Program EASD-SGGD meeting Leiden 2017

Genetics of diabetes and its role in precision medicine
Version: March 2017

Topics:
Type 1 diabetes, Type 2 diabetes, Pharmacogenetics
Precision medicine/therapeutics, Metagenomics
Monogenic diabetes, Clinical utility of genetics
Functional studies, Diabetic complications, obesity
Epigenetics, Animal models, Gene environment interactions
<table>
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<tr>
<th>Time</th>
<th>Event</th>
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<tr>
<td><strong>Thursday May 11</strong> (Amsterdam/Rotterdam room)</td>
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<tr>
<td>12.00 – 13.30</td>
<td>Lunch in the poster area</td>
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<tr>
<td>13.30 - 13.45</td>
<td>Welcome and opening</td>
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<tr>
<td><strong>13.45 – 14.45</strong></td>
<td>Special session 1 Opening session</td>
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<tr>
<td>Chair:</td>
<td>Leen ’t Hart, NL; Mike Weedon, UK</td>
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<tr>
<td>13.45 – 14.15</td>
<td>L Groop, Lund University, Sweden</td>
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<tr>
<td>Title: Genetics of common diabetes- but does common diabetes exist?</td>
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<tr>
<td>14.15 – 14.45</td>
<td>P Njølstad, University of Bergen, Norway</td>
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<tr>
<td>Title: Genetics of monogenic genes - present clinical challenges</td>
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<tr>
<td><strong>14.45 - 16.15</strong></td>
<td>Poster session 1 (groups A to E)</td>
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<tr>
<td>Epigenetics, Screening for monogenic diabetes, Monogenic obesity,</td>
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<td>Type 2 diabetes genetics, Functional genomics</td>
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<td>Coffee/Tea Break</td>
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<tr>
<td>16.15 – 18.05</td>
<td>Session 1 isolated populations</td>
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<tr>
<td>Chair:</td>
<td>Tim Frayling, UK; Ko Willems-van Dijk, NL</td>
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<tr>
<td>16.15 – 16.45</td>
<td>T Hansen, University of Copenhagen, Denmark</td>
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<tr>
<td>Title: Genetics of metabolic traits: Lessons from isolated populations</td>
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<tr>
<td><strong>16.45 – 18.05</strong></td>
<td>Best meeting abstracts session</td>
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<tr>
<td>16.45 – 17.05</td>
<td>OP1 JC Florez, Boston, USA</td>
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<tr>
<td>TCF7L2 genetic variation augments incretin resistance and influences response to a sulfonylurea and metformin: the Study to Understand the Genetics of the Acute Response to Metformin and Glipizide in Humans (SUGAR-MGH)</td>
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<tr>
<td>17.05 – 17.25</td>
<td>OP2 EA Ahlqvist, Malmö, Sweden</td>
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<td>GWAS of glucose-stimulated hormone secretion identifies novel determinants of circulating GIP and GLP-1 concentrations.</td>
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<tr>
<td>17.25 – 17.45</td>
<td>OP3 PB Bowman, Exeter, UK</td>
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<tr>
<td>Sulphonylureas are a highly effective and safe long-term treatment for neonatal diabetes due to KCNJ11 mutations: the first 10-year follow-up study of a large international cohort</td>
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<tr>
<td>17.45 – 18.05</td>
<td>OP4 N Groen, Leiden, Netherlands</td>
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<tr>
<td>Exploring human pancreatic β-cell plasticity using single-cell RNA sequencing</td>
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<tr>
<td>19.00</td>
<td>Dinner in the Garden Restaurant</td>
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## Friday May 12 (Amsterdam/Rotterdam room)

