Stepping Stones towards the Genetics Clinic of the Future

Summary of a CGD-CSG workshop[1]
16-17 January 2014, Club de la Fondation Universitaire, Brussels

An interdisciplinary group of experts gathered in Brussels on 16-17 January 2014 to discuss stepping stones towards the genetics clinic of the future. The main objective of the workshop was to identify field labs that address controversial, interdisciplinary challenges to clinical implementation of next-generation sequencing.

The workshop participants discussed challenges within five areas of the genetics clinic of the future: the living room, the doctor's office, the laboratory, the server room and the main entrance. After introductory presentations to each area, participants prioritised challenges within and across the different areas. Rather than attempting to provide a single comprehensive answer to the varied and complex challenges surrounding the genetics clinic of the future, the workshop participants suggested a stepping stone approach. Suggested field labs included the use of a mobile device as a diagnostic support tool, mapping the roles and responsibilities of the next-generation clinical geneticist, defining opportunities to 'embed' ethicists and legal experts in the laboratory, drafting a survey to map levels of knowledge and awareness on data sharing, and defining more meaningful ways to concretely involve patients in research.

A visual map representing the workshop findings will become available soon. Additionally, the workshop participants will issue a joint statement that discusses the stepping stone approach in further detail and offers concrete proposals for interdisciplinary collaboration.

Addressing controversial, interdisciplinary challenges to the genetic clinic of the future - such as the establishment of appropriate data sharing policies, data protection regulation, data storage in electronic patient records and regulating beyond-the-clinic genetics - requires focused, constructive interactions that take into account the interrelatedness of (partial) solutions: researchers from the natural and social sciences, medical professionals, policy makers, patients and citizens have to engage in dialogue on the design principles of data infrastructures as genomic technologies mature and become embedded in routine diagnostic procedures.

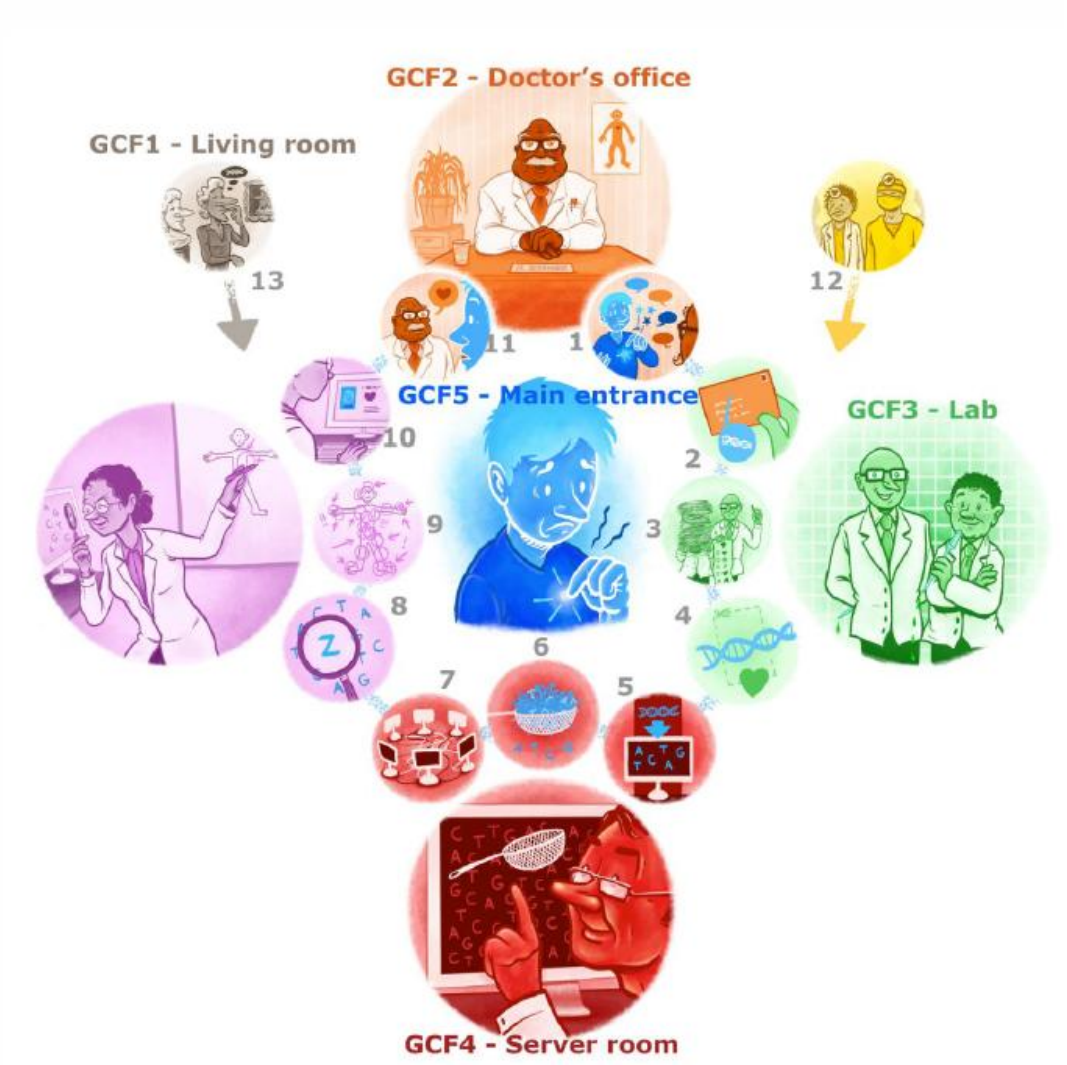
[1] The workshop programme was based on the findings of two Dutch projects, CARDIO and ReDaPeD, funded by the Netherlands Genomics Initiative (NGI) and carried out by the Centre for Genome Diagnostics (CGD) and the CSG Centre for Society and the Life Sciences in collaboration with the Responsible Innovation Collective (RIC). Both projects aimed to map the potential impact of NGS on the future of clinical genetics. Reports are available at: http://www.society-lifesciences.nl/en/projects/dna-data-information/project/artikel/more-data-more-issues.html

