European Mathematical Genetics Meeting 2018
Cagliari, Italy, April 18th to 20th

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 Counsil 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

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Wednesday 18th

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 19th

**9:00 - 10:30**  
*Population stratification and admixture*, Chairs: Justo Lorenzo Bermejo, Hui Guo

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<tr>
<th>Speaker</th>
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<tr>
<td>Jonas Meisner</td>
<td>Inferring population structure and admixture proportions in low depth</td>
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<td>generation sequencing data</td>
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<td>Edmund Gilbert</td>
<td>Shared genetic ancestry of Scotland and Ireland reveals fine scale</td>
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<td>Heidi Hautakangas</td>
<td>Effect of reference panel in LD score regression analysis of lipid</td>
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<td>traits in the Finnish population</td>
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<td>Mart Kals</td>
<td>Population-specific imputation reference panel as a tool for GWAS</td>
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<td>Regina Brinster</td>
<td>analysis</td>
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**10:30 - 11:00**  
Coffee break

**11:00 - 12:30**  
*Results of association studies*, Chairs: Inke König, Svetlana Cherlin

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

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<tr>
<td>James F Wilson</td>
<td>Sex-specific inbreeding depression in humans</td>
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<td>Krista Fischer</td>
<td>Integration of polygenic risk estimates into personalized risk</td>
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<td>prediction algorithms – experience from the Estonian biobank</td>
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<td>Svetlana Cherlin</td>
<td>Prediction of treatment response from genome-wide SNP data in</td>
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<td>rheumatoid arthritis patients</td>
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<td>Sini Kerminen</td>
<td>Evaluating robustness and geographic differences in polygenic risk</td>
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<td>in Finland</td>
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<td>Amke Caliebe</td>
<td>Ancient DNA study reveals HLA susceptibility locus for leprosy in</td>
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<td>medieval Europeans</td>
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**15:30 - 16:00**  
Coffee break

**16.00 - 17:20**  
*Methods for Pedigree Data*, Chair: Marcella Devoto

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<tr>
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<tr>
<td>Suzanne M. Leal</td>
<td>A rare variant nonparametric linkage method for nuclear and extended</td>
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<td>pedigrees with application to late onset Alzheimer’s disease whole</td>
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<td>Alexandra Lefebvre</td>
<td>An Elston-Stewart algorithm for computing exact derivatives of the</td>
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<td>Mohamad Saad</td>
<td>Comparison and assessment of family- and population-based genotype</td>
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<td>imputation methods in large pedigrees</td>
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<td>Renaud Tissier</td>
<td>Ascertainment corrections for secondary phenotypes analysis in family</td>
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<td>Future EMGMs</td>
<td>studies</td>
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**17:20 - 17:30**

**20:00**  
Conference Dinner  
A’mare (c/o Il Lido), Viale Poetto 41 Cagliari
Friday 20th

9.00 - 10:50 **Multiple Phenotypes and Imputation**, Chairs: Zoltan Kutilik, 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 Bickeböller

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

Märk 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

19. 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 ANGPT1 e40k mutation reveals new insights for future drug development

21. Kristina Schlicht  
    Genetic imbalance affects functional outcome after ischemic stroke

22. Gulnara Svischeva  
    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