



House of Lords Science & Technology Committee:
Sub-Committee II

Inquiry into Genomic Medicine

Submission from Roche Applied Science
21 April 2008

EXECUTIVE SUMMARY

1. Roche Applied Science (RAS) provides cutting-edge research equipment to the life science community. We have been at the forefront of innovation in this area for many years.
2. This memorandum draws on our experience working with genetic research laboratories of all kinds and reflects directly conversations we have had recently with academics working in these institutions. Specifically, it focuses on three obstacles that are currently preventing genomic research from being translated into and beyond the development phase:
 - a) The need for further investment in bioinformatics, IT infrastructure and training
 - b) The lack of a nationwide database
 - c) The fact that funding has been focused on research at the expense of development
 - d) The lack of clarity regarding the UK regulatory landscape with respect to diagnostics

INTRODUCTION

About Roche Applied Science

3. Based in Basel, Switzerland, Roche has grown over the past 100 years from a small pharmaceutical laboratory into one of the world's leading research-focused companies in the healthcare sector. Today, Roche employs around 75,000 people and operates in over 150 countries. The business is focused on two healthcare areas: diagnostics and pharmaceuticals.
4. RAS is a global business area within the Diagnostics Division, employing approximately 1,800 worldwide. RAS's headquarters are in Penzberg, Germany; the UK operation is based in Burgess Hill, West Sussex.
5. RAS welcomes the opportunity to submit written evidence to the Sub-Committee. The Roche Group has also submitted a memorandum. Given our different, though complementary, perspectives, we have decided to submit evidence separately rather than combine the two into a single paper.



Our heritage in genomic research

6. RAS has always been at the forefront of innovation in genomic technologies. We provided the first new generation sequencing system on the market, and our products have evolved significantly since then.

7. Our focus has always been on the production of high quality data which can be easily analysed, allowing both research and clinical use alike. This is reflected in the two technologies that currently form the backbone of our product portfolio in this area:

- **the Genome Sequencer FLX (GS FLX) system:** Advances in sequencing technologies such as the GS-FLX will enable information from the Human Genome Project to be applied at an individual level to inform every aspect of patient management. Many diseases occur in genetically distinct subtypes that vary in their clinical course and prognosis. Thus, two patients who seemingly have the same disease and are treated with the same medicine may respond in radically different ways. Next generation sequencing technologies will enable common complex diseases such as cancer, heart disease and diabetes to be tackled. In addition, they have a public health impact by revolutionising pathogen surveillance – e.g. for HIV and for common hospital infections such as MRSA and C. difficile.

Gene sequencing is at the heart of research in this area and RAS has been at the forefront of technological innovation. Our next generation sequencer, the GS FLX, was launched in the first half of 2007. Since then, research using Roche's Genome Sequencer has already been documented in approximately 160 peer-reviewed publications, covering a wide range of research fields from whole genome sequencing to HIV mutation detection.

- **NimbleGen technology:** NimbleGen genomic capture technology, which has only been on the market since April 2008, allows researchers to isolate the genetic material for sequencing much more quickly and accurately than has been possible previously. No comparable technology is currently in existence anywhere in the world.

Our customers

8. RAS's customers include research institutes within both universities and the NHS. One example is Newcastle University's Centre for Life. The Centre is headed by Professor John Burn, its Medical Director, and RAS has worked in partnership with Professor Burn to help the Centre develop software for its gene sequencing research. RAS has also provided some financial support to help it develop upstream capacity and produce some pilot data.

9. The concerns identified below reflect the experience we have gained through dealing with the University of Newcastle as well as other customers. Specifically, we have consulted with the following people as a means of informing our memorandum:

- Chris Mattocks, Wessex National Genetics Reference Laboratory
- Ian Frayling, All Wales Medical Genetics Service Consultant, University Hospital Wales' Institute of Medical Genetics
- Jenny Taylor, Oxford Biomedical Research Centre

The points outlined below are endorsed by them all.

