

Summer School 2021 Genetic Epidemiology of Kidney Function and Chronic Kidney Disease

July 12-16, 2021 ONLINE EDITION

Programme

Day 1

Session	Content	Lecturers
12-1 pm Getting to know each other	Participants informal introduction	Students + Scientific committee
1-1.45 pm Introduction & welcome	General welcoming 1. Course overview 2. Motivational lecture: Genetic modifiers in CKD	Peter P. Pramstaller, Head of the Eurac Research Institute for Biomedicine A. Köttgen, C. Pattaro F. Terzi
2-6 pm Basic principles of genetic association studies	3. 2-2:45 pm: Genetic Epidemiology: [complex] phenotypes first 4. 3-3:45 pm: Principles of population genetics 5. 4-5 pm: Genome-wide association studies 6. 5:15-6 pm: Sequencing studies, gene-based testing	C. Pattaro C. Pattaro C. Fuchsberger E. Koenig

Mon 12

12:00 – 6:00 pm

Day 2

Session	Content	Lecturers
1-1.45 pm exchange	Poster presentation by students, group 1 1. Discovery and fine-mapping of kidney function loci in first genome-wide association study in Africans Segun A Fatumo <i>MRC/UVRI and LSHTM Uganda Research Unit, Ugandan Medical Informatics Centre (UMIC), Entebbe, Uganda</i> 2. Investigating genetic influences on metabolism in a CKD patient cohort Franziska Grundner-Culemann <i>Institute of Genetic Epidemiology, Freiburg University</i> 3. Dissection of HLA-C gene region to investigate its association with complex traits Elena Locatelli, Moron Dalla Tor L, Treccani M, Veschetti L, De Tomi E, Stefani C, Tamburin S, Zipeto D, Trabetti E, Patuzzo C, Malerba G <i>Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona, Verona, Italy</i> 4. An intermediate-effect variant in UMOD confers risk for chronic kidney disease Eric Olinger, C Schaeffer, K Kidd, Y Cheng, I Wilson, <i>Genomics England Research Consortium, K-U Eckardt, A Bleyer, M Wiesener, A Köttgen, J Sayer, L Rampoldi, O Devuyst</i>	Students, C. Pattaro

Tue 13

1:00 – 6:00 pm

<p>2-6 pm From loci to causal variants</p>	7. 2-2:45 pm: Applying fine-mapping for characterizing GWAS loci associated with eGFR and CKD	M. Wuttke
	8. 2:45-3:30 pm: Implementing gene expression and colocalization for identifying genes associated with albuminuria	A. Teumer
	9. 3:45-4:30 pm: Approaches to prioritize causal genes and perform gene-based enrichment analyses using GWAS of urine metabolites	P. Schlosser
	10. 5:15-6 pm Polygenic risk scores - development and applications	M. Scholz

Day 3

Session	Content	Lecturers
<p>1-1.45 pm exchange</p>	Poster presentation by students, group 2	<p>Students, A. Köttgen</p>
	<p>1. Comprehensive cell atlas of the adult mouse kidney Claudio Novella-Rausell, A Mahfouz , DJM Peters, M Grudniewska <i>GenomeScan B.V., Leiden, The Netherlands</i></p>	
	<p>2. Investigation of an LPA KIV-2 nonsense mutation in 11,000 individuals: Mind linkage disequilibrium in Lipoprotein(a) genetics. Silvia Di Maio <i>Institute of Genetic Epidemiology, Medical University of Innsbruck</i></p>	
	<p>3. Induction of selective apoptosis in cyst-lining cells as a novel strategy to delay the progression of autosomal dominant polycystic kidney disease (ADPKD) Carlotta Pioppini, DE Yilmaz, M Krappitz, K-U Eckardt <i>Department of Nephrology and Medical Intensive Care, Charité - Universitätsmedizin Berlin</i></p>	
<p>2-6 pm Exploring causal pathways using in silico approaches</p>	4. Mendelian Randomization for Estimating Causal Effects of Estradiol on Kidney Function Mohammed Nars Kamal <i>Hasso Plattner Institute, Potsdam University</i>	
	11. 2-3:15 pm Mendelian randomization (MR): an introduction	A. Tin, A. Teumer
	12. 3:30-4:10 pm Using MR to test causal molecular pathways	A. Teumer
	13. 4:10-4:50 pm Using MR to assess epidemiological causality at the disease level	A. Tin
	14. 5-6 pm Practical MR session using R	D. Bottigliengo

