FRANCE GENOMIQUE

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International Symposium on Human Genomics

May 5-7, 2025

CICSU Auditorium, 4 place Jussieu, Paris, France

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Monday May 5th, CICSU Auditorium

from 12:30	Welcoming participants, Foyer (no lunch provided)
14:00	Introductory speech, **Pascal Barbry**, France Génomique, IPMC, France ; **Jean-François Deleuze**, France Génomique, CNRGH, France ; **Emmanuelle Génin**, ITMO GGB, Inserm, University of Brest, France ; **Christian Muchardt**, ITMO GGB, INSB, CNRS, France
	Genetics and Pathologies 1/2
14:10	Instability of coding versus non-coding microsatellite sequences in mismatch repair deficient colon tumor cells: the fighting spirit, **Alex Duval**, Inserm Team “Microsatellite Instability and Cancer”, Sorbonne University, UMRS 938 – CRSA, Centre de Recherche Saint-Antoine), Hôpital Saint-Antoine, APHP (France)
14:50	Genome-wide association study of survival in sepsis patients, **Syphax Zeggane**, UFR Health Sciences Simone Veil (UVSQ), INSERM U1173, Infection and chronical inflammation lab (2i) (France)
15:10	Unravelling the molecular mechanisms causal to type 2 diabetes across global populations and disease-relevant tissues, **Ozvan Bocher**, Brest University, INSERM, EFS, UMR 1078 GGB (France)
15:30	Identification of anti-TB therapy induced ADRs genetic markers using In-Silico approaches, **Kamal Kishor**, International Institute for Population Sciences (IIPS) (India)
15:50	BREAK
	Population Genetics and Statistical Genetics 1/2
16:20	The Genome of Europe: Towards Implementing Genetic Information in Health Care and Prevention, **André Uitterlinden**, Laboratory for Population Genomics, Erasmus MC, Rotterdam (The Netherlands)
17:00	Genomic insights into the evolutionary history and metabolic risk of Polynesians, **Etienne Patin**, Human Evolutionary Genetics Unit, UMR 2000 (France)
17:20	Exploring Rare Genetic Variants in French Centenarians: A Path to Understanding Longevity, **Assia Benmehdia**, Human Genomics National Research Centre (CNRGH) (France)
17:40	A critical comparison of clustering methods in structured populations under different spatial sampling schemes, **Maël Guivarch**, Brest University, INSERM, EFS, UMR 1078 GGB (France)
18:00	Improved ancestry and admixture detection using principal component analysis of genetic data, **Florian Privé**, Aarhus University (Denmark)
18:20	Gene-environment interaction in human traits and diseases: a story of misconception, **Hugues Aschard**, Pasteur Institute (France)
18:40-20:30	Cocktail party, 24th floor, Zamansky tower (identity control upon entrance, please have valid ID)

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Tuesday May 6th, CICSU Auditorium

