OMICRON Conference

EASD Study Group on Genetics of Diabetes Meeting

7-9 May 2015

Krakow, Poland

SCIENTIFIC PROGRAMME

omicron
Enabling OMICs high-tHROughput techNologies

JAGIELLONIAN UNIVERSITY
MEDICAL COLLEGE

This project has received funding from the European Union's Seventh Framework Programme for research, technological development and demonstration under grant agreement No 286038

Ministry of Science
and Higher Education
Republic of Poland

KNOW

Dear Friends,

On behalf of the Organizing Committee of the Fifth Meeting of the EASD Study Group on Genetics of Diabetes (EASD-SGGD) and the Jagiellonian University Medical College Omicron Project Conference, I cordially invite you to Krakow – a city of special significance to Poland, Europe and the entire world. With a history of more than a millennium, Krakow was the capital of the Polish kings for over 500 years, and to this day is the seat of the oldest Polish university and the heart of Polish culture and science.

Krakow was the birthplace, workplace and has been the resting place of many great Poles, including Saint Stanislaus of Szczepanow, Casimir III the Great, Hedwig d'Anjou, Nicolaus Copernicus, Tadeusz Kosciuszko, Jozef Pilsudski, Pope John Paul II, Polish Nobel prize winners and many others.

It has witnessed truly momentous events that have been recorded in history books, such as the establishment of the diocese in the year 1000, the martyrdom of Saint Stanislaw (1079), the unification of the state symbolized by the coronation of Ladislav I the Elbow-High (1320), the Prussian Homage (1525), the oath of the Commander of the National Uprising (1794). This was the place where Polish independence was regained (1918) and from where the future Pope John Paul II left to participate in a conclave (1978). The city witnessed thousands of other events which, even though are not found in history books, have made up the life of our nation and formed its scientific and cultural output. The approaching events should be rated among such happenings.

The SGGD-EASD meeting and the Omicron Conference will be held from 7-9 May, 2015 and will be an opportunity to meet hundreds of people – scientists, doctors and other professionals – who actively participate in research studies in the field of diabetes genetics, its complications and related diseases.

I am convinced that the panel of leading diabetes genetics experts will ensure an excellent scientific program, which will include keynote lectures, invited talks and oral presentations given by young scientists as well as poster discussions.

I am looking forward to seeing you in Krakow. I hope that you will leave this place enriched not only with knowledge, but also with many beautiful memories.

Prof. Maciej Małecki, MD, PhD
Chair of the Organizing Committee

HONORARY PATRONAGE

The OMICRON Conference and EASD-SGGD Meeting
are held under honorary patronage of
Prof. Lena Kolarska-Bobińska,
Minister of Science and Higher Education
and
Prof. Wojciech Nowak, Rector of the Jagiellonian University

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JAGIELLONIAN UNIVERSITY IN KRAKÓW

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Congress Topics

- Impact of next generation sequencing technologies on the genetics of diabetes.
- Translation of association signals to molecular mechanisms.
- Integration of genetic and genomic data sets.
- Clinical translation of basic science in genetics of diabetes.
- Biomarkers for diabetes.
- Impact of new data from genomics research and human physiology.
- Animal models and cell research in diabetes.
- New data on genetic architecture and its impact on the understanding of diabetes and diabetic complications.

Invited speakers & chairmen

Fabrizio Barbetti, Italy
Christine Bellanné-Chantelot, France
Miriam Cnop, Belgium
Antonio Luís Cuesta Muñoz, Denmark
Nancy Cox, USA
Józef Dulak, Poland
Sian Ellard, UK
Sarah Flanagan, UK
Jose Florez, USA
Timothy Frayling, UK
Philippe Froguel, UK
Tomasz Grodzicki, Poland
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Wojciech Młynarski, Poland
Pål Njølstad, Norway
Stephen O'Rahilly, UK
Marju Orho-Melander, Sweden
Katharine Owen, UK
Lorenzo Pasquali, Spain
Ewan Pearson, UK
Štěpánka Průhová, Czech Republic
Niina Sandholm, Finland
Naveed Sattar, UK
Robert Scott, UK
Jan Skupień, Poland
Juraj Staník, Slovakia
Ivan Tkáč, Slovakia

Reviewers

We are grateful to Prof. Rob Sladek, Prof. Graeme Bell and Prof. Mark McCarthy for judging the Young Investigator Award Contest, and to Prof. Rob Sladek and Prof. Graeme Bell for the judging the abstracts.

