



Università di Cagliari



Universität Heidelberg

# European Mathematical Genetics Meeting 2018 Cagliari, Italy, April 18<sup>th</sup> to 20<sup>th</sup>

The event is sponsored by and organized under the auspices of  
the University of Cagliari

## *Conference Site*

Università di Cagliari, Facoltà di Scienze Economiche, Giuridiche e Politiche  
Via Sant'Ignazio 76, Cagliari

## *Opening and Lectures*

Room B0-A

## *Poster Area*

next to Room B0-A

## *Course*

Computer Room LISS-D

## **Organizing Committee**

Francesco Cucca	Università di Sassari and Istituto di Ricerca Genetica e Biomedica at the National Research Council of Italy
Marcella Devoto	University of Pennsylvania and Università Sapienza di Roma
Christine Fischer	Universität Heidelberg
Justo Lorenzo Bermejo	Universität Heidelberg

## **Local Organizing Committee**

Claudio Conversano	Università di Cagliari
Alessio Squassina	Università di Cagliari

## **Local Contacts**

*Claudio Conversano, Department of Business and Economics, Università di Cagliari.  
Tel. +39 070 6753337; email: [conversa@unica.it](mailto:conversa@unica.it)*

*Alessio Squassina, Department of Biomedical Sciences, Università di Cagliari.  
Tel. +39 349 2891006; email: [squassina@unica.it](mailto:squassina@unica.it)*

## *Wednesday 18<sup>th</sup>*

8:30 – 9:00

### **Registration**

9:00 - 14:00

### **Preconference course "Introduction to population genetics"**

Benjamin Peter (Leipzig), Christine Fischer, Justo Lorenzo Bermejo (Heidelberg)

16:30 - 17:00

### **Conference Registration**

17:00

### **Opening**

17:15-18:15

### **Keynote lecture: Genetic Architecture of Sardinians**

Francesco Cucca (Sassari)

19:00

### **Welcome Reception**

## Thursday 19<sup>th</sup>

9:00 - 10:30

### *Population stratification and admixture*, Chairs: Justo Lorenzo Bermejo, Hui Guo

Jonas Meisner	Inferring population structure and admixture proportions in low depth next-generation sequencing data
Edmund Gilbert	Shared genetic ancestry of Scotland and Ireland reveals fine scale structure
Heidi Hautakangas	Effect of reference panel in LD score regression analysis of lipid traits in the Finnish population
Mart Kals	Population-specific imputation reference panel as a tool for GWAS analysis
Regina Brinster	Optimal selection of genetic variants for adjustment of population stratification in European association studies

10:30 - 11:00

Coffee break

11:00 - 12:30

### *Results of association studies*, Chairs: Inke König, Svetlana Cherlin

Peter K Joshi	The genomic basis of human lifespan
Zoltán Kutalik	A joint analysis of adiposity genetics unravels subtypes with different metabolic implications
Erin Macdonald-Dunlop	Genome-wide association analysis of 225 metabolites in isolated populations
Marika Kaakinen	Genome-wide association study of 1,124 protein levels in pulmonary arterial hypertension patients identifies a novel trans-pQTL at ELK2AP for death receptor
Eleonora Porcu	Mendelian randomization combining GWAS and eQTL data reveals new loci, extensive pleiotropy and genetic determinants of complex and clinical traits

12:30 - 14.00

Lunch and posters

Thursday poster authors are at their posters 13:15 - 14:00

14:00 - 15:30

### *Autozygosity Polygenic Risk Scores, and more*, Chairs: Michael Nothnagel, Peter Joshi

James F Wilson	Sex-specific inbreeding depression in humans
Krista Fischer	Integration of polygenic risk estimates into personalized risk prediction algorithms – experience from the Estonian biobank
Svetlana Cherlin	Prediction of treatment response from genome-wide SNP data in rheumatoid arthritis patients
Sini Kerminen	Evaluating robustness and geographic differences in polygenic risk in Finland
Amke Caliebe	Ancient DNA study reveals HLA susceptibility locus for leprosy in medieval Europeans

15:30 - 16:00

Coffee break

16.00 - 17:20

### *Methods for Pedigree Data*, Chair: Marcella Devoto

Suzanne M. Leal	A rare variant nonparametric linkage method for nuclear and extended pedigrees with application to late onset Alzheimer's disease whole genome sequence
Alexandra Lefebvre	An Elston-Stewart algorithm for computing exact derivatives of the likelihood in pedigrees
Mohamad Saad	Comparison and assessment of family- and population-based genotype imputation methods in large pedigrees
Renaud Tissier	Ascertainment corrections for secondary phenotypes analysis in family studies

17:20 - 17:30

Future EMGMs

20:00

**Conference Dinner** A'mare (c/o Il Lido), Viale Poetto 41 Cagliari

## Friday 20<sup>th</sup>

9.00 - 10:50

### **Multiple Phenotypes and Imputation, Chairs: Zoltan Kutalik, Marika Kaakinnen**

- Andrew T. DeWan A comparison of univariate and multivariate GWAS methods for analysis of multiple dichotomous phenotypes
- Harmen Draisma Multi-phenotype epigenome-wide association analysis of fasting glucose and insulin in 1,105 Finnish individuals
- James J. Fryett Improving gene expression prediction accuracy in transcriptome-wide association studies
- Saurabh Ghosh Association mapping of multivariate phenotypes in the presence of missing data
- Richard Howey Imputation of missing data for Bayesian network analyses of complex biological data
- J. Asimit A Bayesian joint fine mapping approach that shares information between related autoimmune diseases increases accuracy and identifies novel associations