<table>
<thead>
<tr>
<th>Time</th>
<th>Session/Poster Session</th>
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| 8.45 – 10.30 | **Special Session 2**  
Chair: Jose Florez, USA; Vincenzo Trischitta, Italy                                                             |
| 8.45 – 9.15 | **ER Pearson, University of Dundee, UK**  
Title: Precision medicine approaches in Type 2 diabetes                                                             |
| 9.15 – 9.45 | **AT Hattersley, University of Exeter, UK**  
Title: Novel use of genetic information in the diabetes clinic                                                          |
| 9.45 – 10.15 | **G Nijpels, VU University medical Center, NL**  
Title: A clinician’s view on precision medicine                                                                       |
| 10.15 – 10.30 | **K Dolsma, Erfocentrum, NL**  
Title: Action Programme Early detection of MODY in the Netherlands                                                      |
| 10.30 -12.00 | **Poster session 2 (Groups F to J)**  
Type 2 diabetes, obesity and birth weight; Pharmacogenetics and treatment; Functional studies in MODY; MODY diabetes; Diabetes phenotypes and complications  
Coffee/Tea Break                                                                                                                                 |
| 12.00 – 13.00 | **Session 2**  
Chair: Katharine Owen, UK; Antonio Cuesta- Muñoz, DK                                                              |
| 12.00 – 12.15 | **OP5 KA Patel, Exeter, UK**  
Heterozygous RFX6 protein truncating variants cause Maturity-Onset Diabetes of the Young (MODY) with reduced penetrance |
| 12.15 – 12.30 | **OP6 H Krogh Pedersen, Kongens Lyngby, Denmark**  
Pancreatic Islet Protein Complexes and Their Dysregulation in Type 2 Diabetes                                              |
| 12.30 – 12.45 | **OP7 A Juszczak, Oxford, UK**  
High-sensitivity CRP and antennary fucosylated glycans as biomarkers of HNF1A-MODY                                    |
| 12.45 – 13.00 | **OP8 MB Johnson, Exeter, UK**  
A type 1 diabetes genetic risk score discriminates monogenic autoimmune diabetes from polygenic clustering of diabetes and autoimmunity |
| 13.00 - 14.30 | Lunch                                                                                                              |

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This schedule outlines the events and presentations for the evening of Friday, May 12, in Amsterdam/Rotterdam room. It includes sessions, poster sessions, and presentations on various topics related to diabetes research and treatment.
<table>
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<tr>
<th>Time</th>
<th>Session 3</th>
<th>Epigenetics / microbiome in diabetes</th>
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<tbody>
<tr>
<td>14.30 – 15.00</td>
<td>C Ling, Lund University, Sweden</td>
<td>Chair: Roderick Slieker, NL; Janis Klovins, Latvia</td>
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<tr>
<td>15.00 – 15.30</td>
<td>O Pedersen, University of Copenhagen, Denmark</td>
<td>Title: Epigenetic variation and the pathogenesis of diabetes</td>
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<td>Title: Microbial intestinal dysbiosis in type 2 diabetes – a consequence or a co-pathogenic element?</td>
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<td>Selected oral presentations based on abstracts</td>
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<tr>
<td>15.30 – 15.45</td>
<td>OP9 JV van Vliet-Ostaptchouk, Groningen, Netherlands</td>
<td>Exposure to endocrine disruptors in the general Dutch population and its effect on genome-wide DNA methylation</td>
</tr>
<tr>
<td>15.45 – 16.00</td>
<td>OP10 A Perfilyev, Malmö, Sweden</td>
<td>Impact of fat overfeeding on the DNA methylation in human adipose tissue</td>
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<tr>
<th>Time</th>
<th>Session 4</th>
<th>Functional and translational studies in diabetes</th>
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<tr>
<td>16.00 - 16.30</td>
<td>Coffee/Tea Break in poster area</td>
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<tr>
<td>16.30 – 17.00</td>
<td>A Gloyn, Oxford University, UK</td>
<td>Title: Defining mechanisms for pancreatic beta-cell dysfunction in type 2 diabetes</td>
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<tr>
<td>17.00 – 17.30</td>
<td>J Ferrer, Imperial College London, UK and Univ. of Barcelona (IDIBAPS), Spain</td>
<td>Title: Noncoding genome function in pancreatic islets and diabetes</td>
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<tr>
<td></td>
<td>Selected oral presentations based on abstracts</td>
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<tr>
<td>17.30 – 17.45</td>
<td>OP11 VG Gudmundsdottir, Kongens Lyngby, Denmark</td>
<td>Integrative network analysis highlights biological processes underlying GLP-1 stimulated insulin secretion: a DIRECT study</td>
</tr>
<tr>
<td>17.45 – 18.00</td>
<td>OP12 M den Hoed, Uppsala, Sweden</td>
<td>Zebrafish larvae as a model system for high-throughput, image-based screens in diabetes and insulin resistance</td>
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19.00    Dinner in the Garden Restaurant
### Saturday May 13 (Amsterdam/Rotterdam room)