Conference venue

Auditorium Maximum of the Jagiellonian University
33 Krupnicza Street, Krakow, Poland

SCIENTIFIC PROGRAMME OVERVIEW

THURSDAY May 7, 2015		
13.00	**Registration**	
14.30-15.30	Omicron/SGGD & European Atherosclerosis Society Session	
15.45-16.45	SGGD & European Diabetic Nephropathy Study Group Session WHAT PROGRESS HAVE WE MADE TO DISSECT THE MOLECULAR BACKGROUND OF DIABETIC COMPLICATIONS?	
16.45-18.15	OPENING CEREMONY KEY NOTE LECTURE RISING STAR AWARD/YOUNG INVESTIGATOR LECTURE	
FRIDAY May 8, 2015		
08.30-10.30	UP-DATE ON NGS USE IN RESEARCH AND CLINICAL PRACTICE IN MONOGENIC BETA-CELL DISEASE	
10.30-11.00	**Posters**	
11.00-12.30	OBESITY, INSULIN RESISTANCE AND RELATED TRAITS	
13.15-15.00	ORIGINAL ORAL PRESENTATIONS **(SGGD)**	MEET THE EXPERTS - A CLINICAL SESSION ON MONOGENIC DIABETES **(Omicron)**
15.00-15.30	**Posters**	
15.30-17.00	KEY NOTE LECTURE II OMICRON/SGGD YOUNG INVESTIGATOR AWARDS 2015	
SATURDAY May 9, 2015		
08.30-10.00	FROM GENETICS TO GENOMICS AND HUMAN PHYSIOLOGY	
10.00-10.30	**Posters**	
10.30-13.00	KEY NOTE LECTURE III	
	ORIGINAL ORAL PRESENTATIONS **(SGGD)**	MEET THE EXPERTS - A CLINICAL SESSION ON MONOGENIC DIABETES **(Omicron)**
13.45-15.15	ANIMAL MODELS AND CELL RESEARCH	
15.15-16.15	PHARMACOGENETICS IN DIABETES	
16.15	CLOSING CEREMONY	

All Omicron/SGGD sessions will take place in Room A.

The Omicron *Meet the Experts* sessions will take place in Room C.

THURSDAY May 7, 2015

13:00	**Registration**
14:30-15:30	**Omicron/SGGD & European Atherosclerosis Society Session** Chairs: **Tomasz Grodzicki, Poland, Ryszard Korbut, Poland** Naveed Sattar, UK *Type 2 diabetes, biomarkers and genes: a view from the clinic.* Tomasz Guzik, Poland *Effects of chronic periodontitis treatment on hypertension and metabolic syndrome.*
15:30-15:45	**Coffee**
15:45-16:45	**SGGD & European Diabetic Nephropathy Study Group Session** **WHAT PROGRESS HAVE WE MADE TO DISSECT THE MOLECULAR BACKGROUND OF DIABETIC COMPLICATIONS?** Chairs: **Niina Sandholm, Finland, Marek Kuźniewski, Poland** Jan Skupień, Poland *Research outcomes in epidemiology and genetic of diabetic kidney disease.* Niina Sandholm, Finland *Genetics of nephropathy.*
16:45-18:15	**OPENING CEREMONY** **honored by the presence of the University's authorities and representatives of the local government.** Chairs: **Christine Bellanné-Chantelot, France, Tomasz Klupa, Poland** **KEY NOTE LECTURE** Andrew Hattersley, UK *Gene discovery: the beginning not the end.* **RISING STAR AWARD/YOUNG INVESTIGATOR LECTURE** Sarah Flanagan, UK *New insights from gene discovery in neonatal diabetes.*
18:30	**Welcome Party & Evening Social Event**

