



Consultation Response

Revision of the Directive 98/79/EC on *In Vitro* Diagnostic Medical Devices

Response by Genetic Alliance UK

1. Genetic Alliance UK (formerly Genetic Interest Group) is the national charity supporting all those affected by genetic conditions. Genetic Alliance UK aims to improve the lives of people affected by genetic conditions by ensuring that high quality services and information are available to all who need them. Our membership represents 138 voluntary organisations working for a wide range of conditions, many of which have no cure or treatment.
2. In 2008, Genetic Alliance UK founded Rare Disease UK to stress the importance of strengthening research programmes into rare disease, encourage the development of national rare disease policies, and develop and share common policy guidelines. Rare Disease UK is an alliance between over 125 patient organisations, clinicians, academics, industry and interested individuals brought together in response to the unmet care needs of the estimated 3.5 million people affected by rare disease in the UK.
3. Genetic Alliance UK welcomes this review and the opportunity to respond.

Risk-based classification for *in vitro* diagnostic medical devices

4. Genetic Alliance UK considers the proposal to develop a risk-based classification for *in vitro* diagnostic medical devices to be an appropriate method to ensure the regulation is able to cover unforeseen future developments.
5. We are concerned that classification of risk levels for genetic tests should be based not only on the degree of invasiveness of the test and risks associated with this, but also reflect the impact the potential results of the tests could have upon the patient and their family.
6. A positive test for Huntington's disease for example, gives its recipient a certainty that he or she will suffer from the lethal degenerative brain condition. This information has been shown to have a devastating effect on families when delivered without professional support. The impact of this genetic test upon the test subject is potentially much more severe than a test for Familial Hypercholesterolaemia, a metabolic condition which can be managed with medicine and diet control; thereby enabling the affected individual to avoid the onset of severe disability and early death.

Exemption on "in-house" tests provided for by article 1(5) of Directive 98/79/EC

7. Genetic testing is one of very few services that public sector health services can offer those affected by genetic disease today. It provides invaluable information to health practitioners and patients; allows the planning of the management of the condition; enables valuable advice regarding family planning and reproductive choice; delivers the relief to the patient that comes with a diagnosis;

makes possible the timely provisions of therapies where they exist; and furthers our knowledge regarding the incidence and natural history of the disease in question.

8. In the UK there is a total of 498 genes for which tests have been authorised by the UK Genetic Testing Network (UKGTN www.ukgtn.nhs.uk). There are a further 113 genes for which tests are undergoing assessment for authorisation, a number which reflects the likely growth of IVDs in the genetic testing field.
9. The vast majority of these tests are for rare diseases, and are developed in NHS genetic laboratories for use by the NHS, fitting the definition of an “in house” test. The likely usage rate of the majority of these tests is extremely small.
10. It is vital that these tests continue to be available for use by health services in the UK. The necessary costs, in both time and monetary terms, required for these tests to obtain a CE marking are prohibitive. The benefits in terms of patient safety are negligible given that all UKGTN member laboratories are required to participate in appropriate efficacy and quality assurance schemes. The result of repealing the exemption for “in house” tests would be a severe drop in the availability of genetic tests for rare diseases. A continuation of the exemption in some form or another is therefore essential.
11. In the UK, all genetic tests for medical purposes are evaluated by UKGTN. This organisation ensures the provision of high quality equitable genetic testing services within the National Health Service. The network is a collaborative group of all stakeholders with an interest in the regulation of genetic testing, including laboratories, clinicians, commissioners of genetic services and patient support groups.
12. The UKGTN provides regulation of genetic testing in the UK in a proportionate cost-effective manner. Any measures to oversee quality, efficacy and safety of IVDs exempt from this directive should follow a similar pattern to the UKGTN.
13. In November 2008 the European Commission published its Communication on Rare Diseases: Europe’s Challenges, and in June 2009 the Council of the European Union issued a recommendation on an action in the field of rare diseases. Any over burdensome regulation of “in house” *in vitro* diagnostics would directly contradict the aims of these initiatives by making it less likely that patients with rare conditions would be able to receive an accurate diagnosis and so benefit from effective medical care.

Conclusion

14. We would resist any request to introduce over burdensome regulation such as CE marking for “in house” genetic tests as this will not contribute significantly to patient safety where other measures are in place, it will push up costs and reduce the likelihood of patients receiving a timely diagnosis and therefore accessing subsequent appropriate care and support.



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