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SACGHS Public Consultation Draft Report: Response from the PHG Foundation

Introduction

The UK Foundation for Genomics and Population Health is the successor body to the Public Health Genetics Unit. Its overarching purpose is to foster and enable the application of biomedical science, particularly genome-based technologies for the benefit of human health. Among its specific objectives is the promotion of a social and regulatory environment that is receptive to innovation, without imposing an undue or inequitable public burden. The Foundation has a particular interest in the way that new technologies are translated within health services, in genetic research and its impact upon clinical and public health services. For example, we have recently completed a project on cell-free fetal nucleic acids for non-invasive prenatal diagnosis which was led by a UK expert working group[1] in which we considered the likely effect of patents on the uptake and implementation of this new technology within the NHS. We have also published reports on the regulation and evaluation of diagnostic genetic tests.

Comments

1. We are generally supportive of advances in biomedical sciences that might promote better health and medical care. However, we are particularly concerned about the effect of gene patenting on the future development of multiplex tests based on many genes and whole genome sequencing (lines 2794-2855).
2. The recent plethora of genome-wide association studies has revealed numerous common variants associated with disease. Whilst the risk associated with each of these individual variants is generally very small, their cumulative effects may be significant in the determination of susceptibility to many common complex disorders, including diabetes and coronary heart disease. For example, it has been suggested that genetic risk profiling could be used to effectively target screening programmes towards those at highest risk of disease. Whilst is it not yet clear whether gene-disease associations are themselves patentable, we are concerned that such disease risk profiling techniques might have limited utility for improving health care if they are hampered by gene patent thickets.
3. We are generally supportive of measures to increase transparency and proposals for manufacturers and distributors of tests to be more accountable. Indeed we have pressed for the establishment of a publically accessible database of new and existing laboratory tests which contains evidence of clinical performance of diagnostic tests (including validity of the gene-disease association, and evidence of clinical validity and clinical utility).[2] We see no reason why such a database should not include details of valid and applicable patents, and where appropriate, electronic links to patent documentation. However the process of populating such a database and keeping it up-to-date might present a significant challenge, particularly if patent holders, users or statutory authorities were to seek to rely upon it for enforcement. Moreover, public databases (such as esp@cenet) already exist to allow companies to search for relevant patents.

[1] PHG Foundation (2009) Cell-free fetal nucleic acids for non-invasive prenatal diagnosis at http://www.phgfoundation.org/pages/work2.htm#ffdna

[2] PHG Foundation and Royal College of Pathologists (2008) The evaluation of diagnostic laboratory tests and complex biomarkers. Summary of a Diagnostic Summit 14-15 January 2008.

4. In our view, the suggestion that such a database should include details of licensing agreements is unrealistic, potentially detrimental to commercial partnering, and not consistent with requirements for other (non-genetic) medical tests.
5. Next generation genome sequencing technology is developing very rapidly. Exomic sequencing of the coding regions of the genome (around 1%) has already been used in clinical research and whole genome sequencing is likely to become affordable within the next 5 years. Even if the measures for increasing transparency outlined in the report were to be fully adopted, we believe that it would be unfeasible for developers or providers of such services to be expected to *"obtain licenses for all unexpired patents that claim a nucleic acid molecule… or… a diagnostic process derived from the human genome".* Such a requirement would be both burdensome and impractical, and would only serve to stifle potential research and clinical applications of whole genome sequencing.
6. Regarding the options for statutory change outlined in the final section of the report at Chapter V, Section 8 of the report (lines 3272-3277) our preference is for option E. The UK has an explicit exemption for research, and although in practice there can be difficulties establishing clear boundaries between research and clinical care, on balance, the presence of this exemption helps to encourage innovation.
7. More generally, we favour many of the recommendations arising from the Nuffield Council on Bioethics, outlined in its 2002 report, *The ethics of DNA Patenting* (lines 1550-1555), especially those that advocate raising the bar for obviousness and utility when granting DNA patents, and narrowing the definitions of uses covered by patent claims.

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