

## ***Policy Framework***

- *Who is in charge of setting and reviewing policy in this area?*

Medicines and Healthcare products Regulatory Agency (MHRA), Department of Health, Human Genetics Commission, and parliamentarians (such as members of Health or Science and Technology Select Committees). Also international policymakers (such as the EMEA, Organisation for Economic Cooperation and Development, and the European Commission)

- *Who provides scientific advice on policy development? Who monitors and anticipates potential scientific developments and their relevance to future policy? How effective are these mechanisms?*

Bodies such as the Royal Society and the Nuffield Council On Bioethics.

- *Does the existing regulatory and advisory framework provide for optimal development and translation of new technologies? Are there any regulatory gaps?*

In the EU the European Medicines Agency (EMA) has clarified pharmacogenomic terminology and has introduced a system of Briefing Meetings that allow sponsors and regulators to discuss pharmacogenomic data at an early stage through the Pharmacogenetics working Group. A guidance document explaining how the U.S. FDA and the EMA will process requests for Joint FDA/EMA voluntary genomic data submission (VGDS) briefing meetings has been published (May 19, 2006) ([http://www.ema.europa.eu/pdfs/human/pharmacogenetics/Guideline\\_on\\_Joint\\_VGDS\\_briefingmeetings.pdf](http://www.ema.europa.eu/pdfs/human/pharmacogenetics/Guideline_on_Joint_VGDS_briefingmeetings.pdf))

- *In what way is science and clinical policy decision-making informed by social, ethical and legal considerations?*
- *How does the framework compare internationally?*

## ***Research and Scientific Development***

- *What is the state of the science? What new developments are there? What is the rate of change?*

Genomic-based technologies capable of analysing multiple genetic changes have been validated in a research setting and in some cases in a clinical setting. However progress is extremely rapid in this field and we are likely to see numerous new tests being launched in both an unregulated and regulated fashion within the next three years.

- *Who is taking the lead in the consideration and co-ordination of research and the development of new technologies?*

A coordinated framework does not exist. However individual organizations such as Cancer Research UK are currently developing a set of recommendations in this area. It is clear however that a coordinated approach between government, the pharmaceutical industry, the diagnostic industry and academia is required.

- *How effective is the policy and investment framework in supporting research in this area?*

Not at all effective – There is a lack of understanding across the industry. Meaningful research, capable of impacting on clinical practice is lacking in this field. The results is poorly designed individual studies being funded that lack statistical power and as a consequence any hope of improving patient management. A number of essential criteria should be considered in order to advance meaningful research in this field.

- Identify key areas in which genomic-based diagnostics can impact on the clinical decision making process.
  - Make sure that sufficient biospecimens exist for both the discovery and validation phase of diagnostic test development (in some cases may require appropriate prospective clinical trials).
  - Ensure that the genomic technologies in question (of which there are numerous) are suitably validated (of which there are few) for clinical application.
  - Make sure that the healthcare system is informed and willing to pay for tests based on genomic-based technologies. These tests are likely to be lower volume high costs tests compared to the traditional model of high volume low cost tests.
- *How does research in the UK compare internationally? How much collaboration is there?*
  - *What are the current research priorities?*
  - *What is the role of industry? How much cross-sector collaboration takes place?*

Industry is fundamentally important in translating the new genomic technologies to enhance patient management.

### ***Translation***

- *What opportunities are there for diagnostics, therapeutics and prognostics - now and in the future?*

There are a wealth of opportunities for diagnostics, therapeutics and prognostics given the advances in the elucidation of the human genome. Technologies such as PCR and microarrays are now making their way into clinical practice within the diagnostics industry. The speed of progress has now outpaced corresponding developments in terms of industry regulation and healthcare provision.

- *Who is responsible for translation to clinical practice?*

MHRA currently regulates medicines, devices, and advanced therapy medicinal products/tissue engineering, which may arise from advances in molecular biology, genomics, gene therapy, and cell therapy etc.

Genomic research will lead to a new generation of medicines and an increasing number of specialised and “personalised/ patient stratified” pharmaceuticals and/or (in vitro diagnostic) devices that will require regulation. There are inconsistencies within the European regulations with responsibility for drug regulation being located at the EU 'level' while responsibility for medical and in vitro diagnostic pharmacogenetic/genomic tests residing with the member states competent authority. There is therefore the potential for delay of regulatory approvals of drug and devices which are co-developed or which impact on one another in the marketplace.

- *Given the pace of technological advance, how 'future-proof' is healthcare investment in this area?*

- *How does the UK compare to other countries and what lessons can be learnt?*

There is no specific centralised UK government initiative aimed at genomic research and development per se within the UK. Rather areas or issues 'accessory to/ utilised in' genomic R&D such as tissue sourcing, usage and storage (Human Tissue Act) and Genetically Modified Organisms have been legislated for separately.

In comparison in the United States the FDA have recognised the potential of genomic technology with respect to new therapeutics, diagnostics, prognostics, etc and the potential impact on drug discovery through their Critical Path Initiative (<http://www.fda.gov/oc/initiatives/criticalpath/commentary.html>).

FDA have also released several draft guidance and concept documents around genomic tests (<http://www.fda.gov/cder/genomics/regulatory.htm>) detailing how to make voluntary genomic data submissions with the intention of "... facilitating progress in the field of pharmacogenomics and genetics by helping to shorten development and review timelines, facilitate rapid transfer of new technology from the research bench to the clinical diagnostic laboratory, and encourage informed use of pharmacogenomic and genetic diagnostic devices."

- *How meaningful are genetic tests which use genome variation data? What progress has been made in the regulation of such tests?*

Genetic tests based on genomic technologies have the potential to outperform conventional diagnostic approaches. The ability to look at multiple genetic markers using microarray (DNA Chip) technology will transform the diagnostics industry. At present these types of approaches are covered under the FDA's guidance document for in vitro diagnostic multivariate index assays draft guidance. No similar guidance exists for the UK.

### ***Biomarkers and Epidemiology***

- *In what way do genome-wide association studies contribute to the identification of biomarkers? How is the study of genetic factors and biomarkers integrated for translational purposes?*
- *What impact will genomic data have on data emerging from projects such as UK Biobank, Generation Scotland and other biobanks?*

The provision of genomic data on samples contained within these biobanks will allow the cross referencing of Clinical Data with the genomic data. Linking of genomic and clinical data will provide a data source that will allow the clinical features of the samples and the genomic data to be mined together. This data mining and bioinformatic analysis may lead to the development of new biomarkers and gene signatures for delineating risk of disease, recurrence of disease or even stratifying which patients will respond to existing therapies.