<table>
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<tr>
<th>Time</th>
<th>Session</th>
<th>Title</th>
<th>Details</th>
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<tbody>
<tr>
<td>8.45 – 10.15</td>
<td><strong>Special session 3</strong></td>
<td><em>Obesity / gene x environment interactions/complications</em></td>
<td><em>Chair: Ingrid Dahlman, Sweden; Inês Barroso, UK</em></td>
</tr>
<tr>
<td>8.45 – 9.15</td>
<td><strong>PW Franks</strong>, <em>Lund University and Umeå University, Sweden</em></td>
<td>Title: Will genetics help improve the precision of lifestyle medicine?</td>
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<tr>
<td>9.15 – 9.45</td>
<td><strong>P Froguel</strong>, <em>Imperial College London, UK and Lille University, France</em></td>
<td>Title: New insight in (epi)genetics of obesity and metabolic complications</td>
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<tr>
<td>9.45 – 10.15</td>
<td><strong>J Skupień</strong>, <em>Jagiellonian University, Poland</em></td>
<td>Title: Genome-wide association studies of complex phenotypes of diabetes and its complications – issues, methods and advantages</td>
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<tr>
<td>10.15 – 10.45</td>
<td><strong>Coffee/Tea Break in poster area</strong></td>
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<tr>
<td>10.45 – 12.00</td>
<td><strong>session 5</strong></td>
<td><em>Open session based on abstracts</em></td>
<td><em>Chair: Amelie Bonnefond, France; Ben Glaser, Israel</em></td>
</tr>
<tr>
<td>10.45 – 11.00</td>
<td><strong>OP13 SE Jones</strong>, Exeter, UK</td>
<td>The genetics of objective activity-monitor derived estimates of sleep patterns and the link to obesity and type 2 diabetes</td>
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<tr>
<td>11.00 – 11.15</td>
<td><strong>OP14 A Mahajan</strong>, Oxford, UK</td>
<td>Expanding the spectrum of type 2 diabetes risk alleles through genome-wide association study imputed up to Haplotype Reference Consortium panel</td>
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<tr>
<td>11.15 – 11.30</td>
<td><strong>OP15 RC Sliker</strong>, Leiden, The Netherlands</td>
<td>Poor glycaemic control is associated with altered blood gene expression levels of cell cycle and immune related genes</td>
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<tr>
<td>11.30 – 11.45</td>
<td><strong>OP16 E Valo</strong>, Helsinki, Finland</td>
<td>Genetic background of diabetic kidney disease</td>
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<tr>
<td>11.45 – 12.00</td>
<td><strong>OP17 Y Yi</strong>, Exeter, UK</td>
<td>Identification of alleles associated with higher body fat percentage but lower risk of type 2 diabetes</td>
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<tr>
<td>12.00 - 13.30</td>
<td><strong>Lunch</strong></td>
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13.30 – 13.45  **Best poster and oral presentation Award ceremony**
*Chair: Leen M ’t Hart*

