

# nTAiDA<sup>®</sup> Workshops

*« New Technologies  
for Autoimmune/Inflammatory  
Disease Analysis »*

Workshop 1  
Sponsored by Roche Diagnostics



**i2B** INFLAMMATION  
IMMUNOPATHOLOGY  
BIOTHERAPY  
HOSPITAL UNIVERSITY DEPARTMENT - DHU

# Workshop 1: Transcriptome and Exome

23 Janvier 2015 9h00 - 17h00  
105 Boulevard de l'Hôpital, salle de conférence 6<sup>ème</sup> étage

09.00 Welcome coffee

09.30 **Evolution towards New Technologies in Genomics and Transcriptomics: from Roche-NimbleGen Microarrays to Sequencing.**

*Charleen Courtois*

10.20 **Somatic mutation detection by whole Exome sequencing in non-cancer patients.**

*Laurent Mesnard*

11.10 Coffee break

11.30 **Molecular basis of Mendelian disorders by means of whole genome sequencing and homozygosity mapping.**

*Pr Serge Amselem*

12.20 Lunch

13.30 **Early dendritic cell molecular signature modelling predicts the quality of vaccine-induced adaptive T-cell responses.**

*Nicolas Dérian*

14.20 **Analysis of repeated measurements of gene expression data. Application to an HIV vaccine trial.**

*Rodolphe Thiebaut*

15.10 Coffee break

15.30 **Identification of long non-coding RNAs (lncRNAs) using RNASeq in dogs.**

*Thomas Derrien*

16.20 **General discussion**

17.00 End

# Abstracts

**Evolution towards New Technologies in Genomics and Transcriptomics: from Roche-NimbleGen Microarrays to Sequencing.** The field of genomics and transcriptomics is rapidly evolving in terms of resolution, quality and data throughput. Over the past 5 years, Roche-NimbleGen was able to develop from microarray-based technology to much higher resolution and information throughput with its Sequence Capture technology. Roche-NimbleGen offers now a sensitive, accurate and reproducible detection of variants through genomic, epigenetic and transcriptomic sequence capture solutions, for the majority of sequencing platforms on the market, offering as well high design flexibility for researchers and clinicians. By focusing on sequence capture, Roche-NimbleGen was able to set a new vision on how to decipher genome, epigenome and transcriptome based on the need for targeted and customized research projects.

**Somatic mutation detection by whole Exome sequencing in non-cancer patients.** Whole Exome sequencing (WES) is largely used nowadays for the detection of germinal mutations involving mendelian diseases. Another popular application of WES aims to evidence somatic mutations in cancer patients in which tumor and non tumor DNA samples have been compared. Very few bioinformatic tools are available for the detection of somatic mutations in non-tumoral samples. We exemplified here the research of somatic de novo mutation or mosaicism and their analysis by a dedicated next generation sequencing pipeline (gobyWebSomaticMod <http://campagnelab.org/software/gobyweb/> for details) freely available. [Pitfalls and technical problems associated with the analysis of non tumoral somatic mutations or mosaicism will be discussed and exemplified.](#)

**Molecular basis of Mendelian disorders by means of whole genome sequencing and homozygosity mapping.** Over the past few years, WES has become an efficient means of searching for new molecular defects, especially in recessive diseases. However, it could be quite challenging to point out causative mutations among an average of about 40,000 variants per genome. To improve the efficiency of WES by reducing the number of candidate variants, it is possible to combine this global approach with homozygosity mapping in selected affected individuals born to consanguineous unions. By presenting data obtained on the molecular basis of a rare respiratory disorder, we will illustrate the importance of cautious analysis of sequence variations identified by WES, in order to circumvent possible exome pitfalls.

**Early dendritic cell molecular signature modelling predicts the quality of vaccine-induced adaptive T-cell responses.** High-throughput experimental methods and information technologies have much improved our ability to decipher complex interactions, including in vaccinology. We analysed mouse dendritic cell transcriptome 6 hours after vaccination and produced a *random forest* model to predict antigen-specific T-cell expansion. The model successfully predicted the classification of a new set of vectors, based on spleen dendritic cell, but also peripheral blood mononuclear cell transcriptome. The same model also accurately predicted immune responses from human datasets from the literature.

**Identification of long non-coding RNAs (lncRNAs) using RNASeq in dogs.** Whole transcriptome sequencing technology (RNASeq) has become a standard for transcriptome analysis and allows to catalogue the RNA populations of a given cell line or a tissue at different time points and conditions. Amongst all RNAs, the class of the long non-coding RNAs (lncRNAs) is now emerging as a major component of the non-coding transcriptome but annotating and classifying lncRNAs using RNASeq experiments remains a challenging task. Therefore, we developed FEELnc a workflow for extracting lncRNAs based on a set of RNASeq assembled transcripts as input. To demonstrate the usage of FEELnc, we applied it on canine RNASeq datasets and built a comprehensive catalogue of ~15,000 lncRNAs.

**Analysis of repeated measurements of gene expression data. Application to an HIV vaccine trial.** We present a new method developed to analyse repeated measurements of gene expression data using a gene set approach. The application of the method to the data of an HIV vaccine trial revealed subtle changes of gene expression that were associated with the immune response.

# Upcoming Program

## **Mars 2015**

- \* Workshop 2: High-throughput data, microbiome and next-generation sequencing

## **May 2015**

- \* Workshop 3: Lab Info and Management Systems (LIMS), Database and Big Data

## **September 2015**

- \* Workshop 4: Petscan, Mass spectrometry, Metabolome

## **Novembre 2015**

- \* Workshop 5: Mathematical, statistical modelling and data analysis