From Gene-Targeted Tests to Genome-Wide Data in Cancer Genetics: A Paradigm for the Development Personalised Genomic Medicine?

First workshop

Quality and accuracy of genetic data and knowledge produced by genome-wide strategies

Amphithéatre Marie Curie – Institut Curie
1, rue Pierre et Marie Curie
Paris, June 6-7, 2013

Personalised genomic medicine has little chance of becoming reality if the data and knowledge on which it is based are not reliable. The quality of the data produced by genome-wide strategies, as well as of the science that underlies this activity, is therefore an important preliminary question to providing any form of testing.

Sequencing methods each have their own limitations that need to be specified and taken into consideration when interpreting data. However, it is difficult to determine to what extent differences in interpretation are due to differences in methods or to disagreements in the interpretation. Moreover, all methods produce data that may have no known scientific or clinical validity. How, therefore, does one ensure that the data generated by genome-wide strategies are scientifically valid and clinically useful, and that the clinical utility of data with initially unknown validity is regularly updated? Should complete genomic data be made available for clinical use or should options of analyses be discussed among professionals and with patients, in view of setting standards of practice in clinical settings?

The objective of this workshop is to explore both the epistemological issues raised by big data production and the present practices and potential uses of genome-wide strategies in research and clinical settings, with an emphasis on its relevance for cancer genetics.

Workshop Coordinators:

Simone Bateman, *Senior Researcher, Sociology, CERMES3 (CNRS/University Paris Descartes/EHESS/INSERM)*

Dominique Stoppa-Lyonnet, *Professor of Medical Genetics, Head of the Service of Cancer Genetics (Institut Curie/Hospital and University Paris Descartes)*

Thursday June 6, 2013

9h30h – 10h30 Welcome and Opening Session: On the SIRIC Project and the Five Workshops

Dominique Stoppa-Lyonnet and **Simone Bateman**

10h30- 12h Can high-throughput sequencing produce reliable data? Identifying crucial phases and potential pitfalls of big data production

Ivo Gut, Director of the National Centre for Genomic Analysis, Barcelona (Spain)

Discussion introduced by: **Thomas Rio Frio**, Manager - Next Generation Sequencing platform, Institut Curie

Lunch

14h- 16H30 Why use high throughput sequencing? Examining the contexts and purposes of big-data production

Uses of genome-wide data in population genomics: from natural selection to human immunology

Luis Quintana-Murci : Senior Researcher CNRS, Director of CNRS Human Evolutionary Genetics Research Unit, Institut Pasteur (France)

Discussion introduced by: **Françoise Clerget-Darpoux**, Emeritus Senior Researcher INSERM, Genetics and embryology of congenital malformation, Federative Research Institute Necker Enfants Malades, Paris

Using genome-wide data from Mendelian to complex genetics: the example of infectious diseases.

Laurent Abel, Senior Researcher INSERM, Directeur of U980 – Human Genetics of Infectious Diseases, Federative Research Institute Necker Enfants Malades, Paris

Discussion introduced by: **Dominique Stoppa-Lyonnet**, Professor of Medical Genetics, Institut Curie, University Paris Descartes

Coffee break

17h - 18h30 Data Intensive Science: a new or a renewed paradigm for science?

Bruno Strasser, Professor of Science Education and History of Science, Institute of Genetics and Genomics in Geneva, University of Genève

Discussion introduced by: **Jean-Paul Gaudillière,** Senior Researcher INSERM, Director of CERMES3 (INSERM/University Paris Descartes/EHESS/CNRS)

Friday June 7, 2013

9h - 10h30 Understanding the science behind big-data production: epistemological assumptions

Sabina Leonelli, Senior Lecturer, Department of Sociology, Philosophy and Anthropology, University of Exeter, (UK)

Discussion introduced by: **Soraya de Chadarevian**, Professor, Department of History and Institute for Society and Genetics, University of California, Los Angeles (USA)

Coffee break

11h -12h30 Making sense of the data: transforming DNA sequences into analytically valid and clinically useful information

Catherine Bourgain, Researcher INSERM, Genetic Epidemiology, CERMES3 (INSERM/University Paris Descartes/EHESS/CNRS)

Discussion introduced by: **Jonathan Weitzman**, Professor of Genetics, Centre Epigenetics and Cell Fate (University Paris Diderot/CNRS)

Lunch

14h – 15h30 Putting the person in personalized medicine: historical perspectives on individuality and health

Nathaniel Comfort Associate Professor, Institute of the History of Medicine, Johns Hopkins University (USA)

Discussion introduced by: **Ilana Löwy,** Senior Researcher, History of Science, CERMES3 (INSERM/University Paris Descartes/EHESS/CNRS)

15h30 - 16h15 Overview and Concluding Remarks

Jean Gayon, Professor of History and Philosophy of Science, University Paris 1-Panthéon Sorbonne

Simone Bateman and **Dominique Stoppa-Lyonnet**