**Best poster presentation award.**
*Representative of Illumina*

**Best oral presentation award.**
*Representative of SANOFi*
*mandatory for winners to be present*

<table>
<thead>
<tr>
<th>Time</th>
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| 13.45 – 14.45 | **Special session 4**  **Closing session**  
*Chair: Hanieh Yaghoookar, UK ; Martine Vaxillaire, France* |
| 13.45 – 14.15 | **S Farooqi, University of Cambridge, UK**  
Title: Genetics of Severe Obesity-what have we learned ? |
| 14.15 – 14.45 | **MI McCarthy, University of Oxford, UK**  
Title: Genetics of Diabetes, the past, the present and the future |

14.45 – 15.00  **Farewell and meeting end**
Session A, Epigenetics

Chair: Charlotte Ling, Denmark

PS1 Subcutaneous pre-adipocyte Methylome and T2D susceptibility: new insights

PS2 Changes in DNA methylation in peripheral blood cells from humans threatened with antidiabetic drug metformin
Ilze Elbere, Raitis Peculis, Ineta Kalnina, Linda Zaharenko, Davids Fridmanis, Valdis Pirags, Janis Klovins

PS3 DNA methylation associated with T2D and glycemic traits - a replication study in the LifeLines Cohort
Eliza Walaszczyk, Mirjam Luijten, Annemieke Spijkerman, Marc Jan Bonder, Helen L Lutgers, Harold Snieder, Bruce HR Wolffenbuttel, Jana V van Vliet-Ostaptchouk

PS4 Association between maternal smoking-induced changes to Dna methylation in GFI1 and offspring's cardio-metabolic health
Priyanka Parmar, E Lowry, V Karhunen, P Froguel, T Andrew, L Ala-Mursula, J Miettunen, S Keinänen-Kiukaanniemi, MR Järvelin, S Sebert

PS5 DNA methylation of metformin transporter genes in human liver is associated with diabetes medication
Sonia García-Calzón, Alexander Perfilyev, Ville Männistö, Vanessa D. de Mello, Emma Nilsson, Jussi Pihlajamäki and Charlotte Ling

PS6 Abnormal epigenetic changes during differentiation of human skeletal muscle stem cells from obese subjects
Cajsa Davegårdh, Christa Broholm, Alexander Perfilyev, Tora Henriksen, Sonia Garcia-Calzon, Lone Peijs, Ninna Schiøler Hansen, Petr Volkov, Rasmus Kjøbsted, Jørgen FP Wojtaszewski, Maria Pedersen, Bente Klarlund Pedersen, Dov B. Ballak, Charles A. Dinarello, Bas Heinhuis, Leo A.B. Joosten, Emma Nilsson, Allan Vaag, Camilla Scheele and Charlotte Ling

PS7 IRS1 DNA promoter methylation in human adipose tissue mediates the relationship of rs2943650 near IRS1 to fat distribution and metabolic traits
Kerstin Rohde, Maria Keller, Xuanshi Liu, Matthias Klös, Michael Stumvoll, Arne Dietrich, Michael R. Schön, Daniel Gärtner, Tobias Lohmann, Miriam Dreßler, Peter Kovacs, Matthias Blüher, Yvonne Böttcher
POSTER SESSION 1: THURSDAY MAY 11TH 14:15-16:45

Session B, Screening for monogenic diabetes

Chair: Pål Njølstad, Norway

PS8 Search for variants influencing age of diagnosis in HNF1A-MODY diabetes
Agnieszka H. Ludwig-Galezowska, Jan Skupien, Tomasz Klupa, Maciej T. Malecki, Andrew Hattersley, Stepanka Pruho, Alessandro Doria, Christine Bellanne-Chantelot, Daniela Gasperikova

PS9 Systematic screening for MODY mutations in people of south Asian and African-Caribbean ethnicity: results from the MY DIABETES study
Shivani Misra, Kevin Colclough, Des Johnston, Sian Ellard, Andrew Hattersley & Nick Oliver

PS10 Analysis of large scale sequencing cohorts does not support the role of variants in UCP2 as a cause of hyperinsulinaemic hypoglycaemia
Thomas W Laver, Michael N Weedon, Richard Caswell, Khalid Hussain, Sian Ellard and Sarah E Flanagan

PS11 Monogenic Diabetes accounts for 6.3% of cases referred to 15 Italian pediatric diabetes Centers during 2007-2012.