Centre for Genome Diagnostics

CSG CENTRE FOR SOCIETY AND THE LIFE SCIENCES

De Proeffabriek

Envisioning the Genetics Clinic of the Future



The 'diagnostic cycle' (Figure 1) represents current clinical genetics practice: obtaining a genetic diagnosis generally goes from genetic consult (orange) to DNA sequencing (green) to data analysis (red), to interpretation of results (purple) and post-test consult (orange). The entire process currently occurs within specialised clinical genetic centres, but some aspects may change as a result of the introduction of NGS (Figure 1; 1-13). We highlight five areas within the diagnostic cycle where NGS can be expected to have a significant impact. First, there may be discussions and information gathering beyond the traditional clinical setting (the 'living room'), where aspects like management of own health information (10-13) become increasingly relevant. Second, the role and position of patients that enter the clinical setting (the 'main entrance') may change, involving similar aspects (10-13). Third, information exchange in the doctor's office may change, including aspects like informed consent (1) and counselling (11). Fourth, some processes may change in the laboratory, such as reception and handling of requests (2-4) and interpretation of results (8-10). Finally, the 'server room' will become an increasingly relevant element of the genetics clinic, encompassing aspects like data analysis (5), storage (6) and exchange (7).

Workshop programme

Thursday 16 January 2014

11:00 Introduction: Welcome to the genetic clinic of the future & Tour de table
Dr Terry Vrijenhoek, University Medical Centre Utrecht, NL &
Dr Daan Schuurbiers, Responsible Innovation Collective, NL

12:00 GCF Element 1 - Genomics in The living Room: Beyond-the-Clinic genetics
Dr Barbara Prainsack, Department of Social Science, Health &Medicine, King's College London, UK

12:45 GCF Element 2 - The Doctor's Office
Prof Nine Knoers, University Medical Centre Utrecht, NL

13:30 Lunch

15:00 GCF Element 3 - The Lab
Prof Xavier Estivill, Centre for Genomic Regulation, ES

15:45 GCF Element 4 - The Server Room
Prof Anthony Brookes, University of Leicester, UK

16:30 Introduction to casework: building the genetics clinic of the future? Exploring opportunities and challenges

19.30 Dinner at Cospaia, Capitaine Crespel 1, Brussels

Friday 17 January

09.00 Welcome and introduction to day 2
09:15 GCF Element 5 – The Main Entrance
Cor Oosterwijk, Secretary General, European Genetic Alliances Network, NL

10:00 Funding opportunities in H2020
Dr Lino Paula and Dr Diana Salmen, European Commission, BE

10.45 Casework continued - designing pilots and field labs

13.00 Lunch

14.30 Groups report back, followed by discussion and forward planning: turning opportunity into reality

16.00 Workshop close followed by drinks

Workshop participants

1. Dr Gabrielle Anne Bertier (Centre for Genomic Regulation, ES)
2. Dr Peter Bauer (Human Genetics, University of Tübingen, DE)
3. Dr Christoph Bock (CeMM Research Center for Molecular Medicine, AT)
4. Dr Pascal Borry (Centre for Biomedical Ethics and Law, KU Leuven, BE)
5. Dr Annelien Bredenoord (Medical Ethics, Julius Centre, UMC Utrecht, NL)
6. Prof Anthony Brookes (Dept of Genetics, University of Leicester, UK)
7. Prof Xavier Estivill (Genomics and Disease, Centre for Genomic Regulation, ES)
8. Prof Raoul Hennekam (Pediatrics, Amsterdam Medical Centre, NL)
9. Dr Louise Johnston (Institute of Genetic Medicine, Newcastle University, UK)
10. Prof Helena Kääriäinen (National Institute for Health and Welfare, FI)
11. Prof Nine Knoers (Medical Genetics, UMC Utrecht, NL)
12. Dr Francesco Lescai (Department of Biomedicine, Human Genetics, Aarhus University, DK)
13. Mr Ruben Maalman (Freelance illustrator, NL)
14. Dr Fiona Nielsen (DNAdigest, UK)
15. Prof Gert Jan van Ommen (Department of Human Genetics, Leiden UMC, NL)
16. Dr Cor Oosterwijk (European Genetic Alliances Network, NL)
17. Dr Justin Paschall (Variation Team, European Bioinformatics Institute, UK)
18. Dr Lino Paula (Ethics and Gender, DG Research and Innovation, European Commission, BE)
19. Dr Barbara Prainsack (King's College London, UK)
20. Dr Maud Radstake (Patient Advisory Council, Radboud UMC, NL)
21. Janna Saarela (Insitute for Molecular Medicine Finland, FI)
22. Dr Diana Salmen (Personalised Medicine, DG Research and Innovation, European Commission, BE)
23. Dr Daan Schuurbiers (Responsible Innovation Collective, NL)
24. Dr Ralf Sudbrak (Max Planck Institute for Molecular Genetics, DE)
25. Dr Morris Swertz (Genomics Coordination Center, UMC Groningen, NL)
26. Dr Harriet Teare (HeLEX - Centre for Health, Law and Emerging Technologies, UK)
27. Dr Terry Vrijenhoek (Programme Manager, Centre for Genome Diagnostics, NL)