aviesan

ITMO Genetics, genomics and bioinformatics

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The Institute deals with genetics and genomics of living organisms comprising viruses, microorganisms, plants, animals and humans. The research activities foster the organization, evolution, genome variability, regulation of gene expression, population genetics as well as genetic and epigenetic determinism of diseases and treatment of genetic diseases.

Our missions

Accompany the development of the national strategy in the domain

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- Describe the state-of-the-art of existing research capacities by topic
- Define the main scientific challenges and contribute to the national research strategy
- Support conceptual and technological evolutions with coordination actions
- Introduce the organisation of the French research to international stakeholders

Coordinate actions of different research stakeholders

- Coordinate the contribution to the research programming for ANR (French National Research Agency) and at European level.
- Coordinate the development of NGS (Next Generation Sequencing) platforms, bioinformatic analyses, storage and archiving of NGS and big data at national level
- Foster interdisciplinary approaches between biology, bioinformatics and statistics

Propose and animate the activities

- Visits of research centers and strategic support
- Organisation and support of meetings and thematic workshops
- Communication to general public about scientific, medical and societal impacts of technological advances



Metaphase of mouse chromosomes, with X chromosome in green and repeated LINE sequences in red. Jennifer Chow and Édith Heard, © Institut Curie, Paris

Main scientific and medical challenges

- Characterization of genome, transcriptome and gene/ epigene expression regulation in different species
- Comparison of genomes of different species to better understand evolution and biodiversity mechanisms, and translate to novel approaches for genetic diseases
- Bioinformatics, statistic and biological analysis of simple or unicellular organisms with short life-cycle
- Fostering of interdisciplinary approaches for in silico analysis to model (patho)physiological pathways
- Characterization of genome-phenome and genomeenvironment interactions
- Development of various therapeutic strategies for genetic diseases, based on gene transfer, cell therapy, drug repositioning and therapies targeting pathophysiological mechanisms

The scientific experts

ITMO Directors

Frédéric BOCCARD (CNRS) Catherine NGUYEN (Inserm)

Scientific policy Officer: Françoise PULCINI (Inserm) Administrative assistant: Maryne ANGIBOUST (Inserm)

Assistant director and scientific coordinator of the European Joint Program on Rare Diseases (EJP RD): Daria JULKOWSKA (Inserm) Senior Project Managers: Galliano ZANELLO (Inserm): Immunology - Carla D'ANGELO (Inserm): Genetics -Juliane HALFTERMEYER (Inserm Transfert): Genetics and Oncology Project Assistant: Katerina TZIMA (Inserm) Communication: François URBAIN (Inserm)

Contact Aviesan ITMO GGB: iggb@aviesan.fr

Involvements in national and international programs

- **PNMR3** (3rd National Plan for Rare Diseases), aiming at innovation, diagnosis and treatment for all
- PFMG 2025 (*Plan France médecine génomique* 2025) missioned to investigate establishment of access to genetic diagnosis in France together with a prospective review covering biological and medical research
- **EJP RD** (European joint program on Rare Diseases) launched in January 2019 for five years, under the leadership of INSERM and coordinated by the ITMO GGB. It brings together 130 organisations from 34 countries. The main goal of the EJP RD is to improve integration, efficacy, production and social impact of research on rare diseases through the development, demonstration and promotion of sharing of research and clinical data
- HORIZON 2020. Through research and innovation projects funded by HORIZON 2020 (The EU Framework Program for Research and Innovation.

Main local actions

- Since 2012, organization of the annual bioinformatics workshop of Aviesan - IFB: "Introduction to the processing of genomics data obtained by high-throughput sequencing"
- Since 2016, support and funding of 18 scientific meetings in the field of genetics, genomics, epigenetics, or genetic determinism of human diseases and bioinformatics
- Series of conferences in 2018:
- "Advances in the Epigenetics of Immunity and Infection", March Paris.
- "Challenges and Perspectives in integrative Bioinformatics", September Paris.

Experts committee

- Laurent ABEL (Inserm, Paris)
- Serge AMSELEM (Inserm, Paris)
- Anaïs BAUDOT (Université Aix-Marseille)
- Christophe BEROUD (Inserm, Marseille)
- Jamel CHELLY (Université, Illkirch)
- Mark COCK (CNRS, Roscoff)
- Jean-François DELEUZE (CEA, Évry)
- Bernard DE MASSY (CNRS, Montpellier)
- **Emmanuelle GENIN (Inserm, Brest)**
- Philippe GLASER (Institut Pasteur, Paris)
- Marc HANAUER (Inserm, Paris)
- Édith HEARD (CNRS, Paris)
- Cécile JULIER (Inserm, Paris)
- Stanislas LYONNET (Inserm, Paris)
- Claudine MÉDIGUE (CNRS, Évry)
- Hadi QUESNEVILLE (INRA, Versailles)
- Lluis QUINTANA-MURCI (Institut Pasteur, Paris)
- Ana RATH (Inserm, Paris)
- Hugues ROEST-CROLLIUS (CNRS, Paris)
- Élisabeth TOURNIER-LASSERVE (Inserm, Paris)
- Hélène TOUZET (CNRS, Lille)
- Chantal VAURY (CNRS, Clermont-Ferrand)
- Jacques van HELDEN (Université Aix-Marseille)
- Jonathan WEITZMAN (Université Aix-Marseille)
- Michel WERNER (CEA, Paris)



Multi-scale rewiring of 3D chromatin architecture during mouse brain development.

A Highly spliced genes engage in chromatin contacts across long-range genomic distances. B. The interaction strength between pairs of gene promoters is correlated with their expression levels. C. Dynamic changes in morphology, global 3D genome organization, compartmentalization and regulatory chromatin loops occur during neural differentiation.

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