PS12 MODY diagnostics, the Leiden experience: gene panel based mutation analysis by next generation sequencing
Peter E. Thijssen, Ronelle Snowdowe, Naima Lamzira, Maaike Verschuren, Sander Bollen, Ivo F.A.C. Fokkema, Eelco J.P. de Koning, Nienke van der Stoep, and Monique Losekoot

PS13 Reassessing MODY genes using large scale sequencing cohorts does not support the pathogenicity of BLK, PAX4 and PDX1
Olivia Knox, Matthew Wakeling, Kashyap Patel, Richard C Caswell, Elisa De Franco, Sarah E Flanagan, Sian Ellard, Andrew T Hattersley, Michael N Weedon, Thomas W Laver

PS14 Optimization of a next generation sequencing innovative protocol for the accurate detection of punctual mutations and copy number variant in children with developmental delay
Session C, Monogenic obesity

Chair: Phillipe Froguel, France

PS15 Diet and genetic risk of obesity in the UK Biobank study.

PS16 A homozygous mutation in ROCK1 gene is associated with severe obesity in a consanguineous Pakistani population
Sadia Saeed, Amelie Bonnefond, Qasim M Janjua, Sadia M Din, Mohsin A Chattha, Young-Bum Kim, Muhammad Arslan and Philippe Froguel

PS17 Higher body mass index but lower type 2 diabetes prevalence in New Zealand Polynesian populations carrying CREBF-rs373863828 variant
Rinki Murphy, Mohanraj Krishnan, Tanya Flynn, Nicola Dalbeth, Lisa Stamp, Ofa Dewes, Andrew Shelling, Peter Shepherd, Tony Merriman

PS18 Evidence of decreased osteogenic activity in congenitally leptin deficient but not in MC4R deficient children
Qasim M Janjua, Sadia Saeed, Jaida Manzoor, Sadia M Din, Amelie Bonnefond, Philippe Froguel and Muhammad Arslan

PS19 Evaluation of bioactive and total immunoreactive serum leptin levels in obese children and adolescents
Juraj Stanik, Jürgen Kratzsch, Kathrin Landgraf, Kathrin Scheuermann, Ulrike Spielau, Ruth Gausche, Daniela Gasperikova, Wieland Kiess, Antje Körner

PS20 Evidence that physical activity and genetic risk of obesity interact to influence obesity.

PS21 Functional and clinical relevance of the novel and known PCSK1 variants for childhood obesity and glucose metabolism
Dennis Löffler, Susanne Behrendt, John W. M. Creemers, Jürgen Klammt, Gabriela Aust, Juraj Stanik, Wieland Kiess, Peter Kovacs, Antje Körner
Session D, Type 2 diabetes genetics

Chair: Torben Hansen, Denmark

PS22 Discovery and fine-mapping of type 2 diabetes susceptibility loci across diverse populations
Hidetoshi Kitajima, Anubha Mahajan, Xueling Sim, Maggie Ng, Weihua Zhang, Jennifer E Below, Daniel Taliun, Kyle J Gaulton and Andrew P Morris, on behalf of the DIAMANTE Consortium.