10:50 - 11:20

Coffee break

11:20 - 12:30

### **Heritability**

**Chair: Heike Bickeböllner**

- Arthur Frouin Quantify genomic heritability through a prediction measure
- Anthony F. Herzig What insights can be gained through comparisons of broad-sense heritability estimates in isolated and outbred populations?
- Nicola Pirastu The exposural landscape of coronary artery disease gives new insight in its aetiology and missing heritability
- Xia Shen High-definition likelihood inference for heritability and genetic correlation using GWAS summary statistics

12:30 - 14.00

Lunch and posters

Friday poster authors are at their posters 13:15 - 14:00

14:00 - 15:30

### **Software, platform, and more, Chairs: Stefan Böhringer, Reedik Mägi**

- Emil Jørsboe fastNGSadmix: Admixture proportions and principal component analysis of a single low-depth sequencing sample
- Iuliana Ionita-Laza A semi-supervised approach for predicting cell type/tissue specific functional consequences of non-coding variation using massively parallel reporter assays
- Mauro Pala 3. The eQTLs Catalog and LinDA browser: a platform for determining the effects on transcription of GWAS variants
- Adam Waring Beyond burden testing – association analysis of ultra-rare variants
- Hui Guo Genetic causal pathways: Mendelian randomization, fine-mapping and colocalization

15:30 - 16:00

Coffee break

16:00 - 17:30

### **Statistical methods and modeling, Chairs: Heather Cordell, Eleonora Porcu**

- Märt Möls A statistical method for alignment-free analysis of sequencing reads with applications in copy number determination and plasmid integration detection
- Stefan Böhringer Exact model comparisons when determining parent-of-origin effects
- Zheng Ning A selection operator for summary association statistics reveals allelic heterogeneity of complex traits
- Michael Nothnagel Impact of pathway structures on allelic spectra of diseases
- Stephan Seifert Combining surrogate variables and minimal depth variable importance

17:30

### **Awards & Farewell**

*Poster discussion with authors: odd numbers on Thursday and even numbers on Friday;  
13:15 - 14:00*

- 1 David Almorza Principal components analysis in the phenotypic selection of inbred lines
- 2 Mila D. Anasanti Modern approaches to address missing data in multi-phenotype genome-wide association studies
- 3 Felix Boekstegers Pleiotropic effects of genetic variants on gallstone disease and gallbladder cancer in Europeans and Chileans
- 4 Stefano Calza Cell-specific somatic mutation detection from single-cell RNA-sequencing
- 5 Valentina Cipriani An improved bioinformatics tool for rare disease variant prioritization: the Exomiser 9.0.1 in clinical practice
- 6 Claire Dandine-Roulland Genome-wide data manipulation, association analysis and heritability estimates in R with GASTON 1.5
- 7 Mariza de Andrade An efficient test for gene-environment interaction in generalized linear models with family data
- 8 Damian Gola Classification of CAD status using machine learning approaches
- 9 Rosa González Silos Allele counts: a good alternative for testing genetic association in next generation sequence data
- 10 Katherine A. Kentistou Assessing novel anthropometric indices and their predictive efficacy over metabolic health and disease
- 11 Kristi Läll Comparison of genetic risk scores in the Estonian biobank cohort
- 12 Maarja Lepamets New quality measure for CNV: a multi-omics approach
- 13 Reedik Mägi How well does parental genetic risk predict early menopause genetic risk in offspring?
- 14 Merli Mändul A cluster-randomized trial on personalized feedback on genetic risks: effects on treatment compliance of patients with hypertension
- 15 Ninon Mounier Bayesian genome-wide association study to discover novel lifespan-associated loci
- 16 Carlos Pinto Comparison of the performance of polygene scores and artificial neural networks in the classification of disease status
- 17 Linda Repetto Investigating pleiotropic architecture of plasma proteins using multivariate methods
- 18 Albert Rosenberger Genetic modifiers of radon induced lung cancer risk - a genome-wide interaction study in former uranium miners
- 29 Sina Rüeger Drugs for specific diseases tend to target genes whose expression is causally linked to those diseases
- 20 Sanni Ruotsalainen Phenome wide scan of *ANGPT14* e40k mutation reveals new insights for future drug development
- 21 Kristina Schlicht Genetic imbalance affects functional outcome after ischemic stroke
- 22 Gulnara Svishcheva Region-based association analysis of summary statistics using principal components and functional linear regression models
- 23 Nele Taba Empirically derived dietary patterns – characteristics, phenotypic background, and associations with NMR metabolites and health outcomes in the Estonian biobank cohort
- 24 Tõnis Tasa External evaluation of population pharmacokinetic for vancomycin in neonates with DosOpt
- 25 Hande Topa Searching for genetic variants matching a given multivariate target profile
- 26 Viola Tozzi Coverage of the Oncoarray
- 27 Steffen Uebe Combined CNV and SNP ancestry analysis
- 28 Maren Vens Is the healthy migrant effect heritable? Population structure and demographic history of resettlers and the autochthone German population
- 29 Jonathan Sulc Genetic subtypes of obesity and their impact on health