Wed 14
1:00 – 6:00 pm

Day 4

Session	Content	Lecturers
<p>12:30-2:50 pm journal club</p>	• 12:30-12:40 Introduction	A. Köttgen
	• 12:40-1:50 pm Student interactions within groups: discuss paper	Students

Thu 15

12:30 – 5:30 pm

	<ul style="list-style-type: none">• 2-2:50 pm: JC: Complex disease: polygenic background as genetic modifier	Students, A. Köttgen
3-5:30 pm Translation from the industry perspective	15. 3-4 pm: Using Genetics to Drive Drug Discovery and Development	C. S. Fox
	16. 4:15-5:30 pm: Kidney Diseases Research at Bayer	F. Eitner

Day 5

Fri 16
12:00 – 5:00 pm

Session	Content	Lecturers
12-3:15 pm Experimental identification of causal mechanisms	17. 12-1 pm Translating GWAS findings into molecular insights: the UMOD example	O. Devuyst
	18. 1-2 pm Experimental approaches to identify gene functions and causal mechanisms	M. Köttgen
	19. 2:15-3:15 pm Genetic modifiers of HNF1a/b induced disease	M. Pontoglio
3:30-5 pm exchange	3:30-4:45 Interactive summary session	A. Köttgen, Students
	4:45-5 pm Concluding remarks	C. Pattaro, A. Köttgen

Additional lectures will be available as online material:

- ✓ Integrating Multiomics Data for a Better Understanding of Kidney Disease (A. Köttgen)

Teaching faculty

BOTTIGLIENGO, Daniele
Eurac Research, Institute for Biomedicine, Bolzano, Italy

DEVUYST, Olivier
University of Zurich, Department of Physiology, Zurich, Switzerland

EITNER, Frank
Head of Kidney Diseases, Research & Development, Pharmaceuticals, Bayer AG, Wuppertal, Germany

FOX, Caroline S
Vice President and Head, Genetics and Pharmacogenomics, Merck Research Labs, Boston, USA

FUCHSBERGER, Christian
Eurac Research, Institute for Biomedicine, Bolzano, Italy

KÖNIG, Eva
Eurac Research, Institute for Biomedicine, Bolzano, Italy

KÖTTGEN, Anna
University of Freiburg, Institute of Genetic Epidemiology, Freiburg, Germany

KÖTTGEN, Michael
University of Freiburg, Department of Medicine IV, Freiburg, Germany

PATTARO, Cristian
Eurac Research, Institute for Biomedicine, Bolzano, Italy

PONTOGLIO, Marco
Inserm and Université de Paris, Necker Hospital, Paris, France

SCHLOSSER, Pascal
University of Freiburg, Institute of Genetic Epidemiology, Freiburg, Germany

SCHOLZ, Markus
University of Leipzig, Institute for Medical Informatics, Statistics and Epidemiology, Leipzig,
Germany

TERZI, Fabiola
Inserm and Université de Paris, Necker Hospital, Paris, France

TEUMER, Alexander
University Medicine Greifswald, Institute for Community Medicine, Greifswald, Germany

TIN, Adrienne
University of Mississippi Medical Center, Jackson, USA

WUTTKE, Matthias
University of Freiburg, Institute of Genetic Epidemiology, Freiburg, Germany

Organisers & Partners

eurac
research



This project has received funding from the European Union's Horizon 2020 research and innovation programme under the Marie Skłodowska-Curie grant agreement No 860977