FRIDAY May 8, 2015

08:30-10:30	**UP-DATE ON NGS USE IN RESEARCH AND CLINICAL PRACTICE IN MONOGENIC BETA-CELL DISEASE** Chairs: **Fabrizio Barbetti, Italy, Antonio Luís Cuesta Muñoz, Denmark** Sian Ellard, UK *NGS for genetic testing for monogenic diabetes - are we ready for routine use in clinical practice?* Torben Hansen, Denmark *MODY-X search in the NGS era - are we stuck?* Katharine Owen, UK *Biomarkers in diabetes - from candidates to translation.* Christine Bellanné-Chantelot, France *Congenital hypersinsulinic hypoglycaemia: molecular mechanisms and therapeutic approaches.*
10:30-11:00	**Coffee & posters**
11:00-12:30	**OBESITY, INSULIN RESISTANCE AND RELATED TRAITS** Chairs: **Timothy Frayling, UK, Štěpánka Průhová, Czech Republic** Philippe Froguel, France *Will recent progress in genetics of obesity help us to personalize obesity therapy?* Robert Scott, UK *Genetics of polygenic insulin resistance - discovery and application to complex disease.* Marju Orho-Melander, Sweden *Do not forget about cholesterol! Recent advances in genetics of lipids abnormalities.*
12:30-13:15	**Lunch**

13:15-15:00	**ORIGINAL ORAL PRESENTATIONS/SGGD WORKSHOP 1 (SGGD)** Chairs: **Torben Hansen, Denmark, Katharine Owen, UK** Marine Cauchois-Le Mière, France, Cécile Saint-Martin, France *Minigene-based splicing assay: a useful approach to the interpretation of unclassified variants of the ABCC8 gene in hyperinsulinaemic hypoglycaemia.* Cécile Saint-Martin, France *Application of targeted Next-generation sequencing to the molecular diagnosis of MODY: Lessons from over 600 patients.* Kevin Colclough, UK *Quality improvements in diagnostic genetic testing and reporting of MODY through participation in the EMQN MODY EQA scheme.* Tibor V. Varga, Sweden *Novel genetic loci associated with long-term deteriorations in blood lipid concentrations and coronary artery disease in European adults.* Juraj Staník, Slovakia *Pharmacogenetics in channelopathies causing congenital hyperinsulinism in Slovakia.* Michael Weedon, UK *A novel, inexpensive test can discriminate between type 1 and type 2 diabetes.* Izortze Santin, Spain *The autoimmune disease candidate gene DEXI modulates virus induced pancreatic beta cell inflammation and death.* **MEET THE EXPERTS - A CLINICAL SESSION ON MONOGENIC DIABETES (Omicron)** Fabrizio Barbetti, Italy, Ewan Pearson, UK
15:00-15:30	**Coffee & posters**

15:30-17:45	**KEY NOTE LECTURE II** Chairs: **Andrew Hattersley, UK, Rohit Kulkarni, USA** Stephen O'Rahilly, UK *Current understanding of genetics and metabolism of human severe insulin resistance.* **OMICRON/SGGD YOUNG INVESTIGATOR AWARDS 2015** Nikolay Oskolkov, Sweden *GWAS of histological phenotypes provides insights into the genetic architecture of human skeletal muscle.* Fainia Kavvoura, UK *Can genomic information assist in differentiating aetiology in young adult onset diabetes?* Elisa De Franco, UK *Genomic testing leads clinical care in neonatal diabetes: a new paradigm.* Kashyap Patel, UK *Type 1 diabetes genetic risk score - a novel tool to differentiate monogenic diabetes from T1D,*
19:00	**Dinner**

SATURDAY May 9, 2015

08:30-10:00	**FROM GENETICS TO GENOMICS AND HUMAN PHYSIOLOGY** Chairs: **Sian Ellard, UK, Philippe Froguel, UK** Leif Groop, Sweden *Can genetics of diabetes be translated into the clinic?* Jose Florez, USA *Blurred boundaries between monogenic diabetes and T2DM-related rare variants.* Pal Njolstad, Norway *From GWAS signals to biological mechanisms of diabetes.*
10:00-10:30	**Coffee & posters**
10:30-13:00	**KEY NOTE LECTURE III** Chairs: **Jose Florez, USA, Leif Groop, Sweden** Nancy Cox, USA *New Aspects of Genetic Architecture to Diabetes and Diabetic Complications.* **ORIGINAL ORAL PRESENTATIONS/SGGD WORKSHOP 2 (SGGD)** Reedik Magi, Estonia *Genome-wide association study imputed to 1000 Genomes Project reference panels reveals 17 novel associations with type 2 diabetes.* Matthew Johnson, UK *Recessively inherited LRBA mutations can caus neonatal or infancy onset diabetes with autoimmunity.* Jonathan Locke, UK *A cautionary tale: the non-causal association between type 2 diabetes risk SNP, rs7756992, and levels of non-coding RNA, CDKAL1-v1.* Rashmi Prasad, Sweden *Multiple type 2 diabetes loci influence body composition by parent-of-origin effects.*

