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

Third workshop

Genomic Data: Challenges to the Concept and Practice of Genetic Counselling

Amphithéâtre Marie Curie – Institut Curie
1, rue Pierre et Marie Curie
75005 Paris

January 8-9, 2015

Recent developments in genomics and bioinformatics are making it possible to offer genetic counseling and testing for an ever wider variety of medical conditions, in a variety of settings, to an ever greater number of individuals. Genetic counselling was a practice initially set up and organized around the provision of relevant information to persons potentially concerned by familial disorders; from country to country, the practice has – to a greater or lesser extent – its roots in the eugenic movement of the early 20th century. In many countries today, genetic counselling and genetic screening and testing is provided exclusively by physicians qualified in medical genetics to patients with a genetic or hereditary disease or persons at risk of developing such a disease (potential patients). Some countries also employ professionals who are not physicians, but who often work with them and are specially trained to counsel what they prefer to call "clients", that is persons who may or may not be ill, on genetic-related conditions that affect or may affect them and their family. Advances in genome sequencing have generated new expectations in both clinical and research settings, but have also given rise to new commercial settings that offer what is usually referred to as direct-to-consumer testing (DTC) and advocate the right of individuals to purchase information concerning their personal genome, even without an immediate medical reason to do so. Not all countries allow DTC testing and in those countries where it has developed, there has recently been closer scrutiny as to the way in which genomic information is being provided to the consumer. Next generation sequencing (NGS) techniques are also being used in research settings in which the subjects may be patients or healthy volunteers.

Workshop Coordinators:

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

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

What are the specific benefits and what are the problems raised by each of these settings? Are there any good reasons – scientific, practical, technical or economic – for restricting genome sequencing exclusively to individuals consulting a physician for an identified hereditary disorder? Conversely, if such testing were to respond adequately to present concerns about the validity of the data and the quality of counselling provided to tested individuals in non-clinical settings, would there eventually be any advantages to providing genome sequencing outside of a clinical framework? As genome sequencing becomes available to an increasing number of people in varied settings (research, clinical, commercial) for different purposes (health prevention, treatment management, and reproductive decisions), the question can be raised as to who should provide genetic counselling and even as to whether health professionals other than clinical geneticists and genetic counsellors, especially primary care physicians, are appropriately trained to meet the increasing demand for genetics-related information.

Indeed, how to ensure the quality of the counselling provided to tested individuals before and after genetic testing is not a new issue: many of the aspects related to information, consent to testing, appropriate explanation of test results and patient counselling have been explored over the years. However, the advent of novel, more powerful sequencing techniques raises new questions in this area that need further investigation. Should high throughput genome sequencing be used to generate as much data as possible or should the generation of data be restricted to potentially relevant regions of the genome? How does one ensure that consent to this complex form of testing is appropriately informed? What information should be communicated to the individual after testing: only scientifically valid and clinically useful data or all available scientific data? What in particular should be said about data that has no immediate validity or utility but that may, with time, acquire medical significance? What are the challenges of explaining that sequencing the whole genome or even just the exome might yield incidental findings whose relevance to the person's state of health may sometimes be unknown? Does an individual have a right to genetic information, including information that may have no clinical validity or utility?

The objective of this workshop is to explore the challenges that genome sequencing represents for genetic counselling, screening and testing. The plethora of data potentially available through genome sequencing and the novel settings in which sequencing is provided require a reassessment of the concept of genetic counselling, of the norms that have so far guided its practice and governed decision-making about screening and testing, and of the challenges that such data represent for the management of individual and population health care.

PROGRAMME

Thursday January 8, 2015

8:30 Welcome Coffee

8:45 Introduction to the Third Workshop

Dominique Stoppa-Lyonnet and **Simone Bateman** (workshop coordinators)

9:00 – 13:00 History, diversity and transformation of genetic counselling as a professional practice: a comparative perspective

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

9:00- 9:30 United States:

Alexandra Minna Stern, Professor of History of Medicine**,** Center for Social Epidemiology and Population Health, University of Michigan (USA)

9:30-10:00 France:

Claire Julian-Reynier, Senior Researcher INSERM, UMR912 - SE4S (INSERM/ IRD), Université Aix-Marseille

10:00-10:30 United Kingdom:

Paul Atkinson, Distinguished Research Professor, Cardiff School of Social Sciences, Cardiff University (UK)

10:30 -11:00 - Coffee break

11:00-11:30 Oman:

Claire Beaudevin, Researcher CNRS, Anthropology, CERMES3 (CNRS/University Paris Descartes/EHESS/INSERM)

11:30-12:00 Israël:

Aviad Raz, Professor, Department of Sociology and Anthropology, Ben Gurion University (Israël)

12:00-13:00

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

13h – 14:30 Lunch

14:30 – 18:45 Generating Genomic Data in New Settings: In Which Ways and To What Purpose?

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

14:30 – 15:45 Clinical Setting: Diagnosis, Prevention and Treatment

14:30 – 15:00

Robert Green, Associate Professor of Medicine, Division of Genetics and Department of Medicine at Brigham and Women's Hospital and Harvard Medical School (USA)

15:00 – 15:45

Discussion introduced by **Jenny Hewison,** Professor of Psychology of Healthcare, Leeds Institute of Health Sciences, University of Leeds (UK)

15:45– 17:00 Research on Collections of Patient Biospecimens

15:45 – 16:15

Olivier Delattre, Director of INSERM Research Unit on Genetics and Biology of Cancers, Institut Curie

16:15 – 17:00

Discussion introduced by **Anne Laure Morin,** Lawyer, A.L.M. Avocats, Paris, France

17:00-17:30 Coffee Break

17:30 – 18:45 Direct-to-Consumer Testing: Personal Genomics and Health prevention

17:30 – 18:00

Anne Morriss, Founder and Chief Executive Officer of ***Genepeeks*** (USA)

18:00 – 18:45

Discussion introduced by: **Barbara Prainsack,** Department of Social Science, Health & Medicine, King's College London (UK)

Friday January 9, 2015

9:15 – 11:30 ***Making Choices about Seeking and Providing Information***

Round Table Discussion*: *Is there a right to know, a duty to know, a right not to know? Who Decides?

Introduced and chaired by: **Simone Bateman,** Emeritus Senior Researcher CNRS, Sociology, CERMES3 (CNRS/University Paris Descartes/EHESS/INSERM)

9:30 – 10:30 Three short statements:

Robert Green, Concerning the *ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing* and subsequent debate

Dominique Stoppa-Lyonnet, Concerning a Recent Opinion by the French National Ethics Committee on *Ethical Issues in Connection with the Development of Foetal Genetic Testing on Maternal Blood*

Christel Nourissier, Eurordis – Rare Diseases Europe. *A patient perspective on seeking and providing information*

10:30 – 11:30 Debate with other speakers and audience

11:30 – 12:00 Summary and Concluding Remarks: Simone Bateman and **Dominique Stoppa-Lyonnet**