Gothenburg, June 11, 2010

Changing landscape of genetic testing and its impact on clinical and laboratory services and research in Europe

Talk 1. An overview of the new technologies (array CGH, whole genome sequencing, expression arrays, multifactoral diagnostic services)
Speaker Gert Jan van Ommen
20 min talk: An overview of the different technologies, including application, resolution, limitations. Whether the techniques are currently research or diagnostic and validated diagnostically.

Talk 2. Practical challenges for clinical genetic services
Speaker Dian Donnai
20 min talk: A summary of the issues that clinical genetics service face with the introduction of these high resolution technologies. Practical issues from a clinical geneticist perspective including interpretation of results in relation to the clinical findings or perspective health, wealth of information obtained and how much information must be given to the patients etc

Talk 3. Practical challenges that copy number variation and whole genome sequencing creates for genetic diagnostic labs
Speaker Joris Vermeesch
20 min talk: A summary of the problems of identifying normal variants and clinically significant genomic changes. The interpretation of the significance results for the clinician; minimum platform resolution; whether there is an application for targeted arrays; whether all referral categories should be tested etc.

Talk 4 Ethical and policy challenges in genetic screening
Speaker Lainie Friedman Ross
15 min talk: High throughput technologies are rapidly changing the field of screening. What are the ethical and policy challenges for large scale applications in healthy individuals, for instance in newborn screening. Overview of the various implications for genetic screening and the blurring of the boundaries between a screening and diagnostic test with these new technologies.

Talk 5 The role of professional geneticists in the changing landscape
Speaker Jean Jacques Cassiman
15 min talk: An overview of the role of clinical, scientific and laboratory staff in the development and diagnostic use of these new technologies. Who interprets the array results? What is the role of the laboratory and the role of the clinician given the wealth of information this new technology will produce. Does the laboratory interpret the significance of the results and which genes are of relevant to the clinical referral and the clinical geneticist the clinical implications of this result for patient and family?

Talk 6 Building the evidence of unknown variants
Speaker Martijn Breuning
15 min talk: Laboratories and clinicians are facing the challenge of unsought findings. Some results may even concern unknown variants, so that it is hard to judge what needs to be reported back. How should the evidence of unknown variants be built; can common databases be developed?

Talk 7 Clinical Utility
Speaker Peter Farndon
15 min talk: What is Clinical Utility? How do we define the clinical utility of a test? Should genetic tests be offered because they are available? Multifactorial disease testing is it applicable? What about the patient interests? Private vs public testing; what parameters should be decided before a genetic test is offered.

Talk 8 Patient perspective - new healthcare offers; appropriate testing
Speaker Alistair Kent
15 min talk: This technology has the potential to detect every genetic disease even before symptoms occur. What does the patient expect and/or need? What are the risks of giving too much information or not enough? What protection should there be for the patient? What should the healthcare system offer?

Talk 9 Ethical/legal issues in whole genome diagnostics
Speaker Timothy Caulfield
15 min talk: What are the ethical and legal issues? Who owns the whole genome information and who decides what is relevant for the patient to know. Patient confidentiality, informed consent, giving information that may not be relevant. Presymptomatic testing. Follow up of family members. Paternity issues.

Talk 10 Governance issues in whole genome diagnostics
Speaker Ellen Wright Clayton
15 min talk: How do healthcare providers decide the level of service offered to patients? Who decides what is useful? Who decides what is reported back to patients? Are patients allowed access to their data? The technology is in the public domain; how do you prevent abuse of the technology - inappropriate test results offered to patients (e.g. multifactorial). What governance structures need to be put in place?

Workshop I

Is it morally acceptable to examine the whole genome? What about blurring the boundaries between diagnostics and screening?

Chair: prof. Guido de Wert

Report: Niels Nijsingh

This workshop will focus on the normative implications of the introduction of whole genome sequencing (WGS) in public health care. The practice in clinical genetics used to be indication-based genetic testing such that only a specific question was investigated. WGS generates a wealth of personal genomic data/information that could potentially be used for various health care purposes (screening, diagnosis, personalized medicine). Providing patients with predictive health information not necessarily related to whatever indication they might have, could amount to a form of (population) screening. Thus the distinction between diagnosis and screening is blurred. Will the normative framework for screening also become relevant for WGS? WGS challenges the notion of 'informed consent'. When is it acceptable to offer a test that almost by definition generates unexpected results?