PS23 Identification of chromosomal aneuploidies, large copy number variants, and unbalanced translocations through analysis of off-target reads from targeted next generation sequencing
Matthew Wakeling, Elisa de Franco, Andrew T Hattersley, Sian Ellard

PS24 Novel locus discovery through trans-ethnic association analyses of glycemic traits using densely imputed genetic data
Gaelle Marenne, on behalf of the Meta-Analyses of Glucose and Insulin-related traits Consortium (MAGIC) Investigators

PS25 Insights on the use of super-controls from the UK Biobank type 2 diabetes case-control GWAS.
Benard W. Kulohoma, Anubha Mahajan, Mark I McCarthy

PS26 African T2D Genome-wide Association Study
Ji Chen, Meng Sun, Adebowale A. Adeyemo, Tommy Carstensen, Andrew Morris, Fraser Pirie, Ayesha Motala, Manj Sandhu, Charles N. Rotimi, Mark McCarthy, Inês Barroso, Eleanor Wheeler, Anubha Mahajan

PS27 Fine mapping of T2D linked 12q24 region in Finnish families
Dwivedi OP, Tuomi T, Groop L

PS28 Retracted
POSTER SESSION 1: THURSDAY MAY 11TH 14:15-16:45

Session E, Functional genomics

Chair: Jorge Ferrer, UK, Spain

P29 Combining GWAS and expression analysis to identify eQTLs in pancreatic samples. Amna Khamis, Mickaël Canouil, Afshan Siddiq, Hutokshi Crouch, Mario Falchi, Anke M Schulte, Manon von Buelow, Lorella Marselli, Florian Ehehalt, Krister Bokvist, Ioannis Xenarios, Mark Ibberson, Michele Solimena, Piero Marchetti and Philippe Froguel.


PS31 Variants in Melatonin Receptor 1B gene which are Associated with Type 2 Diabetes Affect Specific Subsets of the Receptor Signaling Modalities. Amélie Bonnefond, Angeliki Karamitri, Bianca Plouffe, Min Chen, Alan Hegron, Mathilde Boissel, Jean-Luc Guillaume, Michel Bouvier, Philippe Froguel, Ralf Jockers.


PS33 Type 2 Diabetes genes with cross-traits relevance: identifying genetic links of common diabetes with its comorbidities. Karla V. Allebrandt, Hartmut Ruetten, Daniel Crowther and Francesca Frau.

PS34 Inuit in Greenland have significantly higher heritability for anthropometric traits. Emil V. Appel, Ida Moltke, Marit E. Jørgensen, Peter Bjerregaard, Allan Linneberg, Torben Hansen, Niels Grarup, Anders Albrechtsen.

Session F, Type 2 diabetes, obesity and birth weight
Chair: Paul Franks, Sweden

**PS36 ASSOCIATION OF EIF2S3 MUTATIONS WITH MEHMO SYNDROME**

**PS37 Maternal genome-wide association study highlights intrauterine influences on birth weight**

**PS38 Rare pathogenic CNVs are associated with BMI and type 2 diabetes risk variation in a very large population based study**

**PS39 Gene-environment interaction on the risk of obesity among ethnic minority populations living in EU and North America: A systematic review**
Zafarmand, M.H.; Tuvay, B.; Spijker, R.; Tajik, P; Agyemang, C.

**PS40 Investigating the association of two lipodystrophy genetic risk scores with anthropometry, insulin sensitivity and MRI derived fat distribution people with pre-diabetes or diabetes: an IMI-DIRECT Study.**
Francesca Frau, Andrea Mari, E Louise Thomas, Wareed Alenaini, Timothy M Frayling, Hanieh Yaghootkar, Hartmut Ruetten, Leen ‘t Hart, Paul W Franks, Karla V Allebrandt, Jimmy D Bell, Ewan Pearson for the DIRECT Consortium

**PS41 Interrelationship of the rs7903149 TCF7L2 gene variant with measures of glucose metabolism and adiposity: the NEO Study**
Raymond Noordam, Charlotte PA Zwetsloot, Renée de Mutsert, Dennis O Mook-Kanamori, Hildo J Lamb, Albert de Roos, Eelco JP de Koning, Frits R Rosendaal, Ko Willems van Dijk, Diana van Heemst

**PS42 Genome-wide study identifies potential parent-of-origin effect on birth weight at ANK1-NKX6-3 type 2 diabetes locus**
POSTER SESSION 2: FRIDAY MAY 12TH 10:30-12:00

