

# RT STATGEN: ANNUAL MEETING

## Tuesday December 9, 8h30-18h30

8h30-9h00 Welcome Coffee

9h00-9h05 Introduction Emmanuelle Génin IT GGB & Robert Barouki IT SP

9h05-9h15 Program overview, Anne-Louise Leutenegger and Emmanuelle Bouzigon

### Session 1: Statistics and genetic epidemiology

**Chair: David-Alexandre Tregouet & Anne-Louise Leutenegger**

- 9h15-9h45: Keynote Speaker - Iuliana Ionita-Laza, Domain-aware phenotype prediction from Electronic Health Record data with applications in genomic research
- 9h45-10h05: Berrandou Takiy, Multi-trait genome-wide and gene-based analyses implicate coagulation and vascular smooth-muscle pathways in spontaneous coronary artery dissection.
- 10h05-10h25: Huang Haibo, Genetic Correlations Between Asthma Subtypes and Neuropsychiatric Disorders.
- 10h25-10h45: Fritsch Humblet Barbara, ATM, medical ionising radiation and breast cancer risk in high-risk women without pathogenic variants in BRCA1 and BRCA2.

10h45-11h15 Coffee Break + Posters

**Chair: Céline Bellenguez & Emmanuelle Bouzigon**

- 11h15-11h35: Palmal Sagnik, Interactions with genetics in Alzheimer's disease.
- 11h35-11h55: Schramm Catherine, Estimation of Alzheimer disease risk curves as a function of age and genetic risk factors.
- 11h55-12h15: Foulon Sidonie, Fantasio: A Case-Control Approach To Detect Rare Recessive Variants In Multifactorial Diseases.
- 12h15-12h35: Herzig Anthony, Impact of study sample composition on supervised admixture modelling.

12h35-14h00 Lunch Break + Posters

### Session 2: Bioinformatics and omics analysis

**Chair: Nicolas Vince & Emmanuelle Bouzigon**

- 14h00-14h30: Keynote Speaker - Antonio Rausell, Novel approaches for disease gene identification: from burden tests to network-based digenism prediction.
- 14h30-14h50: Asgari Yazdan, DNA Methylation and Breast Cancer Risk: An Epigenome-Wide Association Study within the French E3N-Generations Cohort.
- 14h50-15h10: Perrin Aglaé, Assessing the association between human genetic variation and SARS-CoV-2 viral dynamics.

15h10-15h40 Coffee Break + Posters

**Chair: Aurélie Cobat & Anne-Louise Leutenegger**

- 15h40-16h00: Chataigner Lucas, When Ancestry Matters: Visualizing and Profiling Missense Variants in Drug-Response Genes.
- 16h00-16h20: Paes Gabriela, Population-Specific Diversity of KIR2DS1 and KIR2DS4 Activating Receptors: Copy Number Variation and Allelic Patterns Across Global Ancestries.
- 16h20-16h40: Monlong Jean, Association tests in pangenome graphs with STOAT.

16h40-16h45 Conclusion

16h45-18h30 Cocktail, discussion & networking

## Poster Presentations

### *Morning - 10h45-11h15 (odd)*

1. Abani Fatima-Zahra, Post-GWAS Analysis Pipeline for Whole-Genome PacBio HiFi Sequencing: Investigating Non-Coding and Structural Variants in Early-Onset Alzheimer's Disease.
3. Brocard Simon, First genomic study of chronic lung allograft dysfunction discovers two novel associations.
5. Conil Clément, A human YEATS4 variant confers resistance to TST and IGRA conversion despite Mycobacterium tuberculosis exposure.
7. Dowding Julien, Genome-wide association study of IgG response to Ebola virus antigens in survivors of Ebola virus disease.
9. Gélín Morgane, Identification of genetic susceptibility to develop invasive pneumococcal disease in children by whole-exome sequencing.
11. Le Borgne Julie, Gene-sex interaction GWAS for Alzheimer's Disease risk for StatGen 2025.
13. Pluntz Matthieu, Generalized Viterbi algorithm for identifying the multiple most likely paths of hidden states in a HMM and application to detection of homozygosity by descent.
15. Mauduit Vincent, Genome-wide survival study identifies a novel non-HLA donor-recipient genetic mismatch associated with kidney allograft survival.
17. Orsi Laurent, Genes in the Interleukin-6-related pathways of asthma: A meta-analysis of four studies.
19. Sugier Pierre-Emmanuel, Meta-Analysis models with group structure for pleiotropy detection at gene and variant level using summary statistics from multiple datasets.

### *Afternoon - 15h10-15h40 (even)*

2. Barzine Mitra, Fine-mapping of human leukocyte antigen alleles associated with French anti-aquaporin-4 antibodies seropositive Neuromyelitis Optica Spectrum Disorder patients.
4. Bugnon Agathe, Description of molecular interaction networks involved in humoral rejection of kidney allograft.
6. De Walsche Annaïg, Multitrait GWAS to Enhance Signal Detection for a Single Target Trait
8. Durand Axelle, Characterization of genetic risk factors for proteinuria in children in the context of chronic kidney disease and recent African ancestry.
10. Ghestem Florence, Knowledge graph to dissect genotype phenotype association
12. Park Seehyun, Large-scale association analysis identified new susceptibility risk loci for differentiated thyroid carcinoma by integrating the transcriptome and proteome.
14. Martinez Jessica, Multi-trait analysis reveal new variants associated with cytokine levels in asthmatic families.
16. Morin Martin, PBMC mRNA/miRNA profiling identifies MMP9 as a putative non-invasive biomarker and therapeutic target in chronic kidney allograft rejection.
18. Rioux Bastien, Re-evaluating the association between TREX1 variants, lupus and oligoprotein interferon signatures.