

Response to the Department of Health consultation on the framework for the registration of health and adult social care providers and consultation on the draft Regulations.

Response from the PHG Foundation

1. Introduction

- 1.1. 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.
- 1.2 The PHG Foundation has had a long standing interest in the prediction and prevention of future ill health. We have expertise in the evaluation and regulation of diagnostic and biomarker (particularly genetic) tests and advocate for the establishment of scientific and clinical thresholds so that tests are effective and safe. More generally, we have also considered the governance that should apply to laboratories providing such tests (including interpretation of the test result, quality assurance of laboratories providing testing, health and safety, as well as consideration of relevant ethical legal and social issues which apply such as the requirements for obtaining adequately informed consent and policies on privacy and confidentiality).
- 1.3 These issues are becoming increasingly pressing as the potential for direct marketing of genetic tests and diagnostic tests to the public increases. Our response will therefore focus on the provision of diagnostic and testing services, and the drafting of Regulation 11.

2. General comments

- 2.1 We welcome the approach to the regulation of diagnostic procedures set out in the consultation paper. We are concerned that the regulations as drafted fail to cover explicitly some important aspects of laboratory services, such as the use of genetic tests and other predictive tests for the purpose of disease prevention. In particular the draft regulations are unclear as to whether tests based on the analysis of DNA and other nucleic acids, or tests which are intended to predict the risk of future disease and physical or mental disorders, are included within the term 'diagnostic procedures' and thus fall within the remit of the CQC.
- 2.2 For some time we have argued that the existing governance of biomedical and diagnostic tests in the private sector is unsatisfactory, whether offered direct to the consumer or providing a service to physicians and other health care professionals, and in particular that the regulation of companies purporting to provide genetic tests that predict the risk of future disease is inadequate. It seems to us the wording of section 8 could exclude such services if they are managed by a laboratory scientist¹, or by any lay person not falling within the definition of a health care professional. An indirect consequence of these regulations could be that prospective providers of laboratory testing services outside the NHS might expressly reject supervision by a health professional (as defined by regulation 8(11)(c)) so to their

¹ This seems to fall outside the definition of health care professional as defined in the regulations at section 2.

services fall outside the scope of these regulations. This would leave private laboratories offering predictive testing services to the public unregulated.

3. Do the draft Regulations set out at Annex B accurately reflect the policy set out in Chapter 3 and Annex A? (Q.3.1)

A. General Points

3.1 Our first general point concerns the set of activities that will be regulated under the Health and Social Care Act (2008). Regulation 8 (with the exemptions set out in 8(2) to 8(10)) provides only:

(1) for the provision of *treatment* for disease, disorder or injury

and only if it is

(2) by or under the supervision of a health care professional

with the word *treatment* being defined in Regulation 2 as including (a) diagnostic or other investigative procedures (b) nursing, personal or palliative care and (c) the giving of vaccinations and immunisations.

3.2 First, we are concerned that the definition of *treatment* in the regulations is applied too narrowly and fails to reflect the transition in health services from that of 'treating disease' to 'preventing disease' by the giving of advice about the risk of future disease': namely a paradigm of prediction and prevention. We would therefore prefer a more inclusive definition such as 'the provision of advice and management of persons in relation to disease, disorder or injury', to accommodate a wider conception of health care.

3.3 Second, we are concerned that the activity will only come within the scope of the Regulation if it is 'by or under the supervision of a health professional'. (We note that the effect of section 8(12) is to exempt those activities carried out by named professional groups, presumably to avoid duplication). Our concern is that where activities are carried out by neither a health care professional, nor a member of any of the professional groups named in section 8(12), that the current draft allows such activities to continue without any regulation at all, such as where a laboratory scientist or a person without relevant qualification seeks to give advice in relation to the management of disease or the future risk of disease.

3.4 Third, we are concerned that there is insufficient clarity about the scope of Regulations 8 and 11 where both apply. We assume that Regulation 8 is intended to apply where treatment is provided by or under the supervision of a health professional (such as a colonoscopy carried out by a health professional, or an x-ray carried out on the premises of a specialist); and that Regulation 11 applies to pathology specimens that are taken and analysed in a laboratory setting. However, it is not clear which regulation applies where, for example, blood specimens are taken by a health professional and then sent to a third party laboratory. This question may be further complicated by the involvement of overseas laboratory service providers.

B. Regulation 11

Diagnostic Procedure

- 3.5 The term *diagnostic procedure* as used in Regulation 11(a) to (c) of the draft regulations is undefined. A formal definition needs to be considered. On the surface the term may appear self explanatory, namely that the purpose of the procedure is 'to make a diagnosis'. Our view is that this implicit assumption is misleading. The draft needs to make clear by explicit definition that such procedures, often labelled 'diagnostic' as a shorthand, are often carried out with other purposes in mind, such as predicting the risk of future disease or disorder and determining prognosis. For example, imaging and biopsy may be used to assess the spread of a cancer and give a likely prognosis, even though the diagnosis may already be known. Likewise blood tests such as cholesterol are taken to assess the risk of future heart disease.
- 3.6 The assessment and communication of risk is key to the provision of preventive medicine and predictive testing². Biochemical tests results are already used routinely in risk prediction algorithms to inform management and predict future susceptibility to disease, and screening services are already available both privately and publically to assess an individual's risk of developing a particular disease in the future. Our view is that the communication of predictive risk information (including predictive genetic test information) is likely to become a central public health activity within the next decade.
- 3.7 The procedures in Regulation 11(a) and (b) and in the first arm of (c) can be interpreted as having a single step; whereas the procedure in the second arm of (c) involves two steps, the taking of the sample and its subsequent analysis. The drafting should clarify that both these activities should be included within the regulations, and also that samples might include not only blood, but saliva, urine, semen, serum, plasma, tissue and organs.