Session G, Pharmacogenetics and treatment

Chair: Ewan Pearson, UK

PS43 Variation in the Plasma membrane monoamine transporter gene PMAT (SLC29A4) is associated with intolerance to metformin in type 2 diabetes: An IMI-DIRECT study

PS44 Metformin, transport & action: polymorphisms and their efficacy on daily dose of insulin, HbA1cglycaemic control and metformin plasma levels
A post hoc analysis of a 4.3 year randomized placebo controlled trial
Mattijs Out, Matthijs L. Becker, Ron H. van Schaik, Philippe Lehert, Coen D. Stehouwer, Adriaan Kooy

PS45 CYP2C9 and POR gene-gene interaction affects risk of sulfonylurea-induced hypoglycaemia

PS46 Treatment and outcomes of HNF1A-MODY patients from a specialist monogenic diabetes clinic
Agata Juszczak, Paula Bauchan, Amanda Webster, Fainia Kavvoura, Gaya Thanabalasingham, Katharine R. Owen

PS47 Functional characterization of HNF4A variants identified in Norwegian diabetes registries can be important for precision medicine in diabetes clinics
Alba Kaci, Janne Molnes, Bente B. Johansson, Pål R. Njølstad, Lise Bjørkhaug, Ingvild Aukrust

PS48 Early successful hematopoietic cell transplantation (HSCT) in a boy with IPEX syndrome caused by novel c.721T>C FOXP3 mutation
Stepanka Pruhoova, Barbora Obermannova, Renata Formankova, Zdenek Sumnik, Lenka Elblova, Jana Kayserova, Petr Sedlacek, Jan Lebl

PS49 The effect of type 2 diabetes-associated SNPs at the HNF1A locus on age at diabetes diagnosis in HNF1A-MODY carriers.
Jonathan M Locke, Sian E Ellard, Lorna W Harries, Andrew T Hattersley and Michael N Weedon
Session H, Functional studies in MODY

Chair: Anna Gloyn, UK

PS50 In vivo measurement and biological characterization of the diabetes-associated mutant insulin p.R46Q (ArgB22Gln)
Julie Støy, Jørgen Olsen, Soo-Young Park, Søren Gregersen, Claudia U. Hjørringgaard, Graeme I. Bell

PS51 Functional analysis of various HNF4A variants identifies increased transactivation function of R85W-HNF4A causing the mutation specific phenotype of neonatal hyperinsulinism and Fanconi syndrome
Lise Bjørkhaug, Lorentze Hope Hornnes, Alba Kaci, Andre Madsen, Christine Bellané-Chantelot, Andrew Hattersley, Gunnar Mellgren, Ingvild Aukrust, Pål R. Njølstad

PS52 Functional characterisation of novel activating STAT3 mutation observed in two patients with short stature and diabetes and its role in growth hormone signalling

PS53 Old genes, new diseases: Dominant ER stress-inducing WFS1 mutations underlie a genetic syndrome of neonatal/infancy onset diabetes, congenital sensorineural deafness and congenital cataracts.
Elisa De Franco Sarah E. Flanagan, Takuya Yagi, Damien Abreu, Jana Mahadevan, Matthew B Johnson, Garan Jones, Richard Caswell, Sian Ellard, Fumihiko Urano, Andrew T. Hattersley

PS54 Identification and functional characterization of 8 inactivating glucokinase mutations causing GCK-MODY.
Rosa Martínez, Ángel Gutierrez-Nogués, Teresa Velayos, Inés Urrutia, Anibael Aguayo, Idoia Martínez de LaPiscina, Spanish MODY group, María-Angeles Navas, Castaño L.