10:30-13:00	Momoko Horikoshi, UK *Large scale Genome-wide association study for birth weight finds eight novel loci extending the genetic link between early growth and type 2 diabetes.* Anubha Mahajan, UK *Identification of protein-coding variants associated with type 2 diabetes.* **MEET THE EXPERTS - A CLINICAL SESSION ON MONOGENIC DIABETES (Omicron)** Fainia Kavvoura, UK, Katharine Owen, UK, Antonio Luís Cuesta Muñoz, Denmark, Wojciech Młynarski, Poland
13:00-13:45	**Lunch**
13:45-15:15	**ANIMAL MODELS AND CELL RESEARCH** Chairs: **Józef Dulak, Poland, Timothy Frayling, UK** Rohit Kulkarni, USA *Using induced pluripotent stem cells to interrogate the pathophysiology of diabetes.* Miriam Cnop, Belgium *Organelle dysfunction in beta cells and human diabetes.* Lorenzo Pasquali, Spain *Non-coding genome pancreatic function.*
15:15-16:15	**PHARMACOGENETICS IN DIABETES** Chairs: **Wojciech Młynarski, Poland, Juraj Staník, Slovakia** Ivan Tkáč, Slovakia *Pharmacogenomics in type 2 diabetes.* Tim Frayling, UK *Physiology helps genetics take a step closer to mechanism.*
16:15	**CLOSING CEREMONY**
18:00	**Dinner**

POSTERS

P.01 **Pleiotropy or independent genetic effects? - A family study of birth anthropometrics and measures of body composition, beta-cell function and insulin sensitivity in adults.**
A. P. Gjesing, Denmark

P.02 **Parent of origin effects of lipid loci on obesity related traits.**
A. Lessmark, Sweden

P.03 **The low frequency type 2 diabetes protective allele at CCND2 is associated with enhanced insulin secretion and higher skeletal growth from birth to adulthood.**
H. Yaghootkar, UK

P.04 **Genome-wide multi-phenotype rare variant association analysis detects effect of ZNF259 on fasting insulin and triglyceride levels.**
M. Kaakinen, UK

P.05 **High-density imputation and trans-ethnic association analysis reveals a novel locus for type 2 diabetes susceptibility.**
A. P. Morris, UK

P.06 **Value of genetic risk score in personalized risk prediction of type 2 diabetes: analysis of 10273 individuals of the Estonian Biobank cohort.**
K. Fischer, Estonia

P.07 **Comparing polygenic risk scores for type 2 diabetes.**
K. Läll, Estonia

P.08 **The impact of known type 2 diabetes associated variants on circulating levels of GLP-1, GIP and glucagon during an oral glucose tolerance test.**
A. Jonsson, Denmark

P.09 **PLCL2 - the only gene without LoF mutations in 61K individuals that has islet expression associated with HbA1c and islet ß cell specificity.**
J. Fadista, Sweden

P.10 **Identification of functional effects of common and low frequency genetic variants in the ADIPOQ gene using individuals recruited by genotype.**
B. P. Lee, UK

P.11 **Variants in candidate genes and glycaemic response to metformin treatment in Slovak type 2 diabetes patients: association study.**
M. Javorský, Slovakia

P.12 **A Recombined diabetes gene as risk factor for chronic pancreatitis.**
K. Fjeld, Norway

P.13 **Screening for genes involved in beta-cell proliferation and resistance to death by lipids.**
L. Boquete Vilariño, UK

P.14 **Expanding the clinical and mutational spectrum of IPEX syndrome with FOXP3 mutations.**
J. A.L. Houghton, UK