Workshop II

When should the whole genome be examined?

Chair: Brian Fowler, David Barton & Ros Hastings

This workshop will discuss the challenges of WGS from a laboratory perspective. New syndromes/genetic diseases as well as normal variants of no clinical significance will be identified through WGS. The identification and interpretation of 'normal variants', together with the increased complexity of cases, will require that all genetic laboratories interpret the clinical significance of the data obtained. There is a need to discuss how best to re-structure the laboratory services logistically, as well as determine and interpret the data/information made available to clinicians so that patients can receive appropriate advice and genetic testing. Should all genetic referrals be offered WGS or are there exceptions where a targeted approach to WGS is more appropriate e.g. prenatal testing? Are there referrals where WGS is not the first-line test to use? Should genetic reports contain all the genomic information including diseases/disorders that the patient is at low risk for which there is no medical or family history? As the Cytogenetic and Molecular Genetic service technologies are now merging closer, perhaps now is the time to provide an integrated lab service? Will there still be a need for specialist discipline training?

Workshop III

How do we build the evidence of clinical utility?

Chairs: prof. Jörg Schmidtke & prof. Michael Krawczak

Whole genome sequencing (WGS) generates personal genomic data/information that could potentially be used for many health care purposes (diagnosis, prediction, prevention, personalized medicine). Generally speaking, information that will contribute to a longer and healthier life should be reported back to patients, but how do we build the evidence of clinical utility for all genetic variants? Regulation of new test or publicly accessible quality indicators may help guide both public and professionals in differentiating between useful, useless or even harmful tests. However, who can or should decide on evidence thresholds remains as yet an unresolved issue.

Afternoon Workshop IV:

How do we prepare our primary health care service to incorporate whole genome testing?

Chairs: prof. Martina Cornel, Eric Vermeulen & Carla van El

Until today, incorporating currently available genomics knowledge and testing in primary care has proven to be a difficult task. The introduction of new techniques, such as whole genome testing, promises to be even more challenging. In this workshop we will reflect on possibilities, prerequisites and barriers for incorporating new techniques and testing. Different models for integrating genetic medicine into primary care have been proposed, depending on the organization of health care and type of services offered. The partnership between medical genetics and primary care needs creative rethinking and fortification. Overall, primary health care professionals have insufficient knowledge about genetics. Training and facilitating easily accessible sources of information is paramount. Websites, knowledge centers, ambulatory genetic counselors or prevention consultants regularly visiting primary care units may be helpful innovations. The availability of direct-to-consumer genetic testing may lead to unnecessary use of scarce health care resources because of the need to help interpret test results..
This workshop aims to provide learning through focused discussion on these elements among participants with knowledge of different health care structures. The goal is to explore necessary and feasible adaptations and an agenda for future initiatives.

Workshop V: How do we prepare our genetic services to incorporate whole genome testing?

Chairs: prof. Bert Bakker & prof. Joris Vermeesch

This workshop starts from clinical genetic services, which have to rethink their role in health care structures. Whole genome sequencing (WGS) will generate data/information that could potentially be used for other health care purposes (screening, diagnosis, personalized medicine). Providing patients with predictive health information not necessarily related to whatever indication they might have, would amount to a form of (population) screening. Are clinical geneticists prepared to discuss screening results? Will they have to collaborate with public health services? How can they give advice on life style and/or pharmacogenetics? If genetic laboratories interpret the raw data, are clinical geneticists prepared to discuss all the wider implications of the results to the patients?

Workshop VI: Blurring boundaries between research and clinical services

Chairs: dr. Pascal Borry & Yrrah Stol

Whole genome sequencing (WGS) will generate data/information that could potentially be used for other health care purposes (screening, diagnosis, personalized medicine). Providing patients with predictive health information not necessarily related to whatever indication they might have would raise similar issues as biobanks. The increase in the collection, storage and use of genetic data has led to a number of questions related to issues concerning anonymity, privacy, confidentiality, ownership and informed consent. To what extent can anonymity, privacy and confidentiality still be guaranteed? To what extent are current informed consent procedures sufficient to define what researchers can and cannot do with the material and data, while also enabling more or less unknown applications and uses of this data? How can guidelines for re-contacting and informing patients of unexpected results be developed?