



**SGGD**

9th Meeting of the Study Group  
on Genetics of Diabetes

**EXETER 17-19TH APRIL 2024**



## **9<sup>th</sup> Meeting of the Study Group on Genetics of Diabetes Full programme**

Wednesday 17<sup>th</sup> April - Friday 19<sup>th</sup> April 2023  
Peter Chalk Centre, University of Exeter, Stocker Rd, Exeter EX4 4QD

### **Wednesday 17<sup>th</sup> April**

- 12:00 - 13:15**      **Lunch + Registration**
- 13:15 - 13:30**      Welcome and opening address (Andrew Hattersley)
- 13:30 - 15:00**      **Invited session 1- Using common genetic variants to diagnose and understand disease across ancestries**  
*Chairs - Tim Frayling (Geneva) and Miriam Udler (Boston)*
- 13:30 - 14:00**      Diagnosis and prediction of Type 1 diabetes (Richard Oram, Exeter)
- 14:00 - 14:30**      African genomes hold the key to accurate genetic risk prediction (Segun Fatumo, LSHTM)
- 14:30 - 15:00**      Applying Mendelian Randomization in obesity (George Davey Smith, Bristol)
- 15:00 - 15:30**      **Coffee Break**
- 15:30 - 16:30**      **Abstract selected oral presentation session 1**  
*Chairs Inês Barroso (Exeter) and Jose Florez (Boston)*
- 15:30 - 15:45**      Polygenic risk scores for the prediction of type 2 diabetes and complications in diverse ancestries (Alicia Huerta-Chagoya, MGH/Broad)
- 15:45 - 16:00**      A South Asian-specific missense variant in PIEZO1 may distort relationships between HbA1c and blood glucose (Miriam Samuel, QMUL)
- 16:00 - 16:15**      Identifying rare non-coding regulatory regions for HbA1c using Whole Genome Sequences (Gareth Hawkes, Exeter)
- 16:15 - 16:30**      Rare variant associations with birth weight highlight genetic links with later metabolic health (Robin Beaumont, Exeter)
- 16:30 - 16:45**      **Break**
- 16:45 - 17:45**      **Keynote** - Telomere to telomere human genomes and improved genotyping of complex structural variation (Evan Eichler, Washington)
- 17:45 - 19:00**      **Drinks Reception and poster viewing**

## Thursday 18<sup>th</sup> April

- 09:00 - 10:15**      **Invited session 2 - What's new in monogenic gene discovery**  
*Chairs - Pal Njølstad (Bergen) and Sadia Saeed (Lille)*
- 09:00 - 09:30**      Gene discovery in neonatal diabetes to understand beta-cell biology  
(Elisa De Franco, Exeter)
- 09:30 - 10:00**      From gene discovery to mechanism (Miriam Cnop, Brussels)
- 10:00 - 10:15**      Protein-truncating variants in BSN are associated with severe adult-onset obesity, type 2 diabetes and fatty liver disease (Yajie Zhao, Cambridge)
- 10:15 - 10:45**      **Highly Commended Poster Flash Talks**
- 10:45 - 12:15**      **Poster session and Coffee**
- 12:15 - 13:00**      **Lunch**
- 13:00 - 13:15**      **Sponsor talk - Oxford Nanopore Technologies**
- 13:15 - 14:45**      **Invited session 3 - From variant to function**  
*Chairs - Jorge Ferrer (Barcelona) and Endrina Mujica (Uppsala)*
- 13:15 - 13:45**      Translational Genomics of Type 2 Diabetes (Ele Zeggini, Germany)
- 13:45 - 14:15**      Linking metabolic disease polygenic scores to disease biology (Melina Claussnitzer, Boston (MGH/Broad))
- 14:15 - 14:45**      What can we learn from stem cell derived islets? (Timo Otonkoski, Helsinki)
- 14:45 - 15:45**      **Abstract selected Oral Session 2**  
*Chairs - Emma Ahlqvist (Lund) and Inga Prokopenko (Surrey)*
- 14:45 - 15:00**      Ultra-deep targeted transcriptome sequencing identifies isoform diversity across human pancreatic development (Ailsa MacCalman, Exeter)
- 15:00 - 15:15**      Age-related DNA methylation dynamics in insulin-sensitive tissues and their implications in metabolic health and obesity (Amna Khamis, Lille)
- 15:15 - 15:30**      Unravelling the interplay between type 2 diabetes, genetics and metabolite levels (Ozvan Bocher, Munich)
- 15:30 - 15:45**      Genetic architecture of oral glucose-stimulated insulin release provides biological insights into type 2 diabetes aetiology (Anne Madsen, Copenhagen)
- 15:45 - 16:15**      **Coffee Break**
- 16:15 - 17:15**      **Abstract selected Oral Session 3**  
*Chairs - Cecile Saint-Martin (Paris) and Antonio Cuesta (Malaga)*
- 16:15 - 16:30**      Homozygous mutations in SREK1 linked to a new condition of syndromic obesity (Sadia Saeed, Lille)
- 16:30 - 16:45**      Disease-causing low-level mosaic variants can be detected in blood samples from individuals with hyperinsulinism (Tom Laver, Exeter)
- 16:45 - 17:00**      From 58 type-2 diabetes candidate genes to 5: A validated 2-step in vivo prioritization system (Endrina Mujica, Uppsala)
- 17:00 - 17:15**      Homozygous and Heterozygous INS Mutations Cause Divergent Clinical and iPSC-Derived  $\beta$ -Cell Phenotypes (Yue Tong, Brussels)
- 19:00**              **SGGD dinner + live music**  
Holland Hall, University of Exeter, Clydesdale Road, Exeter, EX4 4SA  
**Dinner talk - Tom Staniford**

## Friday 19<sup>th</sup> April

- 09:00 - 10:30**      **Session 4 - Lessons from other diseases and technologies**  
*Chairs - Torben Hansen (Copenhagen) and Daniela Gašperíková (Slovakia)*
- 09:00 - 09:30**      Finding new diagnoses in the Deciphering Developmental Disorders Study  
(Caroline Wright, Exeter)
- 09:30 - 10:00**      Genetics and Genomics in Drug Discovery and Development (Rob Scott, GSK)
- 10:00 - 10:30**      Epigenetic and transcriptional pathways to brain disease (Jon Mill, Exeter)
- 10:30 - 11:00**      **Coffee Break with Traditional Devon Cream Tea**
- 11:00 - 13:00**      **Session 5 - Translating genetics to the clinic**  
*Chairs - Tiinamaija Tuomi (Helsinki) and Leen 't Hart (Leiden)*
- 11:00 - 11:30**      Precision medicine in diabetes (Ewan Pearson, Dundee)
- 11:30 - 12:00**      Are genetic diabetes clusters ready for the clinic? (Miriam Udler, Boston)
- 12:00 - 12:30**      The impact of rare variants in common diabetes and obesity towards precision  
medicine (Amélie Bonnefond, Lille)
- 12:30 - 13:00**      **Oral and poster awards + closing remarks**
- 13:00 - 14:15**      **Lunch & Meeting close**



**SGGD**

9th Meeting of the Study Group  
on Genetics of Diabetes

**EXETER 17-19TH APRIL 2024**

Supported by:

**RANDOX**

 Oxford  
**NANOPORE**  
Technologies