BARRIERS TO THE TRANSLATION OF GENOMIC RESEARCH

10. In his review of health research funding in the UK, Sir David Cooksey identified an insufficient focus on the translation of research as a key weakness¹. There has been much progress in this area since Sir David published his report – the establishment of the Office for Strategic Coordination of Health Research, and its focus on translating medical research, is particularly welcome, as is the emphasis on translation in the MRC's Delivery Plan to 2010/11².

11. Effective translation requires every link in the chain to be working efficiently and for 'gaps' to be plugged. Our dealings with geneticists have exposed three such obstacles that, we believe, are preventing the successful translation of genetic data into the development of new products and approaches for treating disease:

a) The need for further investment in IT infrastructure and training

12. The complete human genome cannot be sequenced in a single run using existing sequencing technology, but next generation sequencing equipment is likely to make this possible within the next five years or so. We are getting closer to this point all the time – for example, over the summer, RAS will launch new 'consumables' which will allow five times as much DNA to be sequenced by the GS-FLX system in a comparable timeframe, at about a fifth of the cost.

13. These technological developments represent a major opportunity and could significantly accelerate the progress of genomic medicine. However, they also present challenges. Specifically, the amount of raw data that is produced by genetic laboratories will continue to grow and grow at a rapid rate, but it will need to be stored and analysed if it is to be used effectively. This has raised two key concerns within the research community:

- that investment in IT infrastructure has not kept pace with the development of technology, and, in many research laboratories, the existing infrastructure that is in place will not be capable of coping with the volume of data which will be generated; this appears to be a particular problem in clinical genetics facilities in the NHS.
- that there are no bioinformaticians within the NHS to analyse the data and the environment and a lack of IT infrastructure will hinder attempts to recruit such specialists.
- that there are not enough trained individuals capable of running the IT equipment.

14. Unless both of these issues are addressed, these weaknesses will become more and more exposed. In turn, researchers will be unable to benefit from innovation in sequencing technology that is continuing apace.

¹ http://www.hm-treasury.gov.uk/media/4/A/pbr06_cooksey_final_report_636.pdf

² <http://www.mrc.ac.uk/Utilities/Documentrecord/index.htm?d=MRC004269>

b) The need for a nationwide database

15. A second major barrier to the translation of genomic research is the lack of a UK-wide database for storing sequencing information. Such a database would handle data from wherever it was produced and be accessible to researchers up and down the country. In the absence of this kind of resource, some laboratories have forged links independently and are sharing data, but there has been no consistency of approach.

16. A nationwide database would require significant investment to establish and a permanent funding stream to maintain, but the idea enjoys widespread support from across the research community.

c) Research and development funding has been focused on the 'R' at the expense of the 'D'

17. Over the last few years, funding for medical research has risen steadily, and will increase by a further £1.7 billion by 2010/11 under the terms of the Comprehensive Spending Review. This is welcome, but spending on development has not kept pace with spending on research – it is generally acknowledged that funding has been focused on the 'R' at the expense of the 'D'.

18. Again, this has hampered efforts to ensure the translation of genomic research into routine diagnostic tests, and is a crucial weakness that has yet to be addressed. The increased funding for translational research and the Biomedical Research Centres with an interest in genomics research will help to address this.

d) The lack of a clear regulatory role

The pathway for approval of new drugs in the UK is well established with the National Institute for Health and Clinical Excellence (NICE), but there is no NICE equivalent for diagnostics. The lack of clarity regarding both the regulatory and commissioning pathways presents a serious barrier to making novel molecular diagnostics available for clinical evaluation and use.

CONCLUSION

19. Our key recommendations are that investment in NHS bioinformatics and IT infrastructure is made to allow the NHS to reap the clinical benefits which novel genomics technologies could provide. This is necessary if the UK is to capitalise on the translational funding which has been committed following the Cooksey Report. In addition and related to this, the regulatory and approval processes for diagnostics need to be clarified and streamlined so that the process has the same clarity as that which exists through NICE for pharmaceuticals.

20. We would be very pleased to clarify any of the points made or to provide further information on request.