Disorder and Disease

- 3.8 Regulations 8 and 10 refer to the treatment of *disease*, *injuries* and *disorders* but the definition (as applied to the examination of tissues, fluids and cells) in Regulation 11 seems to exclude obtaining information on *disorder* and *injury*. This is inconsistent. Any attempt to try to distinguish between *diseases* and *disorders* for these purposes seems futile and arbitrary. We would like to see the definitions in section 11 widened to include injury and disorder, or clarified such that, for the purposes of the regulations, *diseases* and *disorders* are treated in the same way.

Purpose of the Procedure

- 3.9 The purpose of a procedure set out in Regulation 11(c) is in our view too narrow. We would include (for the reasons first alluded to in our Para 3.2 above) in the list of purposes not just 'obtaining information on the *causes and extent of disease* and the *response to a therapeutic intervention*', but also 'obtaining information about *the risk or probability of future disease*'. The practice of preventive medicine will increase over the coming decades and much medical practice will be aimed at preventing disease before its development. Moreover, much of the concern about direct to consumer testing has been to do with private screening providers and predictive genetic tests.

Exemption for Low Risk Tests

² For example NICE has recently recommended that the cascade genetic testing of first, second and third degree relatives of every individual diagnosed with familial hypercholesterolaemia is standard practice.

- 3.10 The consultation paper states that whilst certain diagnostic procedures, such as pathology and cytology tests, are within the remit of the CQC, that 'low-risk or non-invasive diagnostic procedures should not be within the scope of the registration'³. We agree with that sentiment. However, our concern is that some types of predictive test, which may currently be excluded by the Regulations, are 'high risk' in that they convey information about serious health problems which will inevitably occur in the future. The archetypal case is the test for Huntingdon's disease. We therefore wish the regulation to give greater emphasis on establishing criteria that serve to distinguish between those tests which will be regulated and those which will fall outside the scope of regulation
- 3.11 Regulation 11(3) was drafted with the intention of excluding such low risk tests. Blood tests which are EITHER taken with a pin prick OR however taken, but which do not need to be sent to a specialist facility, are exempted. Although, at present, this may adequately serve the purpose of the regulations, we suggest that in the not too distant future, desktop analytical tools may be available for detecting genetic variants and for testing in other high risk situations. The criterion of having or not having 'to send to a specialist facility' does not therefore reliably serve to distinguish high from low risk tests. Moreover, the method of securing the specimen, whether by pin prick or otherwise, is an inadequate guide to the risk of the test. For example, all genetic tests can be performed without any direct risk associated with obtaining the sample itself, such as by using saliva or buccal swabs, but may nonetheless produce a highly predictive result that has profound implications for the future health of the individual. Such tests therefore clearly represent a 'high risk', and activities which depend on the use of such technology need to come within the scope of regulation if they are used for highly predictive tests for serious disorders.
- 3.12 We have one other comment with regard to Regulation 11(3), concerning the use of the word *procedure*. In 3.7 above we noted, in the context of pathology tests, that the *procedure* would involve both the taking of the specimen and its analysis. It might therefore be more correct to substitute 11(3)(a) the word *procedure* with the phrase *the taking of the sample*.

C. Additional clarification

- 3.13 We would wish to see additional clarification as to:
- the extent to which genetic (and other forms of predictive) tests are to be included within the CQC's remit and how, if at all, this varies with the predictive ability of the marker in question;
 - whether laboratories that process specimens (i.e. receive and analyse specimens but have no direct contact with patients) will be regulated, and in particular to consider the situation that such laboratories are run by individuals who are neither medical practitioners nor a health care professional as defined in Section 60(2) Health Act 1999 – for example, laboratory scientists ;
 - the need to discriminate between those services that are provided direct to consumer from those offered via a registered physician or other appropriately qualified and professionally regulated health care professional.
4. If not, what changes are needed to the draft Regulations to ensure they reflect the policy set out in Chapter 3 and Annex A? (Q3.2)

³ At page 105.

- 4.1 We have alluded to some of the changes in Para 3 above but in particular we wish to emphasise the following points:
- (a) the term 'diagnostic procedure' should be extended to include disorder or injury;
 - (b) it should explicitly include the prediction and risk of future disease, disorder or injury as well as an assessment of prognosis;
 - (c) since it is common for DNA to be extracted from saliva samples, these should be mentioned specifically in section 11(c).
5. **Do the draft Regulations set out at Annex D accurately reflect the policy set out in Chapter 5 and Annex C? (Q5.1)**
- 5.1 As argued above, our view is that laboratories offering a medical or health-related testing service to the public comprising of analysis and/or interpretation should be regulated by the CQC. At a minimum, the criteria for successful registration should include evidence of the following:
- participation in an agreed quality assurance scheme;
 - health and safety;
 - that those providing samples have given an informed consent as to their use;
 - that adequate privacy and data security policies are in place;
 - that there is robust scientific evidence supporting a link between the test and the disease;
 - that if an interpretation service is provided, those providing either technical or clinical interpretation, including the assessment of disease risk, have the necessary qualifications and competencies to provide such a service.
6. **If not, what changes are needed to the draft Regulations to ensure they reflect the policy set out in Chapter 5 and Annex C? (Q5.2)**
- 6.1 Our comments are already detailed above.

PHG Foundation
23 May 2009

Contact details: alison.hall@phgfoundation.org