	Advanced omics analyses 1/2
09:00	The new biology revealed by single-molecule sequencing of the transcriptome, **Ana Conesa**, Institute for Integrative Systems Biology, Spanish National Research Council, Paterna (Spain)
09:40	DNA long-read sequencing, an interest for genetics predispositions to breast and ovarian cancer, **Crystal Renaud**, INSERM U1245, Cancer and Brain Genomics Lab, Centre François Baclesse (France)
10:00	Wastewater-based epidemiology of human viruses by nanopore sequencing, **Juejun Chen**, Institute of Molecular and Cellular Pharmacology, IPMC (France)
10:20	K-mer-based-genome-wide association studies of the gut microbiome, **Raphaël Malak**, Statistical Genetics, Department of Computational Biology, Pasteur Institute (France)
10:40	BREAK + POSTER SESSION (1/2) (ODD NUMBERS)
	Openness to Society/Science Communication
11:40	Science in times of uncertainty: investigating and communicating on the origin of the COVID-19 pandemic, **Florence Débarre**, Institute of Ecology and Environmental Sciences, IEES-Paris, UMR 7618, CNRS, Sorbonne Université, UPEC, IRD, INRAE (France)
12:20	LUNCH (Foyer)
	Genetics and Pathologies 2/2
13:40	Title TBC, **Christine Petit**, Hearing Institute, Pasteur Institute (France)
14:20	Graph neural networks reveal digenic disease candidates through biological network analysis, **Romain Nicolle**, Laboratoire de Bioinformatique Clinique, Imagine Institute (France)
14:40	VIOLA: Variant PrIOritization using Latent spAce to improve mitochondrial diseases diagnosis, **Justine Labory**, Université Côte d'Azur, Institut Sophia Agrobiotech (France)
15:00	Metanalysis of germline whole exome sequencing in 1,435 cases of testicular germ cell tumour to evaluate disruptive mutations under dominant, recessive and X-linked inheritance models, **Zeid Kuzbari**, The institute of cancer research London, University of Oxford (United Kingdom)
15:20	BREAK
	Population Genetics and Statistical Genetics 2/2
15:50	Approaches to prioritize non-coding disease risk variants, **Steven Gazal**, University of Southern California, Los Angeles (USA)
16:30	Rare variant aggregate association analysis using imputed data is a powerful approach, **Suzanne M. Leal**, Columbia University (USA)
16:50	Detecting rare recessive variants involved in multifactorial diseases: validation and power of the Fantasio method, **Sidonie Foulon**, CESP Inserm U1018, Université Paris-Saclay (France)
17:10	LDAK-PBAT: A Novel Pathway-Based Analysis Tool for Decoding the Genetics of Complex Diseases, **Takiy Berrandou**, Université Paris Cité, Paris Cardiovascular Research Center, Inserm (France)
17:30	rcRS algorithm: Incorporating complex genetic model into risk estimation, **Fabien Laporte**, Nantes Université, CNRS, Inserm, Thorax Institute (France)
	Immunogenetics
17:50	TBC
18:30	IMGT® Population Analysis of the Human IGH Locus: Unveiling Novel Polymorphisms and Copy Number Variations Across Diverse Genome assemblies, **Ariadni Papadaki**, **Maria Georga,** IMGT, Institute of Human Genetics, IGH (France)

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Wednesday May 7th, CICSU Auditorium

	Advanced omics analyses 2/2
09:00	Combinatorial DNA-Pools targeted-sequencing as a robust cost-effective method to detect rare variants: analysis strategy and application to dilated cardiomyopathy genetic diagnosis, **Claire Perret**, INSERM UMRS1166, ICAN (France)
09:20	Long-Read RNA sequencing in cardiomyopathies: a new approach for genetic diagnostic with strong potential?, **Laëtitia Rialland**, INSERM UMRS1166, Medical school, Pitié-Salpetrière Hospital (France)
09:40	Innovative insights on the genetic architecture of the human plasma proteome through meta-analysis of English and Italian protein Quantitative Traits Loci studies, **Solène Cadiou**, Human Technopole (Italy)
10:00	Lifting the veil on Challenging Medically Relevant Genes, **Victor Grentzinger**, GIGA Human Genetics, Liège University Hospital Center (Belgium)
10:20	BREAK + POSTER SESSION (2/2) (EVEN NUMBERS)
	Epigenetics / Regulome
11:20	Searching for biologically consequential and inconsequential miRNA/target interactions using the evolutionary history of vertebrate miRNA genes, **Hervé Seitz**, Institute of Human Genetics, Montpellier GenomiX, IGH (France)
12:00	Impaired RNA Polymerase II Elongation Reveals Novel Molecular Mechanisms in Multiple Sclerosis, **Christian Muchardt**, IBPS, Sorbonne Université, CNRS Biologie (France)
12:20	Identifying causal cell types for human diseases and risk variants from candidate regulatory elements, **Artem Kim**, University of Southern California (USA)
12:40	LUNCH (Foyer)
	Single Cell/Spatial Transcriptomics
14:00	Multi-modal learning methods for single-cell data integration, **Laura Cantini**, Pasteur Institute (France)
14:40	pyROMA, a python software for representation and quantification of module activity from single cell and bulk transcriptomic data, **Altynbek Zhubanchaliyev**, Computational Systems Biology group, Learning Planet Institute, Université Paris-Saclay, CEA, CNRGH (France)
15:00	Imagine the Medicine of the Future Now, **Mickaël Ménager**, Imagine Institute (France)
15:40	Early COPD single-cell and spatial transcriptomics, **Morgane Fierville**, Institute of Molecular and Cellular Pharmacology, IPMC (France)
16:00	Single-nucleus transcriptomic analysis of ageing in the mouse lemur prefrontal cortex, **Clémence Su, Eric Bonnet,** Human Genomics National Research Centre (CNRGH) (France)
16:20	Conclusions, Best poster award ceremony