nytt*, uke 19, 2019.
85: **Njølstad PR.** Forskningsgruppene ved Klinisk institutt 2. Leder, *K2 Nytt*, uke 23, 2019.
86: **Njølstad PR.** God sommer. Leder, *K2 Nytt*, uke 25, 2019.
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88: **Njølstad PR.** Medarbeiderssamtaler. Leder, *K2 Nytt*, uke 36, 2019.
89: **Njølstad PR.** Tid for registrering av sidegjøremål. Leder, *K2 Nytt*, uke 38, 2019.
90: **Njølstad PR.** Om RETTE. Leder, *K2 Nytt*, uke 40, 2019.
91: **Njølstad PR.** Nobelprisen i medisin eller fysiologi 2019. Leder, *K2 Nytt*, uke 42, 2019.
92: **Njølstad PR.** Verdens diabetesdag 14. november. Leder, *K2 Nytt*, uke 46, 2019.
93: **Njølstad PR.** Genmodifiserte organismer (GMO). Leder, *K2 Nytt*, uke 47, 2019.
94: **Njølstad PR.** Strammere økonomi i 2020. Leder, *K2 Nytt*, uke 48, 2019.
95: **Njølstad PR.** God jul! Leder, *K2 Nytt*, uke 50, 2019.
96: **Njølstad PR.** Godt nyttår. Leder, *K2 Nytt*, uke 1, 2020.
97: **Njølstad PR.** Forskerskolen i vinden. Leder, *K2 Nytt*, uke 4, 2020.
98: **Njølstad PR.** Konsul Søren Falch og øyenlege Sigurd Falchs fonds. Leder, *K2 Nytt*, uke 6, 2020.
99: **Njølstad PR.** Hvad nuh, når søknaden din blir avvist? Leder, *K2 Nytt*, uke 8, 2020.
100: **Njølstad PR.** Koronavirusepidemien. Leder, *K2 Nytt*, uke 10, 2020.
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102: **Njølstad PR.** Om å ta vare på hverandre og forhåndsregler ved utlån av personell. Leder, *K2 Nytt*, uke 12, 2020.
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112: **Njølstad PR.** Midlertidig rokkering i K2-ledelsen. Leder, *K2 Nytt*, uke 38, 2020.
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114: **Njølstad PR.** Godt nyttår! Leder, *K2 Nytt*, uke 1, 2021.

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116: **Njølstad PR.** Tid for ARK igjen. Leder, *K2 Nytt*, uke 7, 2021.
117: **Njølstad PR.** Valg av rektor og prorektor og nye medlemmer til universitetsstyret. Leder, *K2 Nytt*, uke 9, 2021.
119: **Njølstad PR.** Midler til utdanning og stipendiatstillinge. Leder, *K2 Nytt*, uke 11, 2021.
120: **Njølstad PR.** Universitetet i Bergen er 75 år! Leder, *K2 Nytt*, uke 15, 2021.