PS55 Anhydroglucitol and C-reactive protein – alone or together in MODY diagnosis?
M. Szopa, M. Kapusta, B. Matejko, J. Skupien, J. Hohendorff, B. Zapała, T. Platek, W. Glodzik, MT Malecki, T. Klupa
Session I, MODY diabetes

Chair: Andrew Hattersley, UK

PS57 Contribution of Mutations in MODY and Neonatal Diabetes Genes on Multigenerational Diabetes Mellitus of Adult Patients.
Serena Pezzilli, Ornella Ludovico, Tommaso Biagini, Luana Mercuri, Federica Alberico, Eleonora Lauricella, Hamza Dallali, Giorgio Basile, Elide Miccinilli, Pamela Piscitelli, Maria Giovanna Scarale, Salvatore De Cosmo, Massimo Carella, Tommaso Mazza, Vincenzo Trischitta, Sabrina Prudente

PS58 Decreased ventricular contractility in Hif1a deficient offspring of diabetic mothers.
Radka Cerychova, Frantisek Papousek, Frantisek Kolar, Pavel Abaffy, Vladimir Benes, Romana Bohuslavova, Gabriela Pavlinkova

PS59 Impact of diabetes caused by digenic inheritance of Glucokinase Gene and Hepatocyte Nuclear Factor-1 Alpha mutations on glucose metabolism and incretin secretion.

PS60 Biliary Anomalies in Patients with HNF1B-Diabetes.
Jarno LT Kettunen, Helka Parviainen, Päivi J Miettinen, Martti Färkkilä, Marjo Tamminen, Pia Salonen, Eila Lantto, Tiinamaija Tuomi

PS61 A novel WFS1 variant with ambiguous inheritance pattern.
Martina Skopkova, Juraj Stanik, Lukas Varga, Ivica Masindova, Emilia Jancova, Iwar Klimes, Milan Profant, Daniela Gasperikova

PS62 A novel HNF4A mutation c.427-1G>A causing congenital hyperinsulinism and glycogenosis -like phenotype
Juraj Stanik, Martina Skopkova, Katarina Brennerova, Daniel Danis, Monika Rosolankova, Anna Salingova, Vladimir Bzduch, Iwar Klimes, and Daniela Gasperikova

PS63 Novel p.Tyr60Asn mutation in CTLA4 causing familial autoimmune diabetes combined with autoimmune cytopenias
**POSTER SESSION 2: FRIDAY MAY 12TH 10:30-12:00**

Session J, Diabetes phenotypes and complications

Chair: Leif Groop, Sweden

**PS64 Retracted**

**PS65 A genome-wide association study of IVGTT-based measures of first phase insulin secretion refines the underlying physiology of type 2 diabetes variant**

Andrew R Wood, Anna Jonsson, Anne Jackson, Nan Wang, Nienke van Leewen, Nicholette D Palmer, Donald W Bowden, Torben Hansen, Mark Walker, Richard M Watanabe, Leen M t’ Hart, Robert L Hanson, Timothy M Frayling

**PS66 POST-LOAD GLUCOSE CURVES AND THEIR ASSOCIATION WITH GENETIC PROFILES AND CARDIOMETABOLIC RISK FACTORS OVER TIME.**


**PS67 Identification of four type 2 diabetes associated genes modulating pancreatic function and insulin secretion**


**PS68 Homozygous carriers of the TCF7L2 rs7903146 T-allele show altered postprandial response in triglycerides and triglyceride-rich lipoproteins**


**PS69 Negative Pressure Wound Therapy of Diabetic Foot Ulcer results in different Gene Expression Profile than Standard Approach**


**PS70 Genome-wide association study for diabetic nephropathy in American Indians with type 2 diabetes**

Ivica Masindova, Paolo Piaggi, Yunhua L. Muller, Sayuko Kobes, Peng Chen, Stephanie K. Tanamas, Jennifer E. Weil, Guozhi Jiang, Emma Ahlqvist, William C. Knowler, Ronald C. Ma, Juliana C. Chan, Leif Groop, Robert G. Nelson, Clifton Bogardus, Leslie J. Baier, Robert L. Hanson

**PS56 An improved T1D GRS for diagnosis of T1D**

S A Sharp, M N Weedon, A T Hattersley, K A Patel, W Hagopian, R A Oram