P.15 **The length of the deletion in region 17q contributes to the individual variability of the phenotype of patients with renal cysts and diabetes syndrome (RCAD, HNF1B-MODY)**
Š. Průhová, Czech Republic

P.16 **Contribution of salivary and pancreatic amylase enzymatic activity to metabolic traits in the French cohort D.E.S.I.R.**
A. Bonnefond, France

P.17 **Qualitative analysis of the bacterial flora of the duodenal biopsy specimens in patients with obesity and type 2 diabetes using Next generation sequencing method.**
A. Sroka, Poland

P.18 **Search for the markers of disease progression among patients with Wolfram syndrome.**
A. Zmysłowska, Poland

P.19 **Prevalence of retinopathy in adult patients with GCK-MODY and HNF1A-MODY.**
M. Szopa, Poland

P.20 **Comparison of glomerular filtration rate estimation from serum creatinine and cystatin C in HNF1A-MODY and other types of diabetes.**
M. Szopa, Poland

P.21 **Quality of life assessment in patients with genetic diagnosis of HNF1A-MODY and GCK-MODY.**
M. Szopa, Poland

P.22 **Circulating ghrelin level is higher in HNF1A-MODY and GCK-MODY than in polygenic forms of diabetes mellitus.**
N. Nowak, Poland

P.23 **The comparison of the bacterial flora of gastrointestinal tract in patients with type 2 diabetes mellitus and HNF1A-MODY diabetes.**
S. Mrozińska, Poland

P.24 **Using different workflows to investigate monogenic diabetes in Slovakia.**
M. Skopková, Slovakia

P.25 **Molecular diagnosis of monogenic diabetes by targeted Next-generation sequencing in Slovak MODY patients - first results.**
L. Valentinová, Slovakia

P.26 **DNA testing of M.3243A>G mutation in patients with clinical suspicion on MODY or MIDD in Slovakia.**
M. Skopková, Slovakia

P.27 **Prevalence of monogenic diabetes in the Norwegian childhood diabetes registry estimated by targeted deep sequencing.**
B.B. Johansson, Norway

P.28 **Type 1 diabetes prediction program: additional source of MODY patients?**
P. Dusátková, Czech Republic

P.29 **The first two probands diagnosed by whole exome sequencing through Slovakia WES project.**
D. Gasperiková, Slovakia

P.30 **Diagnosis of Maturity Onset Diabetes of the Young at presentation.**
A. Juszczak, UK

P.31 **From whole exome sequencing data to pathogenic variants using linux text formating utilities and publicly available tools.**
D. Danis, Slovakia

P.32 **Women with gestational diabetes diagnosed by IADPSG criteria have a different genetic predisposition from those diagnosed by previous WHO criteria.**
R. M. Freathy, UK

P.33 **Exome sequencing identifies a novel mutation in STAT3 gene associated with neonatal diabetes and early onset-autoimmune disease.**
T. Velayos, Spain

P.34 **Hyperglycemia preceded by neonatal hyperinsulinemic hypoglycemia in infants with novel HNF1A mutations.**
J. Malíková, Czech Republic

P.35 **Specific effects of PTPN22 c.1858T>C and FCRL3 -169C>T polymorphisms co-occurrence in children with newly diagnosed type 1 diabetes from Pomeranian Region of Poland.**
M. Pawłowicz, Poland

P.36 **Sex-specific associations of PTPN22 c.1858T allele with diabetic ketoacidosis and worse clinical course of disease in children with newly diagnosed type 1 Diabetes from Pomeranian Region of Poland.**
M. Pawłowicz, Poland

P.37 **Joint effects of selected GCK and FCRL3 polymorphisms on dynamics of residual β-cells function in children with early clinical phase of type 1 diabetes.**
M. Pawłowicz, Poland

P.38 **Human population variations in GRP94 chaperone impact on production of insulin-like growth factors.**
Y. Argon, USA

P.39 **Reduced soluble CD93 are not a result of T2D but might be a cause.**
R. J. Strawbridge, Sweden

**A full list of authors is available online*

Basement of the building located at 7 Kopernika St, Krakow before its full renovation in 2011

A current view of the OMICRON Laboratory, at the end of the Project